Understanding of genetic test results for osteogenesis imperfecta by attorneys and child welfare workers in non-accidental injury cases

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Abstract

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This dissertation research explored the ways in which child welfare workers and attorneys involved in non-accidental injury (NAI) cases understand and use genetic test results for osteogenesis imperfecta (OI). 14 semi-structured expert interviews with 14 professionals known to be involved in OI vs. NAI cases were recorded, transcribed, coded, and analyzed, in order to help inform a larger, more wide-scale quantitative survey. The on-line survey included 46 questions, and participants were drawn from a number of sources. A total of 192 participants (both attorneys and child welfare workers) took the survey, 102 of which completed every question, 185 of whom completed more than 70% of the survey. Results suggested that some professionals involved in OI vs. NAI cases may not be equipped to understand genetic test results that they are returned, and they may not be able to get help from an appropriately trained scientific expert to aid them in understanding the result. The majority of participants agreed that educational or policy/practice recommendations could be useful in OI vs. NAI cases, and that most professionals in their field would be interested in learning more about OI and its relationship to NAI. Finally, a set of suggestions are proposed, outlining different recommendations for education, policy, and practice. Many limitations in this study were identified that limit the ability to generalize the findings beyond the study population.

Dedication

I would like to thank my friends and family who have supported me along the way, many of them from quite a distance. Their unflagging encouragement and belief in me and my endeavors is priceless and so appreciated. My department peers have provided an unremitting source of academic dialogue, friendship, commiseration, and laughs, and for that, I am forever grateful. A special thanks to Ben Wiggins, a great source of inspiration and mentorship. He always pushed me to be a better teacher, and inspired in me a passion for science education that infuses exploration, collaboration, and joy in the classroom. Lastly, thanks to the swing community, dancers and musicians alike, for keeping me balanced in so many ways.

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Chapter 1: DISSERTATION AIMS

Children with injuries consistent with abuse represent an urgent problem for health care providers and others involved in protecting children. In 2015, the Department of Health and Human Services recorded that in the U.S. there were over 119,000 confirmed cases of child physical abuse[1] and it is suspected that many other cases went unreported and undocumented. In the U.S., when there is substantial suspicion by a physician or others that a child is being abused, the child is removed from the home and placed into foster care by law enforcement and/or child welfare workers, to avoid any possibility of further injury while the investigation is underway. The amount of evidence required for the removal of a child from the home is not standardized.

Although child physical abuse, or non-accidental injury (NAI), is common and many injuries result from NAI, it is not the only cause of childhood injuries. Certain genetic conditions can lead to a predisposition to bone fragility and skeletal fractures, which could be difficult to distinguish from injuries due to NAI. The inherited condition osteogenesis imperfecta (OI) is characterized by fragile bones that break easily[2] and it is the most common genetic condition that leads to predisposition to skeletal fracture.[3] Previous research has indicated that the incidence of OI among children 0-3 years old who have been hospitalized with skeletal fractures, do not have head trauma, and have been suspected of NAI, is about 0.85%.[4]

Other conditions, such as genetic forms of rickets, disorders of vitamin D metabolism,[5] disorders of copper metabolism,[6] and hypophosphatasia,[7] can also result in signs that can be confused for NAI.[8] Since OI is the most common inherited syndrome that predisposes children to skeletal fractures, this dissertation focuses on OI as a test example, although other conditions should not be ignored. NAI cases are often emotionally charged, and a mishandling of a case could lead to emotional trauma within the family. Therefore, OI and other medical conditions

that could be mistaken for NAI should be ruled out in a time-efficient manner as part of an evaluation of suspected child abuse.

Clinical evaluation alone is not always sufficient to diagnose or to rule out OI.[9] A genetic test for OI can be done, although the genetic test results for OI are not always easy to understand. 90% of all OI cases are due to mutations in the genes *COL1A1* or *COL1A2*. DNA-based testing for variants in these genes can provide unequivocal diagnosis of OI (benign or pathogenic), but can also identify variants of uncertain significance (VUS). In other words, a DNA variant could be found in a collagen gene, yet it is unclear whether the variant is causative of OI. A VUS test result should not be considered as the equivalent to either a positive or negative test result, but for an individual without formal training in genetics, the distinction may not be obvious.[10]

Although previous research has pointed to the fact that more professions are now using genetic information, it has not translated into providing those working professionals with a parallel increase in genetics education.[11,12,13,14,15] In the legal system, genetic test results are being used more commonly as evidence. Thus far, no research has been done on how attorneys and child welfare workers understand and use genetic test results in the context of NAI cases. A misappraisal or misrepresentation of the genetic evidence could have significant consequences on the family under investigation.

In 46 states in the U.S., children suspected to be victims of child physical abuse can be removed from their home without a court order by law enforcement if a clear and present danger to a child's health, safety, or welfare if suspected.[16] About 20 U.S. states give this same authority to Child Protective Services.[17] If an allegation of child abuse is determined to be founded, a child may be placed into state custody or foster care for protection. Alternatively, if there is a plausible medical explanation for the child's fractures, the child may be safely kept within the family and

provided with appropriate medical care. The separation of a child and parents can cause a total disruption of the family, including a loss of familiar people and surroundings for the child, which may be traumatic.[18] While the determination of whether a child has suffered physical abuse takes into account a variety of evidence, genetic test results can play a pivotal role.9 For this reason, it is critical that information obtained through genetic testing for OI is properly understood and represented.

Learning more about how much awareness attorneys and child welfare workers have of OI, and their understanding and use of the genetic test results will help indicate whether there are any potential problems in this system that require addressing. If there are, this information could help inform potential educational and policy suggestions regarding genetic testing for OI in the context of NAI cases.

Three aims were developed to help aid the investigation of this research topic. This study was designed as an exploratory study to begin to investigate a variety of potential issues that might be pertinent to stakeholders involved in OI vs. NAI cases. The results of this dissertation are not intended to be generalized beyond the study population.

Aim 1: To interview attorneys and child welfare workers who had experience with child physical abuse cases that involved genetic testing to learn about their experiences and to get advice regarding what questions should be included in a broader survey on the same topic. The aim of the study was accomplished via expert interviews with attorneys who represent parents in child abuse cases and with child welfare workers. Conversations explored the participant's experiences on OI vs. NAI cases, their thoughts regarding phrasing for the survey, and question topics that

should be included in the survey. The interviews were audio recorded, transcribed, and coded thematically.

Aim 2: To understand the ways in which genetic test results for OI are understood and represented by child welfare workers and attorneys in OI vs. NAI cases. This aim was accomplished by using a mixed methods internet survey. This group that was invited to participate in the survey was identified via state offices of public defense and through the National Association of Social Workers (NASW). Questions on the survey represented themes that emerged from the interviews in Aim 1. The hyperlink to the survey was distributed via email, and all responses were anonymous.

Aim 3: To create a list of suggestions for education and support of working professionals in child welfare or child/parent advocacy on OI, in regards to any potential issues that may be identified during Aim 2. Findings that emerged from the survey were used to draw up a list of suggestions that could be helpful for professionals involved in OI vs. NAI cases.

Chapter 2: BACKGROUND

Non-accidental injury statistics

Non-accidental injury (NAI), or physical abuse, is a major public health problem. There are over 119,000 cases of confirmed child physical abuse every year in the U.S. and it is likely that many more cases go unreported.[1] Fractures are the second most common injury from NAI after bruising, and the majority of fractures occur in children less than three years old.[19] Fractures in children account for up to one-fourth of all pediatric injuries;[20] it is estimated that 18% of all children will have a fracture by age nine.[21] In children under 16 years of age, 1.3% of femur fractures were attributed to NAI, approximately 75% was attributed to falls or motor vehicle accidents, and the remaining fractures were unexplained.[22] In the U.S., the prevalence of NAI with fractures is approximately 24:10,000 children in the birth to three year range (as of 2002).[23] In 2004, in children less than three years old, 24%-40% of all fractures were suspected to be related to non-accidental injury.[24, 25, 26, 27] Among children hospitalized each year for fractures (about 150,000), the percentage of those in which NAI could be a cause for fractures (and without traumatic brain injury) is highest among children less than one year of age (20.4%).4

However, not all children who present in a clinical setting with injuries suggestive of abuse were actually abused. Due to the fact that NAI is so common, it is not difficult to see why health providers often suspect NAI when a child presents with unexplained fractures. The inherited condition osteogenesis imperfecta (OI) is characterized by fragile bones that fracture easily with little trauma. Given that the history and explanation of the fracture(s) often insufficiently explains the severity of the injury, children with OI are often suspected victims of abuse before an OI diagnosis is made.[28]

OI prevalence statistics

OI is a relatively rare condition with a prevalence of about 1:10,000,[29] and due to the range of severity of OI, only cases on the mild end of the spectrum are likely to be confused with NAI. In other words, there is a very small fraction of children that are suspected of NAI that will actually have OI. At the moment, it is not clear what the true incidence of OI is among those evaluated for NAI, as there is no registry or database for this. Leventhal et al estimated that OI accounts for about 0.85% of hospitalized children with fractures but no head trauma who are evaluated for NAI. Many patients are referred for genetic testing for OI as a means to help rule out a genetic cause of unexplained fractures,[1] but, of course, OI and NAI are not mutually exclusive, so a positive OI test result does not exclude purposeful harm as the reason for injury.

It is quite difficult to determine the number of children who are suspected of NAI and *with* OI who could be missed by diagnostic testing. The genetic test for OI and its ability to accurately detect OI in a child suspected of NAI depends on the test's sensitivity (the sensitivity of the genetic test for OI has been estimated at 95%),[2] and the a priori likelihood that a child could have a mild form of OI with no other clinical signs that would indicate the condition (2%). Therefore, an infant who is suspected of NAI and is tested for OI but has a *normal* test result has a 1/980 chance of having OI (in one of the two genes most commonly associated with OI).[30] In other words, the probability that a child who is suspected of NAI but really has OI and is diagnostically tested for OI is quite unlikely to be missed by diagnostic testing.

Professional stakeholders in OI vs. NAI cases

There are four primary groups of professional stakeholders that have significant interest and involvement in OI vs. NAI cases: child welfare workers, attorneys, physicians, and law enforcement.

Child welfare workers: this group includes child protective services (CPS) workers, hospital social workers, and social workers that work for Army, Navy, Air Force and other armed forces or personnel supported by the Federal Departments of Public Defense. These professionals receive and evaluate NAI reports to determine if a child is at risk. They also help counsel the family, support and rehabilitate the family, or help coordinate foster care or an adoptive home. While many of these professionals have a master's degree in social work, it is not a requirement to work in child protective services.

Attorneys: This group includes attorneys that work in both civil cases as well as in criminal cases. Another key legal professional is the judge, who controls how trials progress, and makes important decisions regarding how the law should be interpreted. Importantly for OI vs. NAI cases, judges must assess evidence presented to them, such as genetic test results. If scientific experts from opposite sides (defense vs. prosecuting) disagree on the meaning or significance of genetic evidence, it is often the judge who makes the final decision. All of these professionals have completed a juris doctor (JD) degree and passed a state bar examination. Judges have completed additional judicial training and have been elected or appointed as judges.

Physicians: this group includes medical geneticists as well as pediatricians and emergency room (ER) doctors. Pediatricians or ER doctors are often brought into an NAI case to do a physical examination on a child. The child may then be referred to a physician that

specializes in child abuse, or if there is concern that the child could have a medical condition causing the fractures, they could be referred to a medical geneticist or other specialist, depending on the concern. In all states, doctors are required under law to report any child that they suspect has been abused to CPS or law enforcement. If a doctor fails to make this report, he or she could face penalty. Physician's training includes completing a doctor of medicine (MD) degree and finishing a multiyear residency. Many would have also completed a fellowship. Medical geneticists have done a specialized fellowship in genetics.

Law enforcement: This group refers to anyone charged with making sure that people are following the law, including state or local police officers, or certain federal agencies like the FBI. Law enforcement often gets involved with NAI cases in order to help investigate whether the child has been harmed and who the perpetrator was, or whether the child is at risk. These professionals are usually involved after an accusation is made, before a trial begins. Their background and training involves passing the law enforcement entrance exam, and then completing academy training, which varies depending on the type of law enforcement.

Each stakeholder will get involved in an NAI case at a different time in the investigation depending on the situation. For example, if the child is brought to the emergency room for an illness, the ER doctor could find that the child has multiple healing fractures. As a mandatory reporter, the doctor could then contact CPS, who will decide whether to get law enforcement involved for an investigation. A decision will then be made as to whether the case will require legal action. If it does, attorneys will get involved at this point. There are two separate courts: one for criminal law and one for civil law. There is no rule about which order the trials must proceed in.[31] Civil cases involve dependency action to determine whether the accused parent(s) should be able to retain custody of the child. CPS workers are involved in civil cases. A criminal

case will determine whether the parent(s) should face a criminal conviction for the abuse. CPS workers are not involved in these cases. These trials may or may not involve a medical expert to help suggest any additional tests that need to be done or to help attorneys or child welfare workers understand a test result. Each trial will be adjudicated by a judge. This particular scenario is outlined pictorially in a flow chart in Figure 1. Other scenarios may also arise.

Current state of awareness of OI among professionals

The use of genetic tests in everyday life is increasing at an exponential rate, but the rate of genetics education among professionals that use genetic test results is not keeping pace. Sometimes people with no scientific training are asked to understand and use genetic results to make important decisions,[32] such as when genetic technology is used in legal cases to determine the presence or absence of OI.[10] Research indicates that non-scientists need help using genetic data correctly,[33] and understanding how they use genetic data is crucial. Child welfare workers and attorneys in OI vs. NAI cases play a significant role in child abuse cases, yet their knowledge of genetics is limited and their awareness of OI is currently unknown.

Preliminary data from my master's thesis[34] indicated that genetic test results are sometimes given directly to defense and prosecuting attorneys without help from a physician in understanding the test results. Nearly one in four (14 out of 60, or 24%) of the medical geneticists that answered this question indicated that this "sometimes" happens. This highlights the importance of attorneys and child welfare workers to be able to understand in broad terms what conclusions can be drawn from the genetic test results; they are not expected to have in-depth technical knowledge. However, empirical evidence of how much they do understand about the test results and how they are used in the context of NAI cases has not previously been gathered.

Reasons that low awareness of OI among these specialists could present a problem

Data from my master's thesis research has suggested that awareness of OI among attorneys and child welfare workers is low.[34] If these professionals are not aware of alternative causes of skeletal fractures, they may not have the child examined by an appropriate professional who may be able to rule out OI or request genetic testing.[34] Greater knowledge in this area could help inform policy or educational content that can be implemented to ensure that all people involved in suspected NAI cases are aware of the possibility of OI as a potential alternative cause of unexplained fractures.

Current process of identification of child physical abuse[28]

Medical examination:

Accurate identification of children who have been abused can facilitate appropriate referral, investigation, and outcomes for the patients and their families. A clinical evaluation of the child, including a careful physical assessment and investigation of suspicious injuries may help to identify NAI. Previous research has indicated that the physical evaluation of a child suspected as an NAI victim should include detailed documentation, by way of either body diagrams and/or photographs, of any concerning findings, and the evaluation should include a comprehensive search for any other physical signs that may suggest a non-traumatic cause of the fractures. Involving a pediatrician with a specialty in child physical abuse evaluations could be useful and could help obviate the need for unnecessary invasive testing.

A child that presents with skeletal injuries should be evaluated for any signs of restraint or "grab marks" that could help indicate whether the fractures were likely accidental. Other injuries discovered on the child could also be helpful in gathering information for an evaluation.

In contrast to accidental injuries, non-accidental skin injuries are more likely to occur in non-bony areas of the body, such as the neck, head, buttocks, or upper arms.[35,36] Bruises in particular shapes or size could help identify the mechanism of injury. The intercanine distance of bite marks could also help determine in the injury was inflicted by an adult or a child. If any injuries suggestive of NAI are found, it makes it more likely that the fractures were also due to NAI. However, as mentioned previously, OI and NAI are not mutually exclusive, so a diagnosis of NAI does not rule out the possibility that a child could also have OI.

Social history:

Parents or caregivers suspected of NAI may also be interviewed as part of the evaluation. Documentation should be kept throughout the interview, particularly in regards to the caregivers' explanation of the injury, how consistent their story is with the injury, how consistent their details within the story are, and how consistent their story is compared to witness accounts of the same injury(ies). The interview should also include past medical history, past family history, familial patterns of discipline, child temperament, history of abuse in the family, substance abuse history, social or financial stressors in the family, and any violent interactions between any other family members.

Diagnostic tests for patients suspected to be victims of NAI:

The current recommendations for diagnostic testing for children with skeletal fractures include skeletal surveys of the humeri, forearms, femurs, lower legs, hands, feet, skull, cervical spine, thorax and lumbar spine, and pelvis.[37] There is currently no protocol for determining whether and when a child with skeletal injuries should be tested for OI.

Legal issues:

In the U.S., all physicians are required to report suspected child abuse and neglect, and the physician is not required to prove NAI before a report is submitted. Some states allow physicians to evaluate a child for physical abuse without parental consent, others do not. The physician who submits a report of suspected child abuse may be required to testify in civil or criminal court trial proceedings. Judgments in civil cases are based on a "preponderance of the evidence" in regards to the likelihood that NAI occurred. Judgments in criminal cases are based on testimony regarding the guilt or innocence, and the occurrence of NAI must be proven "beyond a reasonable doubt". The burden of proof in a criminal hearing is higher than that of civil hearings. Literature by the Committee on Child Abuse and Neglect suggests that physicians should not testify to anything that is beyond their level of knowledge or expertise.

Current procedure for suspected NAI court cases

Suspected NAI cases can go through either civil or criminal court, and often goes through both. Civil court cases pertain to whether parents are fit to care for their child(ren) and should be allowed to retain custody of them, and criminal cases answer the question of whether the parent is guilty of the criminal act of having abused their child(ren). In criminal court, the process is relatively straightforward in which there is a criminal proceeding held to determine whether the parent(s) are guilty of the crime of child abuse and neglect. Civil cases are far more complicated and involve a number of steps, as outlined below:

1) A report of suspected child abuse is made and submitted to CPS
2) CPS decides whether to investigate the case. In a case involving a young child/infant with a suspicious broken bone, it is likely that the case will be investigated and it will receive a risk tag for a quick investigation

3) If the CPS investigator believes that the child is in imminent danger, the child may be removed from the home even before the CPS investigation is concluded

4) If the child is removed from the home, the dependency proceeding starts. A petition is filed with the court alleging that the parent has abused the child and therefore the child needs to be in state care until the parent has addressed and corrected his or her behavioral problems.

5) Within a specified about of time (in Washington State, the time frame is three days), a hearing is held to see if removal is (or was) appropriate (depending on the likelihood of risk of further injury at home) and if the child should remain outside of the parent's care while the dependency proceeding is pending (if the child was already removed).

6) Within 75 days of the petition (to remove the child from the home) being filed, it must be decided whether the child is dependent on the state due to the alleged child abuse. This happens regardless of whether the CPS investigation is concluded by then or not.

This decision can, and often does, happen through a settlement. Usually parents will agree to a dependency under another provision which allows the court to declare that they are unable to care for their children rather than that they abused their children.

Alternatively, the decision regarding whether the child is dependent on the state can happen through a fact-finding hearing. The court will then have to find whether the child was abused and whether the child needs to remain in state care. If they remain in state care, they can be placed either with a relative or a licensed foster care provider, but in either case, they are supervised by the state.

7) If the child is found to be dependent, then the parent continues to have the opportunity to get their child back by cooperating with the service plan and making progress to prove that they are fit to have the child return home.

8) If the child has been out of home for 15 of the last 23 months, then the department moves to terminate parental rights, which involves another trial. At that point, the question is no longer about abuse but rather about whether the parent has made sufficient progress to have the child returned and whether it is in the child's best interest to terminate parental rights.

Current practice on removing children from the home

Previous research indicates that when a child is suspected of being abused, and if the risk of serious child maltreatment is high, he or she is often removed from the home and the parents, before an investigation to confirm NAI is completed. The purpose of this removal is to protect the potential abuse victim from further harm. In 2014, approximately 264,746 children in the US entered foster care due to suspected child abuse, neglect, abandonment, or their behavior posed a threat to themselves or others.[38]

Consequences of inappropriate child removal

Although removing a child from their home can be appropriate, it is not without additional risks and consequences for the child. Removal can cause considerable psychological trauma to the child, as it often removes children from their families, or feeding and napping schedules. In addition, removal causes emotional trauma for the parents who have their child(ren) taken away.[39] Furthermore, if the child's injuries are a result of a medical condition like OI, rather than NAI, removal from the home could delay recommended therapy.[40] Using genetic testing to determine whether OI could be a cause of the child's injuries can be helpful, but examining the child for OI may take time, and understanding the test results is not always straightforward.

Challenges of understanding OI genetic test results

Correctly using the genetic test for OI in court cases can be quite challenging, as there are complexities when testing for genetic conditions. For example, one such complexity is the locus heterogeneity of OI. This means that mutations (disease causing variants) in different unrelated genes can result in the same disorder. In OI, 90% of cases are due to a mutation in either *COL1A1* or *COL1A2*, the genes that encode type I procollagen. Over 500 different pathogenic mutations in these genes that cause the phenotype have already been identified[41, 42]. Other genes in which mutations result in OI include the following genes: *BMP1, CREB3L1, CRTAP, FKBP10, IFITM5, P3H1, P4HB, PLOD2, PLS3, PPIB, SEC24D, SERPINF1, SERPINH1, SP7, SPARC, TMEM38B* and *WNT1*.

Another layer of complexity in OI testing is that when results are returned, the test results are not always easy to understand.[43] Sometimes the results come back as clearly positive for OI, which would then offer one explanation for the unexplained fractures, and sometimes the test comes back as negative, making it less likely that genetics plays a role in the fracture phenotype. It is also possible for a patient to have a variant in *COL1A1* and *COL1A2* whose clinical significance is unknown. This may be because the variant is novel, rare, or atypical for the type of alteration that usually gives rise to the condition. [44] In this case, the test is inconclusive and the clinicians are unable to determine if that variant could be contributing to the bone fractures. A result of this type is aptly called a variant of unknown significance (VUS)[45]. An example of a genetic test for OI whose end result was a VUS is shown in Figure 2. Ideally, a laboratory physician or genetic counselor will ensure that whoever receives the test results properly understands what conclusions can be drawn from the test results. However, in some cases, an explanation may not reach the attorney or child welfare worker on the case, or the only

interpretation that the professional receives could be the interpretation write up that accompanies a result report. In those situations, if that professional does not have a scientific expert available to get additional help, he or she must do their best to understand the test result and the implications it has the NAI investigation.

Genetic test results for *COL1A1* and *COL1A2* that come back as inconclusive, or a VUS, are not uncommon. At the Collagen Diagnostic Laboratory (CDL) at the University of Washington (UW), about 6% of all tests for *COL1A1* or *COL1A2* come back as VUS.[46] A VUS may be reclassified at a later date in time if further knowledge about the phenotypic effect of the variant is acquired. It may be then reclassified as one of the following: benign, likely benign, likely pathogenic, or pathogenic. There does not currently exist any regulation as to whether it is the responsibility of the provider to alert patients of a change in VUS status, or if it is the responsibility of the patient to obtain the information themselves. Over time, the diagnostic accuracy of genetic tests for OI is likely to improve, and VUS that are found will be able to be reclassified as either benign or pathogenic mutations, as more information about the functionality of each variant is uncovered.

Confusion surrounding VUS among non-geneticists

Previous research indicates that for many non-geneticist clinicians, the difference between a VUS and a causative mutation may not be clear,[47] and they may proceed as if they have the same value. For a non-medical professional, the distinction is likely to be even more confusing. A participant (medical geneticist) from my master's thesis research stated, "The agencies in charge of protective services are not sophisticated [regarding] genetic knowledge. The judges have difficulty understanding what a VUS is."[34] The Dutch Society of Clinical

Genetic Laboratory Specialists (VKGL) also touches on this issue in a recent article stating, "Extreme caution should be taken when issuing a report of a UV (uncertain variant) to any professional who is not conversant with the complexities of such information. In these cases it is *essential* that careful unambiguous wording is used and it is *essential* to suggest discussion with a clinical geneticist."

Current estimates of VUS in OI vs. NAI cases

Preliminary data collected during my master's thesis showed that just among the physicians that refer patients to the Collagen Diagnostic Laboratory (CDL) at UW, at least 535 patients are referred each year with a question of OI or NAI, but the upper limit could be as high as 1,302. Data from this same research project also indicated that about 37% of these types of cases result in litigation, yielding a range of about 200 – 500 OI vs. NAI court cases per year. As mentioned previously, we calculated from unpublished data from the UW's CDL, that approximately 6% of all tests for OI among patients are returned as a VUS.[34] Given the numbers of OI cases that are estimated to end up in court, this could mean that there are up to 29 OI vs. NAI court cases each year in the U.S. in which the outcome of the genetic test is a VUS. If the professionals that are asked to understand and/or represent the result in court may have difficulty understanding it on their own, and may not be able to get help from an appropriate professional to help them understand it.

International policy on dealing with questions of OI

In Europe, there are recommendations for how to evaluate a child suspected of having OI and what to do if the result is inconclusive. The European Molecular Genetics Quality Network

(EMQN) recommends that *COL1A1* and *COL1A2* are sequenced first when a patient is evaluated for OI with genetic testing. If no mutation is identified and the patient is still believed to have a clinical diagnosis of OI, other genes, including the recessive genes previously mentioned, can be tested for. If an unclassified variant is found in *COL1A1* or *COL1A2*, segregation and functional analysis can be performed to help determine whether the unclassified variant is compatible with a diagnosis of OI. Figure 3 in this dissertation, from van Dijk et al.'s paper, helps to illustrate this. Segregation analysis relies on genetic information from the patient's family members, particularly, the parents, to determine if a pathogenic mutation has been identified. For example, if one parent also has the same variant identified in the patient but the parent does not have frequent fractures, the probability that that variant is responsible for the frequent fractures in the child is reduced. Not identifying a mutation in *COL1A1* or *COL1A2* makes the diagnosis of OI less likely but does not entirely exclude the diagnosis; if the test is negative, the chance of the child having OI is diminished, making NAI a more likely explanation. On the other hand, if genetic testing establishes a diagnosis of OI in a child, the unexplained fractures may be due primarily to the genetic condition. Thus, a diagnosis of OI does not exclude the possibility of NAI, as the two are not mutually exclusive.

Study Hypothesis

I hypothesized that the attorneys and child welfare workers that are involved and invested in suspected child physical abuse cases are aware of OI at a moderate level. I also hypothesized that when genetic test results for OI are returned, there is often some confusion regarding how to understand the test result and the implication the result has for the investigation. To test my hypotheses, I chose to start by conducting interviews and collecting qualitative data, because a qualitative study allows a rich assortment of perspectives to emerge. I then used the themes that

emerged during the qualitative stage to develop the questions that were used for the second (mostly) quantitative aim of the study, the survey. A quantitative approach allowed me to obtain further, detailed information about each theme, and due to the larger sample size, greater power. A quantitative approach also allowed me to test whether there are differences between groups.

Innovation

This study helped to elucidate the general level of knowledge that attorneys and child welfare workers have of OI, the general ways in which the genetic test results for OI are used in NAI cases, and what the participants' experiences with medical experts in NAI cases has been like. No previous studies had been done on this topic. It was also a unique study in that it was be multidisciplinary, and included policy, law, social work, genetics, and bioethics.

Chapter 3: EXPERT INTERVIEWS

ABSTRACT

Introduction: Non-accidental injury (NAI, or physical abuse) is a worldwide pediatric concern. The prevalence of NAI with fractures is ~24/10,000 in the 0-3 year range. Osteogenesis imperfecta (OI), which is characterized by fragile bones, is the most common genetic condition confused with NAI (prevalence ~1/10,000). Genetic testing can be used to help identify OI in the context of evaluation for NAI.

Materials and Methods: To understand how genetic test results for OI are used by professionals involved in child welfare or child abuse law, one-on-one semi-structured telephone interviews were conducted with 6 child welfare workers and 8 public defense attorneys (representing parents) who were known to have some experience or involvement with this issue. Interviews were recorded, transcribed, and coded thematically by two separate coders.

Results: On the basis of these interviews, these professionals are not always trained about causes of skeletal injury besides NAI. Given that the overall genetic education background of the participants is low, there is the possibility that test results could be misunderstood and misrepresented in a case. Medical experts are not always helpful in offering a non-biased assessment of whether abuse has occurred. The use of genetic testing might be limited by financial constraints, policy, or guidelines.

Conclusions: While genetic testing can identify children with OI among those investigated for NAI, testing also could come at some risk. Misunderstanding of genetic test results in the hands of a professional untrained in genetics could lead to deleterious consequences for a family. This study was a small, exploratory study that provides responses and opinions from a very specific

group of participants. The findings from this research cannot be generalized beyond the study participants, and further research on this topic is advised.

Background/Introduction:

Children typically have relatively strong bones that only fracture after physical trauma such as an accident, fall, or inflicted injury. However, some children have a medical condition that result in fragile bones that break more easily than they should. Upon a child's entry to the hospital, fractures due to physical abuse, or non-accidental injury (NAI), and fractures due to a medical condition that causes predisposition to fractures are not always easy to distinguish from one another. Genetic testing can determine if a heritable condition, such as osteogenesis imperfecta (OI), could provide an alternative explanation for the fractures.[9]

OI is a rare genetic disorder characterized by increased risk of bone fractures. The incidence is about 1:10,000, which corresponds to the birth of about 400 children each year with OI in the US.[48] It is the most common inherited condition leading to fracture predisposition.[3] Leventhal et al. estimated that in 2003, approximately 12% of children hospitalized with fractures had fractures due to NAI, however, among children hospitalized with fractures due to abuse (and no head trauma) about 0.85% of them have OI.[19] If the populations of these children are combined, we expect that about 1.3 per 100,000 or about 7.5% of this combined group of children with unexplained fractures will have an underlying bone disease.

The clinical determination of whether a child has suffered physical abuse can take into account a physical examination, social history, and other factors (see Chapter 2 for a more detailed explanation). However, a clinical examination alone may not be enough to determine whether OI could play a role in the fracture phenotype.[1] For this reason, genetic testing is sometimes needed to help rule out OI.

Many people are involved when an allegation of child abuse is made, and the timing during the investigation in which the stakeholders are brought into the picture could vary. For

example, if a child is brought into the emergency room with unexplained fractures, the emergency room (E.R.) doctor would be the first professional involved in the case. In the U.S., legal statutes require medical doctors to report any suspicion of child abuse to law enforcement or child protective services, so these professionals would be brought onto the case at that point. If the case progresses to court, two attorneys are needed (both prosecuting and defending, for the state and for the parents), and medical experts for the case may also be brought in. If genetic testing is considered, the test can be requested by the attending physician, and the judge can demand that a test be arranged. We know from previous research that when genetic tests are done in cases in which there is a question of OI vs. NAI, the test results are sometimes returned back to the attorneys or child welfare workers who are working on the case, and they do not always go through the medical expert who originally requested the test.[34] For this reason, it is useful for those involved in the representation or explanation of the genetic test results to have an understanding of what conclusions can or cannot be drawn from the results.

This qualitative study examined the opinions of defense attorneys and child welfare workers about cases involving children with unexplained fractures, in which there is a question of whether the fractures should be attributed to NAI, OI, or both. The study explored the following topics: (1) Extent of academic or vocational training about genetics or genetic conditions that could predispose a child to fractures, (2) Awareness of and experience in dealing with OI cases, (3) Levels of comfort with reading scientific literature and confidence in colleagues to understand genetic reports, (4) Experience working with medical experts, and (5) Efficacious ways to raise awareness of OI and other disorders that could lead to fractures among attorneys and child welfare workers.

Methods:

Identification of Study Participants

To capture robust participant considerations on this topic, both purposive sampling* and snowball sampling* were used identify subjects who were believed to have opinions regarding the questions. In terms of purposive sampling, both the first child welfare worker and the first attorney that were contacted were recommended by an advisory committee on the research team. Then a snowball sampling approach was employed: each of these two subjects suggested at least one other professional to contact, and each of those suggested at least one other. In total, fourteen individuals participated in the study, six child welfare workers and eight attorneys. Each individual was initially contacted by email, usually with the help of an email introduction from the professional who had recommended them, with a request to participate in a telephone interview to assess their opinions on the use of genetic testing in child abuse cases. Participants were asked to email me back if they were interested in participating. A maximum of two follow-up emails were sent to any one potential participant. If participants did choose to email me back with a statement that they were interested in participating in the study, a time to call was coordinated by email.

Interview Guide

An interview guide was written before interviews commenced and was reviewed by an advisory committee for clarity, depth, and time appropriateness. Modifications to the questions were made where necessary to increase the likelihood of capturing the most informative responses. Numerous additional probes were included for each question on the guide.

All interviews were conducted over the telephone by a single interviewer (EY) trained in standard interview techniques. Each interview lasted between twenty minutes and an hour. Study identification numbers were assigned to participants to protect confidentiality. Interviews were recorded, transcribed, and de-identified. Two coders (EY and CE) individually coded each interview for content analysis using the digital transcripts. Research has indicated that multiple coders are suitable for mid-size datasets where there is adequate similarity between responses.[49] Emergent, thematic analysis* was also conducted by the first author. (* See Qualitative Research Terminology Glossary in Appendix 1 for definition.) This combination of qualitative and content analysis helped to identify novel participant responses and to obtain a more thorough understanding of those responses, while the quantitative aspect offered by both coders allowed for increased precision and ability to confer intercoder reliability.*[50]

Qualitative content analysis* has been described in many ways. It is used primarily as a quantitative research method in which text data is coded into distinct categories and described using statistics.[51] More specifically, it has been defined as a research method for the interpretation of the content of text data through the methodical classification process of coding* and characterizing themes that emerge from the data.[52] Researchers consider content analysis as a flexible method of analyzing text data,[53]and the coding procedure allows for qualitative data to be reported as quantitative frequencies.[9]

The thematic analysis* approach used for this study has been described as subtle realism.*[54,55] Theoretical and methodological perspectives were realized after two thorough readings by each coder, and by the creation and revision of a content analytic codebook.* During the first reading of the transcripts, focal themes were identified for each participant. Theme identification was done immediately after each interview took place to help researchers decide

whether content saturation had been met, defined by a repetition of themes or cessation of novel themes. This process formed the basis of the decision to terminate interviews after eight attorneys and six social workers had been recruited and interviewed. Before the second reading of the transcripts, the themes that were gleaned during the first reading were classified into ten clusters. During the second reading, each transcript was assessed for the presence of each of the themes and clusters of themes. This was done as a quality control measure to ensure that any one theme was not missed in the first read.

Using the identified themes and thematic clusters, a content analytic codebook was developed (EY) and reviewed by both coders. Previous research has indicated that this form of content analysis is appropriate for ascertaining relatively specific themes, such as those characterized in this study. A standardized codebook and numerous repetitions of coding to achieve coder agreement are necessary to obtain greater accuracy in the codes, precision in the clusters, and further intercoder reliability.[6]

The first draft of the codebook was tested on the first six transcripts by the second coder, who at the time was uninvolved with the study. Revisions were made to the codebook to improve clarity, and transcripts that had been coded with the first drafts of the codebook were reread and recoded with the final version of the codebook. Code results from both coders were compared for intercoder reliability. For any cases in which kappa was less than 0.80, coders met for a resolution discussion, changes to the codebook were made, and the items were recoded until that level of agreement was achieved.

Results

Participants:

Six of the participants self-identified as child welfare workers, eight were attorneys. Half (n=3) of the child welfare workers interviewed were employed by a county or state office of public defense providing services to children and families. Two of the child welfare workers were affiliated primarily with a university, and one was affiliated primarily with a hospital. Three were men. Among the attorneys interviewed, five were under contract with a state or county office of public defense to represent parents in dependency proceedings, and the other three were employed by a private firm that represents parents in dependency proceedings. Three of the eight were men. See Table 1 for demographic details.

Responses:

Twenty-five codes* were created by the end of the final coding and conflict resolution round. Over fourteen participants, this resulted in a total of 350 coded items. Percentage agreement between coder one (EY) and coder two (CE) for all items coded was 99.43%.

Genetics Education

In response to the question asking participants to describe their history of education around genetics or genetic testing, 71% (n=10) had never had any training in genetics, while 29% (n=4) had an overview of genetics as part of their undergraduate biology class. Seven (50%) had done some self-education in genetics. For example, P8 (numbered codes were assigned to participants to protect anonymity and for brevity) stated that self-education comes in the form of "books, magazines, peer review journals…related to the human body, genetics".

Awareness of OI

All but one of the participants agreed that their overall awareness of medical conditions that could contribute to bone fractures was low and should be raised. Stakeholders inquired about included attorneys and child welfare workers (including CPS workers), but participants also brought up the need for awareness among law enforcement officers and judges. Four participants made statements specific to the lack of attorney's ability to comprehend genetic test results.

Participant P11 said of CPS workers,

"*I believe that most child protective services in most areas are lacking in [awareness of OI]…I shouldn't immediately jump in and say who's to blame… I should be thinking about what else is going on here- I don't think that's a thing*".

Although not explicitly asked about, some participants brought up law enforcement officers as an important group to train about medical conditions such as OI, as this group is often involved in NAI investigations before a trial begins, when it is easier to request genetic testing, according to one participant. Those that mentioned the need for training among law enforcement also acknowledged a current deficit regarding genetic knowledge among this group. P11 (attorney) stated,

"*law enforcement agents are in no way trained on any thought process, you know, about medical, biological, genetics, sort of, issues*".

Other similar sentiments were expressed. Judges were also brought up by participants as stakeholders that should have greater awareness of OI, and three participants indicated that judges are the most important profession to train, as it is ultimately up to them to make the final decision regarding the presence of a genetic cause of the injuries.

Three participants indicated that judge awareness is of the utmost importance in order to make an informed decision in situations in which there is disagreement between scientific

experts, and a "battle of the experts" ensues. In explaining prior experiences with battles of the expert, P8 (child welfare worker) stated,

"*sometimes it's literally the judge, who presumably doesn't have a medical background, relying on whatever [genetic] data's in front of him [to make a decision]".*

The one participant (child welfare worker) out of the fourteen that did not agree that overall OI awareness among all stakeholders was low stated,

"*Especially if you're doing CPS work…I'd be SO surprised if they didn't know [about OI]. Like, stunned*".

This participant was also very confident in the ability of a medical expert to properly detect whether a child did or did not have OI. Other participants were not as convinced, as explained below.

Use of medical experts

Thirteen participants agreed that medical experts are not used as frequently as participants felt that they should be in these cases, despite twelve of the fourteen indicating that they do have easy access to a medical expert upon request. While three participants cited the expense of the medical expert as a reason that medical experts are not used, three other participants stated that getting a medical expert is too much of a hassle for already overworked professionals. Six others indicated they have no idea why more people do not get experts, particularly because many states have a budget specifically set aside for scientific experts. If experts are not requested, in some states this means that

"*tests can be done, but then there's no one to help interpret what the tests mean*", according to P12 (attorney). Indicating that non-genetics professionals might not understand what conclusions can be drawn from the test results.

On the other hand, some participants indicated that using medical experts can sometimes be detrimental to a case, depending on the expert. Four separate participants indicated that some medical experts are given too much authority, or what one called "an abuse of power" within the court system that allows for experts to assume that a child's injuries are due to NAI. It was stated of the experts,

"*they refuse to acknowledge that the bone fractures could have been caused by anything other than abuse*".

According to one attorney, this last statement is true even in children with a family history of "weakened bones", genetic disease, or low vitamin D. Another attorney referred to her local medical experts as "villains" and "so called child abuse doctors", in reference to two of her cases in which she believes that evidence was clear that her clients were innocent, and due to the experts belief that,

"*the villains…have already made up their minds that unexplained fractures in infants happen…because their parents were pounding on them…they call the cops and they call CPS and that's it…These doctors come up with these things, and it turns out to be crap, but you know, the people they've harmed aren't really in a position to do anything about it*".

Consequences of the "abuse of power"

Participant P13 (attorney) recounted one case in which a child was removed from his mother due to a doctor's suspicion of NAI after the mother's first child died soon after having been removed from her, despite having been found to have a chromosomal abnormality causing physiological defects. According to this interviewee's account, the court made the mother feel as though,

"*all this is her fault as opposed to the doctor's fault, and it's just, it's really a nightmare… [there was] no evidence that the mom had ever done anything wrong or no*

concerns from anybody, and [the doctor] can look at that report…and say she killed that kid?...The idea that you can be in that position to say that and say it with a lot of certainty as a doctor is terrifying to- it's just scary".

P14 (attorney) recounted two cases in which families were separated on account of suspected child abuse, and in both cases, the defense won (after genetic testing was done) and the families were able to be reunited. This participant said of the situation,

"*In both cases, I believe my client was entirely innocent. They suffered huge harm, they were separated from their child for well over a year in both cases, they had to endure humiliation, they had to endure counseling for domestic violence and mental health issues, they are out thousands of dollars, and you know, their family was traumatized because, you know, tiny children were placed with strangers…they're terrible cases, they're so frickin' unjust….I can't give these people a year of their children's lives back, and at the end of the day, the system is patting itself on the back because they fixed this family*".

Two participants indicated that because of the position of authority that child abuse experts have, once they declare that a child has been abused, the court will often side with them and take steps to remove the child, and it becomes infinitely more difficult for the defense to prove to the court that their client is innocent. Participant P5 (child welfare worker) remarked,

"*It shouldn't be like, alright the kid's got broken bones, let's find out who did it and then we're done and we don't look at any information that got gathered afterwards. That's not reasonable, that's not realistic*",

yet participant P13 stated that this often happens because there is "zero incentive" for child abuse experts to admit that they made a mistake or they overreacted, and

"*as long as [the child abuse expert] acted in good faith, he's off the hook liability-wise…they have no problem adopting a child out, you know, based on [the expert's] say-so*".

Medical experts as mandatory reporters

Many participants expressed some doubt as to the qualifications of a medical expert to distinguish between NAI and medical conditions that could cause predisposition to fractures. A few indicated that they believe that more medical experts are quick to report NAI in children

because of the legal statute in all states that require numerous professional groups including doctors, social workers, and teachers, to report any children with signs of abuse to law enforcement or a similar authoritative body. Participant P11 (attorney) commented,

"*once all these statutes went into place about all these doctors being mandatory reporters…they don't take the step of checking…for a biological answer, they just report*".

While participant P6 referred to this as using, "an abundance of caution", participants P13 and P14 (attorneys) referenced the child abuse "panic" that is causing many professionals to quickly conclude NAI without a further examination, citing that this very "panic" or "heyday" happened in the past with Munchausen's, sexual abuse, shaken baby syndrome, and is now happening with NAI. P12 (attorney) commented that in some states,

"*physicians (are) weighing in as child abuse experts, and a lot of people are saying that's not necessarily qualification…especially without examining a child*".

P4 (child welfare worker) stated that often, social workers choose to work with local physicians rather than experts because they already have a relationship with their local physicians, and P13 (attorney) highlighted doctors' overuse of the res ipsa loquiter doctrine to try to prove NAI. (The res ipsa loquitur doctrine--the thing speaks for itself, in Latin--from the common law, concludes that if the injury happened in the first place, that proves that the defendant must have been negligent.)

<u>Selected anecdotes from participants on their perception of biases within the legal system</u>

Nearly half (n=6) of participants highlighted issues that they have personally faced within the defense system that made it difficult for their clients' families to receive the best care. The reasons were varied. Participant P10 (attorney) made the following statements when asked why genetic testing for conditions like OI isn't used more often when a child is suspected of NAI,

"*people are afraid of the consequences if they, kind of, mount a strong defense against these cases….clients have pled guilty rather than going to trial, getting the experts to look at the genetic issues, because somebody's offered them something that's easy- that's easier a lighter sentence or some kind of an agreement with CPS…if you keep saying you didn't [cause the injury] you're in denial, so then, you're still at risk. You pose a risk to this child, therefore, you don't get your child*".

In other words, participants indicated that if a defense is mounted against an allegation of child abuse, it can end up resulting in a worse outcome for the parent because it seems that the parent is in denial of their actions. If the verdict is that the parent abused the child, the outcome is worse for the parents than if they had confessed from the beginning, because now they are believed to have been both abusive and in denial about their behavior.

Participant P9 (attorney) said,

"*[Testing] doesn't always happen. I think sometimes attorney's get faced with the fear of, you know, my client could face criminal charges, we should just agree to the dependency*".

<u>Selected anecdotes from participants on social biases within the system</u>

Two participants indicated that they believe that some families are more likely to be suspected of child abuse. Participant P12 (attorney) remarked,

"*family court has created a whole class of parents who are always suspect…abuse is generally related to poverty…just finding out how much they're penalizing people that don't have money, based exclusively on that fact is nauseating*".

Another participant indicated that they have experienced a problem in some situations in which defense attorneys are not allowed to speak to their social workers, due to historic bureaucracy. P12 explained,

"*there are places where defense attorneys don't even know the first thing about their social workers…they're asking on cross what their qualifications are, they don't even know those".*

Case examples from participants

Seven different case studies were recounted by interviewees during the course of this study. All indicated that they believe there is a need for professionals involved in NAI cases to have greater awareness of alternative causes of bone fractures other than NAI. Participant P9 (attorney) recounted a case study in which a mother was sent to prison for child physical abuse because fractures were discovered in the child; she was later exonerated due to genetic test results, but it's been over six years and she is still fighting to get her child back. Participant P10 (attorney) recounted a case in which two siblings had signs indicative of child physical abuse so they were removed from the home and adopted by a different family. It was later determined that the fractures were due to OI, but the parents are still unable to get their children back who now only know their adoptive family. Participant P11 explained a criminal case in which the child was removed from the mother on suspicion of child abuse, despite the mother having OI. It was difficult to get the child tested for OI because the child was not in front of the court. Testing never happened on the child because the parents decided to plead guilty.

Many participants pointed to fact that the consequences for mistakes are very high. Participant P14 exclaimed,

"It's a child!...Children die when somebody makes a mistake!", and participant P12 commented that "termination of parental rights…is basically the civil death penalty".

Discussion:

This study was undertaken to explore the opinions and experiences of a small group of attorneys and child welfare workers that have been involved in OI vs. NAI cases. No other studies on this topic have previously been done.

The findings indicate that the levels of awareness and specific knowledge of genetics and OI are varied among attorneys and child welfare workers. All interviewees highlighted the need for those involved in child welfare cases to be knowledgeable about genetic issues pertinent to child abuse investigations, and all but one believed that within their own profession, the current level of OI familiarity was lower than they believe that it should be. Attorneys in this study were on average more willing to provide more information than child welfare workers in this study, and were more willing to indicate how they think other professionals within their field would answer the research questions.

Participants' comments regarding the frequency with which medical experts are not used in NAI investigations were surprising. It was also surprising that participants indicated that even if there is no medical expert available to help an attorney or child welfare worker understand a genetic test result, the tests might still be done anyway. When experts are used, a number of participants indicated the frequency with which experts from the defense and prosecuting sides of a case disagree about what a test result means, resulting in a battle of the experts. Participants also indicated that often in those cases, it is up to the judge to decide which expert is making the strongest case, regardless of the judge's previous education or training in genetics. Relating to this point were a number of participants' comments indicating doubt in the experts' ability to understand a genetic test result. Since there are no federal guidelines for expert qualifications, selection of experts differed by region. In some areas, participants stated that attorneys prefer to use local physicians to help them understand genetic test results because they have a personal relationship with them, despite the fact that the local physician may not be a geneticist.

Four participants were insistent that child abuse specialists have too much authority in child abuse cases, and that they too frequently assume that a child's fractures can be due to no

other cause than NAI. This could lead, as one participant pointed out, to attorneys and child welfare workers opting not to use medical experts for fear that they will invariably conclude that the child's injuries should be attributed to NAI.

Some interviewees indicated that in certain situations, after an accusation of NAI is made, the family is advised by their attorney that they should not pursue genetic testing. This advice might be given because, often, refuting an NAI allegation could indicate that the defendant(s) is in denial of their actions which could ultimately lead to a more severe outcome. One participant indicated that if a family refutes an allegation they could be told, "*you're in denial, so then, you're still at risk. You pose a risk to this child, therefore, you don't get your child*". This leaves parents then with the option of either confessing to having abused their child(ren) and hoping for an easier outcome, such as having the children be put into care with a relative for a short period of time, or entering into a trial that could result in a more severe penalty for that family.

Overall, participants agreed that changes in education or policy could be useful. Professional groups that participants indicated were most in need of greater awareness included attorneys, judges, child welfare workers, and law enforcement officers. One participant summarized the way attorneys sometimes deal with child abuse, and this sentiment was echoed by other participants (in reference to other professions as well): "*here's the crazy part…child abuse is such a hot button issue that…if someone says there's no other reason for that injury to exist in a child, a lot of defense attorneys kind of throw up their hands...that's certainly not ideal public defense*".

Conclusion:

Given the complex nature of NAI cases, and the monumental effect that the outcome of the case can have on a family, it is important that all aspects of the case are handled correctly. Findings from this exploratory study suggest that perhaps there are components of OI vs. NAI cases that could be improved upon, but the findings also indicate that more research is needed on this topic. Since the study sample size was quite small and the participants were recruited using purposive sampling, these findings cannot be generalized beyond this study population. The aim of this project was to collect different ideas, perspectives, and opinions on this topic from a small group of professionals in order to help inform a more robust quantitative study (explained in chapter 4). While a qualitative study allowed for a rich assortment of perspectives to emerge during the interviews, it also presented challenges in data analysis and the ability to perform statistical analyses on the responses. The next chapter contains details on an anonymous survey that was informed by findings from the study described in this chapter, and was able to reach a significantly larger sample size.

CHAPTER 4: SURVEY DATA

ABSTRACT

Background: Osteogenesis imperfecta (OI) is a genetic condition that could be confused for child physical abuse in the context of evaluation of unexplained injury in a child. Preliminary data on a small population suggested that professionals involved in evaluation of these children are not always trained to consider medical causes of skeletal fracture, such as OI, and when a genetic test result for OI is returned to a non-geneticist professional involved in the case, it has the potential to be misunderstood. This study was designed to further explore the findings from previous qualitative research done on a small sample size.

Methods: A multiple (mixed) methods* survey was designed and distributed to attorneys and child welfare workers involved in non-accidental injury (NAI) cases. The survey included 46 questions, and participants were drawn from a number of sources, all of which included an email invitation from a third party explaining the purpose of the survey and a link to the web-based survey. The survey was divided into five primary topics, each including a primary question and one or two ancillary questions. Point estimates were calculated for each of the quantitative questions, and survey results were summarized using descriptive statistics.

Results: A total of 192 participants started the survey, 185 of whom completed more than 70%, and 102 of whom completed 100%. Findings from this research suggest that most study participants are aware of OI, although they estimate that OI awareness among others in their profession is low. While study participants are not typically educated in genetics, they rated their confidence in their ability to understand scientific literature highly. While slightly more than half of participants have easy access to a medical expert to help them interpret genetic test results,

only about two-thirds of those are likely to be able to get help from a geneticist. Overall, participants wanted to learn more about OI and genetic testing and agreed that a policy or practice change could be beneficial to their field.

Conclusions: A lack of awareness of conditions that could be responsible for unexplained fractures, or a misunderstanding of their diagnostic tests could be problematic in the context of a NAI case. Education about OI or other causes of fracture and policy and practice guidelines and protocols might help professionals in their assessment of children with unexplained injuries in which NAI is a major consideration. However, numerous limitations in this study restrict the generalizability of the findings and further research on this topic is required.

Introduction:

Genetic discoveries are occurring at an unprecedented rate, and the use of genetic information and genetic testing is quickly becoming incorporated into many aspects of our everyday lives. One such example of this is the use of genetic testing for the inherited condition osteogenesis imperfecta (OI) during investigations in which a child is suspected of having been physically abused. Since OI is characterized by a fragile skeleton, unexplained fractures often occur in infants that have OI, though it may not be apparent that he or she has a genetic condition if the case is mild and the child has no other accompanying features, such as blue sclerae, bowing of the long bones, or joint hypermobility. OI is a rare condition, with an incidence of about 1 in 10,000 live births, which corresponds to the birth of about 400 affected children each year in the US.[56]

Non-accidental injury (NAI, or child physical abuse), on the other hand, is quite common. In 2003, the estimate of the incidence of hospitalization with fractures due to abuse among children aged 12 months or less was 36.1 per 100,000. The incidence was 15.1 per 10,000 for children aged 0-3 years.[19] In a population of children hospitalized with fractures (and with no head trauma), approximately 12% have fractures that can be attributed to abuse, and slightly less than 1% of them have OI.[4] Unfortunately, upon admission to a hospital, the cause of the fractures is not easily discernible. A previous study found that clinical evaluation alone may not be enough to distinguish fractures due to NAI from fractures due to OI.[9] This emphasizes the potential benefit of genetic analysis in this population of children with unexplained fractures.

The difficulty in genetic testing for OI is multifold. Genetic test results are not always easy to understand, even for some medical professionals.[33] When genetic testing is used as evidence in a courtroom, the people charged with presentation of the test result are often non-

geneticist professionals. For example, previous research has indicated that sometimes the test results could be returned back to a defense attorney, a prosecuting attorney, a child welfare worker, or a scientific expert involved with the case.[34] Other research has shown that when non-geneticist professionals make attempts to handle genetic information without help from a geneticist, tests could be misunderstood and cause negative outcomes.[33] Previous qualitative interview-based research has been done (chapter 3 of this dissertation) to explore what attorneys and child welfare workers know about OI, how much education they have had in genetics, and how they navigate OI vs. NAI cases. Interview results from that study suggested that attorneys and child welfare workers involved in NAI cases are not always trained about causes of skeletal injury besides NAI, and that testing might be limited by financial constraints, policy, or guidelines. Results also indicated that when genetic testing is done, attorneys and child welfare workers do not always have access to a medical expert that is qualified to help them understand the test results, and medical experts brought into a case are not always helpful in offering non-biased judgment regarding whether NAI has occurred. More research is required to further examine these preliminary findings.

Previous research on attorney and child welfare worker comprehension of genetic test results

Little research has thus far been done on how attorneys and child welfare workers interpret genetic test results to be used in court. A research study from Australia found that reports of DNA evidence for use by non-geneticists attorneys contained language that was difficult for attorneys to understand, which was compounded by the use of specialist terms. Moreover, the sections of the report were not in a logical sequence, and the appendices of the report used very small font, with single spacing, and tables that spanned several pages, which could lead to difficulty comprehending the material.[57] Other research concluded that scientists

have an ethical responsibility to assist non-scientists in understanding scientific evidence to be used as decision-making aids in court, and that the communication of scientific information to non-scientist audiences is an under-researched issue. This same group found that scientific reports are often written at a level difficult for any non-scientists to read, for reasons such as high lexical density (information content per clause), and could therefore impede their understanding of the results.[58,59] No previous research has been done on child welfare worker's comprehension of scientific reports.

Purpose of the Study

The purpose of this study is to learn more about what child welfare workers and attorneys involved in child abuse cases know about OI vs. NAI, how much education they've had in genetics and how comfortable they feel reading scientific literature, what it's like working with medical experts during a case, and whether they are interested in learning more about OI. I hypothesize that the study will reveal a moderate awareness of OI among relevant professionals, and a low scientific literacy among these professionals. I hypothesize that the professionals will be receptive to learning more about OI.

Preliminary data

Qualitative Interviews: In the spring of 2016, 14 one-on-one semi-structured telephone interviews were conducted with participants that were known to have some experience or involvement with this child abuse cases. Six of the participants were child welfare workers and eight were public defense attorneys. Interviews were recorded, transcribed, and coded by two separate coders. Emergent, thematic analysis was done on responses. Full methods for this study

were described previously (chapter 3). The information obtained from this study was used to inform the multiple (mixed) methods survey used in this study.

Methods:

Design

A cross-sectional multiple (mixed) methods approach was used that incorporated quantitative and qualitative components in a single survey. This study was approved by the University of Washington Institutional Review Board Minimal Risk Committee. A 46-item internet survey was developed using data and key points obtained from the qualitative interviews.

Target population

Our target population for this aim was child welfare workers and attorneys who are actively involved in NAI cases. The sample size for this aim was determined by using the department for social services (DSS) sample size online calculator,[60] and with help from the University of Washington's Biostatistics consulting services. A sample size calculation was computed using one-sample (average) values, two-way testing, with alpha error level (confidence level) set at 5% (95% confidence interval) and beta error level (statistical power) set at 80% (1-beta). Standard deviation was set at 1.0 as is typical for sample size calculations. A sample size table was constructed for various estimates of the test value and the sample average, with increments set at 0.25 to capture the range of sample size required to meet the 80% power and 95% confidence interval parameters. See Tables 2a and 2b for complete sample size details. The two tables both contain sample sizes with identical parameters and 0.25 increments of estimates for test value and sample average, but Table 2a spans test and sample values from 0-

0.225, and Table 2b spans test and sample values from 0.7-0.9. Both tables indicate that if the test value differs from the sample average by 0.075 or more, than the sample size needs to be equal to or greater than 115. If the test value differs from the sample size by 0.1 or more, the sample size should be equal to or greater than 65.

Recruitment and Procedure:

Participants were recruited for this study from a variety of sources. To recruit attorneys for the study, state offices of public defense were contacted to ask for help in distribution of the survey, but responses were only received from Louisiana and Washington State. While state offices include both attorneys as well as child welfare workers, only attorneys responded to this method of recruitment. Other attorneys were recruited with the help of a defense attorney from Louisiana who sent the survey out to a small private listserv of public defenders that represent parents in child welfare cases. Each entity that agreed to send out the survey on my behalf sent out a short email blurb explaining the study and including a link to the survey hosted on Qualtrics Survey software. After realizing that no child welfare workers were responding to the survey, a monetary incentive was added to the survey, and a plan to target child welfare workers was launched. State chapters of the National Association of Social Workers (NASW) were contacted in all 50 states to request that they survey go out to each state chapter, but only the states of Ohio and Michigan were willing to include the survey information in their newsletter. An email blast was also sent out on our behalf to a subset of a listserv managed by the NASW that included only social workers involved in child welfare. 1000 email addresses belonging to child welfare workers on this listserv were selected at random. The email that was sent out to these different entities can be found in Appendix 2. While the NASW mailing list is largest mailing list available for social workers working in CPS, it is not representative for all child

welfare workers as most of them do not have a social worker degree. Despite this, it is a very solid source of social workers with experience in child welfare and child protective services.

Measures

Questions were constructed to address key issues raised by interview participants or suggested by members of the dissertation committee. A full set of the questions can be found in Appendix 3. For the quantitative data component of the survey, a combination of multiple choice, likert scale,* numerical fill in the blank, and matrix-style questions were used. For the qualitative data component of the survey, open textboxes were available at the end of many questions in order to allow participants to provide more detailed information and comments to enrich the analysis, and one single short answer question was asked. The responses to all text-entry questions/responses were evaluated for emerging themes by qualitative content analysis.[52]

Question topics

Questions that were asked in the survey included the following research questions in five categories. Each category includes a primary and an ancillary question. The ancillary question is designed to add more information to the primary topic.

<u>OI Awareness</u>

Primary question: How aware are attorneys and child welfare workers about OI?

Ancillary question: Do attorneys and child welfare workers receive training on the job about OI?

<u>Genetics/Science Familiarity</u>

Primary question: To what extent have attorneys and child welfare workers had formal education in genetics?

Ancillary question: How comfortable are attorneys and child welfare workers with reading scientific literature?

Ancillary question: Do attorneys and child welfare workers know what a variant of unknown significance (VUS) is?

Factors that contribute to whether a child will be tested for OI

Primary question: Why doesn't genetic testing happen when it is suggested?

Ancillary question: At what stage of an investigation or trial should genetic testing be requested if OI is being considered?

Medical experts in OI vs. NAI cases

Primary question: How easy is it for attorneys and child welfare workers to get help from a medical expert?

Ancillary question: How likely is it that they will be able to get help from a geneticist?

Ancillary question: Are the medical experts unbiased in their opinion of differential diagnosis of OI vs. NAI?

Direction of Future suggested policy

Primary question: Are attorneys and child welfare workers interested in learning more about OI?

Ancillary question: Are attorneys and child welfare workers interested in suggestions for policy or practice guidelines surrounding OI vs. NAI cases?

Analyses

Quantitative Analysis

Point estimates, or prevalence rates, were calculated for four of the five primary and ancillary questions using numeric values assigned to each answer choice (calculations were not done on the questions pertaining to 'Factors that contribute to whether a child will be tested for OI' which is described qualitatively). To determine the "prevalence" of each of the quantitative questions, values for each answer choice were recoded to make each question outcome binary, if they were not already. For example, for the question relating to the amount of genetics education that participants had had, participants' responses were separated between those that had had at least one entire class in genetics education and those that had had less than one complete genetics class. This point estimate of the study population gives an estimate of the "prevalence" of a background in genetics education among the participants. For questions in which binary responses were divided into Yes or No categories, the value one was assigned to affirmative answers while the value zero was assigned to negative answers.

Qualitative Analysis

Open text responses were analyzed using a coding framework to identify broad themes in the participants' experiences and opinions. Only one question was entirely open response, the others include optional write in boxes if participants desired to further explain their answers to a close-ended question. The data collected from the open text boxes was used to enrich the quantitative analysis and provide commentary on the trends seen in the statistics.

Results:

Survey completion

The survey was emailed to approximately 10,405 potential participants via direct email or via a work-associated newsletter. The methods used are listed below:

- 150 direct emails with survey links were emailed out, with the help of an attorney from Louisiana, via a small private listserv of public defenders that represent parents in child welfare cases (attorneys).
- 200 direct emails with survey links were sent through the Washington State Department of Public Defense, (40 of whom were attorneys, the rest of whom were child welfare workers),
- 6000 links were sent through the Michigan State National Association of Social Workers chapter newsletter (emailed)
- 3055 were sent out to the Ohio State National Association of Social Workers chapter newsletter (emailed),
- 1000 were sent through a purchased portion of a listserv of child welfare workers (purchased from the National Association of Social Workers (NASW) via InFocusMarketing).

Because of the third party emailers that were involved, the only email addresses that I had access to were those few people that agreed to forward the email on for me. Only one email was sent out to all of these listserv groups. In addition, because of the third party involvement, there was no way to track how many of these direct emails bounced back (undelivered), routed to spam, or were unopened. From the listservs that included surveys sent through the NASW Ohio and Michigan State chapters' newsletters, it was recorded that their open rate was 29%, and 40%,

respectively. From the listserv purchased through NASW, 12 recipients unsubscribed. If I exclude these unopened and unsubscribed emails from the total potential participants,[61] but assume the other emails had a 100% open rate, the total number of potential subjects that were invited to participate in the study was 4,624 rather than 10,405.

The survey was partially or fully completed by 192 (4.15% total consent rate). The total proportion of participants by profession was about 34.3% attorneys (n=66), and 36.5% child welfare workers (n=70). The response rate within each professional group was 18.9% for attorneys, and 1.6% for child welfare workers. The most probable reason for the discrepancy in response rate between the two professions was that there were specialized listservs available to contact the most appropriate attorneys who were interested in the subject material. Such specialized listservs for this area of social work do not exist, so a slightly wider net was cast in order to capture enough of the appropriate individuals. The final response rate among child welfare workers was not unexpected given that only 3% (2050 out of 56,175) of the entire NASW listserv comprises professionals involved in child welfare. A more extensive discussion on this can be found in the limitations section where the effects of non-response bias are discussed.

29.1% of participants (n=56) chose not to answer the question regarding the specialty that they most identify with. The survey was completed by 102 (53% completion rate). 43% of total participants (n=83) responded to at least 70% of the 46 questions. The full raw results from the survey can be found in Appendix 4.

Backgrounds of the Participants

Of the total 192 participants, 34.4% (n=66) identified as attorneys. Of those, 22.7% (n=15) identified as criminal attorneys, and 77.3% (n=51) identified as civil attorneys. 36.5% of total participants identified as child welfare workers (n=70), with 10% (n=7) identifying as working for a service that represents parents, and 14.29% (n=10) identifying as having some other child welfare related profession. Table 3 illustrates the professional demographics of participants.

States represented

36 states were represented by participants. The most well represented state was Washington State with 27.87% of participants (n=34), followed by Ohio, with 9.02% of participants (n=11), and New York and California with 5.74% of participants (n=7) each.

Responses to survey questions on five topics of interest

1. OI Awareness

Primary question: How aware are attorneys and child welfare workers about OI?

Participants were asked to select how they first became aware of OI. 24% of participants (n=30) were unfamiliar with the condition prior to learning about this study. Overall, the majority of respondents that had heard of OI before had first heard about it on the job (49%, n=61). 13% (n=15) had learned about it from some form of media or a friend, and 11% (n=14) had learned about it at university.

The survey included a question asking participants to choose which of the listed statements best described the level of familiarity they had with OI and its relationship to NAI cases. Of all the participants, 27% (n=33) indicated that they have little to no understanding of OI. The majority of participants, 67% (n=82) stated that they know that OI can be confused for

child abuse, and 15% (n=18) indicated that they can determine when it is appropriate to request diagnostic testing for OI. Only 2% (n=3) indicated that they have no problem interpreting genetic test results for OI. When numeric values were assigned to each of the four answer choices with 1 being the least amount of familiarity (little to no understanding of OI) and 4 being the most familiar (no problem interpreting genetic test results for OI), the average value among all participants was 1.7.

Participants were asked to estimate the percentage of professionals in their field (law or social work) they expect would have heard of OI before. Using a combined sample (n=118), the average was 42.41% with a standard deviation of 28.39. Among attorneys (n= 57), the average percent was 42.89% with a standard deviation of 27.09. Among child welfare workers (n= 61), the average was 38.21% with a standard deviation of 28.46. Note these are large standard deviations that indicate large variations in opinion.

Ancillary question: Do attorneys and child welfare workers receive training on the job about OI?

The total percentage of participants that had attended a work related training, conference or seminar in which OI or other genetic conditions were discussed was 30% (n=37). Most (70%, n=85) had not attended a training of this type. There was no difference between the aggregate response to this question between participants that identified as attorneys and those that identified as child welfare workers.

2. Genetics/Science Familiarity

Primary Question: To what extent have attorneys and child welfare workers had formal education in genetics?

A series of survey questions were asked related to the participants' previous genetics education history. Most participants (n=113; 82%) had had very little formal education in genetics (covered as part of their undergraduate biology or less). The remaining 18% had taken at least one full course on genetics.

Ancillary question: How comfortable are attorneys and child welfare workers with reading scientific literature?

The survey asked participants to select which response best described how comfortable they felt reading scientific literature. This question was asked because previous research has indicated that genetic test results are often written in a scientific way that is too complicated for attorneys to understand. We also asked this question in response to interview data that indicated that some attorneys and child welfare workers engage in genetics self-education using scientific journals and related literature. This question was not intended to be a proxy for professionals' abilities to interpret a genetic test result.

Combined among participants, 1% (n=2) indicated that they have never attempted to read a scientific journal before, 18% (n=25) do not feel very comfortable with scientific literature, 58% (n=82) state that they are fairly comfortable reading a scientific journal, and 23% (n=32) indicate that they have no problem reading a scientific journal. When assigned numeric values where 1= no attempt to read a scientific journal and 4= no problem reading a scientific journals, the average value among all participants was 3.14.

Ancillary Question: Are attorneys and child welfare workers aware of a variant of unknown significance (VUS)?

83% (n=89) of participants indicated that they had never heard of a VUS before. 17% (n=18) indicated that they had. Responses to the question asking about participant's familiarity

with VUS results were varied. 77% (n=78) indicated that they had never heard of a VUS or had no idea what it was. 3% (n=3) indicated that a VUS should not be considered admissible evidence in court, 3% (n=3) thought a VUS should be treated as the equivalent of a positive test result, 10% (n=10) believe that a VUS needs to be followed with parental DNA testing, 2% (n=2) stated that a VUS should be treated as the equivalent of a negative test result.

3. Factors that contribute to whether a child will be tested for OI

Primary question: Why doesn't genetic testing for OI happen in a case when it could be helpful?

Participants were invited to rank the reasons that genetic testing for OI does not happen in a court case when, genetic testing for OI could or would have been helpful (either hypothetically guessing that it could have been helpful or retrospectively concluding that it would have been helpful). The reasons that were selected in order from most common response to least common response is as follows: lack of awareness about OI: 91% (n=99), insufficient funds to cover diagnostic testing: 60% (n=65), other: 29% (n=32), parent's resistance to going through with genetic testing: 28% (n=31), testing not relevant due to guilty plea: 24% (n=26), no access to child for diagnostic testing: 21% (n=23), lack of confidence in the validity of the genetic test: 18% (n=20), concern about emotionally harming the child by the testing process: 9% (n=10), concern of physically harming the child by the blood draw: 5% (n=5). Participants were given a free text box at the end of this question to leave any additional comments that they wished researchers to read. Responses that were recorded from those that indicated "other" included the following:

"lack of interest in discovered it [OI] and its potential impact on the prosecutor of child abuse",

"state and law enforcement have no interest in the truth",

"CPS believes the parents(s) are guilty of abusing their child and don't seek alternative explanations",

"Overdelegation of diagnostic responsibility to "child abuse" pediatricians who are underqualified to rule out this diagnosis",

"Prosecutor, CPS and pediatricians aversion to believing it is an issue and preference to jump to the conclusion that it is abuse",

"Cases are resolved by agreement prior to trial, so no testing",

"Doc for state are biased",

"Court is inclined to follow medical doctor's opinion when abuse is suggested upon initial investigation and look no further".

Figure 4 shows a diagram of participants' responses to this question.

Respondents were also asked to indicate some of the concerns that they have about using genetic testing for a case. The most frequently selected answer (40%; n=43) was potential misunderstanding of the genetic test results. However, 39% (n=42) indicated that they have no concerns related to genetic testing. Other concerns that were selected in order from most to least common included: 36% (n=39) selected fear of introducing ambiguity or confusion especially with inconclusive test results, 29% (n=31) selected cost of the test as a concern, 21% (n=23) indicated skepticism about the accuracy of the test results, and 19% (n=20) selected fear of increasing the likelihood of losing the case. Other answers that were not commonly selected (10% or fewer of the participants) included: potential genetic discrimination (10%), harm/trauma to the child (9%), fear that the test result will reveal something else that was not anticipated (e.g. health related condition) (8%), and potential breach of confidentiality (7%).

Ancillary question: At what stage of an investigation or trial should genetic testing be requested if OI is being considered?

Regarding the timing during a case that genetic testing for OI should be requested, the majority of respondents (89%, n=98) agreed that testing should be requested during an investigation, before charges are pressed. However, not all agreed with this. 7% (n=8) of participants indicated that they think testing should happen after charges are pressed, but before the trial, while 3% (n=3) indicated that it is case dependent and should be tested "whenever OI becomes relevant." No one believed that testing should happen during the trial.

4. Medical experts involved in OI vs. NAI cases

Primary question: How easy is it for attorneys and child welfare workers to get help from a medical expert?

Participants were asked whether they have easy access to a medical expert. 59% (n=64) of the respondents indicated yes, 28% (n=30) indicated no, 13% (n=14) said they weren't sure. The average value for this question was 0.68, where yes=1, no=0, and "not sure" answers were excluded. One person left a comment stating, "Funds are available to pay for experts, but the process of identifying the appropriate expert and lining up funding can be burdensome".

Ancillary question: How likely is it that attorneys or child welfare workers will be able to get help from a geneticist?

A question was asked to try to elucidate the probability that if a case requested and received a medical expert, the expert would be knowledgeable in genetics. 24% (n=25) stated that it would not be likely, and 19% (n=20) indicated that it would be likely. 49% (n=51) stated that they had the option to choose to specifically request a genetics expert if necessary. When

collapsed into binary categories where experts would either be likely to be a geneticist or a geneticist could be requested, the average value among participants was 0.72.

In exploring more about how medical experts are selected to testify for a case, participants were asked where the majority of the medical experts that are selected to work on a case usually come from. 35% (n=33) of participants indicated that local physicians were chosen, whether or not their area of expertise was genetics. 46% (n=43) stated that the medical experts come from a variety of different sources, and 47% (n=44) stated that anyone with expertise in the pertinent area can be chosen.

A follow up question was included to gauge participant's confidence in medical experts' abilities to understand a genetic test result. 29% (n=31) indicated that they are very confident in a medical expert's abilities, while 41% (n=44) are only somewhat confident in their abilities. 7% (n=7) indicated that they have little to no confidence in medical experts' abilities. 18% (n=19) were neutral on the issue.

Ancillary question: Are the medical experts unbiased in their opinion of differential diagnosis of OI vs. NAI?

Participants were asked about the experiences they have had working with medical experts. Only one attorney answered this question, and 40 child welfare workers answered the question. The one attorney indicated that the medical experts are usually unbiased and that they consider all possibilities of unexplained fractures equally. Among the 40 child welfare workers that answered this question, 80% (n=32) of them indicated that the medical experts are usually unbiased, and 20% (n=8) indicated that the scientific experts are usually biased towards injuries happening from abuse. No one indicated that the experts are usually biased towards injuries happening from accidents or medical causes.

5. Direction of Future suggested policy

Primary question: Are attorneys and child welfare workers interested in learning more about OI?

The majority of participants (90%, n=93) agreed that professionals that involved in cases where OI could occur would probably be interested in further education regarding OI. However, comments left by participants included the following:

"Although I think some might be resistant to changing their views"

"Lawyers and CPS workers, yes; "child abuse" pediatricians are zealots and not neutral or impartial medical experts"

"They should be very interested and willing to keep an open mind but I'm not sure that is actually the case."

Participants were also asked to indicate which methods of education they thought would be most beneficial for increasing awareness of OI among attorneys and child welfare workers. In order, the following methods from predicted most effective to predicted least effective were: work-related conference/seminar/training, webinar, website, email, change in academic curriculum, and social media.

Ancillary question: Are attorneys and child welfare workers interested in suggestions for policy or practice guidelines surrounding OI vs. NAI cases?

A question was asked to gauge whether participants think it could be beneficial to have suggestions for policy changes regarding genetic testing of OI in NAI cases and what type of policy changes might be beneficial. The majority of participants (75%, n=76) selected yes. 25% (n=26) selected no. Many of the participants in favor of a policy change gave some suggestions regarding conditions under which a policy might be recommended, including:

"Yes for all bone breaks, especially with no other injuries"

"In cases with multiple fractures, it should be mandated to cover all bases"

"When there is no clear perpetrator, no other injuries, and no history of injuries."

Participants that indicated that a policy change would not be helpful stated comments including the following:

"It would likely be another unfunded mandate"

"The child safety workers in my state don't follow their own policies"

"Policy changes rarely have real impact due to lack of training on said policy".

In regards to whether participants think it would be beneficial to have practice guidelines pertaining to genetic testing of OI in NAI cases, the majority of participants (91%, n=91) selected yes. 9% (n=9) selected no. Many of the participants in favor of practice guidelines gave suggestions regarding the guidelines including:

"checklist to go through, or a decision tree based on information gathered during investigation"

"one page sheet"

"Increased education and trainings on medical conditions that can be confused for child abuse for mandated reporters".

Participants that were not in favor of practice guidelines provided comments including:

"As a CPS worker, I rely on my medical staff to know this stuff"

"I feel that medical professionals should decipher the results/make suggestions for the child's wellbeing".

Discussion:

My hypotheses that the level of OI awareness among child welfare workers and attorneys is at a moderate level, their understanding of genetics is low, and they have interest in learning more about OI, were all supported.

OI awareness

Overall, the participants that responded to this survey had a moderate awareness of OI (75% of participants had heard of it, and 67% of those knew that fractures due OI could be confused for NAI). However, despite these professionals having this level of awareness of OI, they indicated that other professionals in their same subspecialty likely had a lower awareness of OI and indicated that other professionals in their field would probably be interested in learning more about OI. This combination of responses indicates that the group of participants that answered this survey may not be representative of the greater population of child welfare workers or attorneys involved in NAI cases, which limits the generalizability of the findings. This issue is further elaborated on in the limitations section of this chapter.

Genetics education and comfort reading scientific literature

Despite the fact that participants on average had low levels of genetics education in their academic background, they rated their level of comfort with reading scientific literature as quite high. Given that participants interviewed as part of Aim 1 of this research indicated that although they do not have formal education in genetics, they felt as though they had done self-education in

genetics by reading scientific literature, these responses raise the question of how well they actually understand the papers or journals that they are using for self-education, and how much they retain.

Genetics knowledge and medical expert availability

A question related to genetics knowledge regarding their awareness of VUS in genetic testing was included in the survey, which revealed that 78% of participants had never heard of VUS before and would need help understanding what it means. Unfortunately, we also know from the results of this survey that only 59% (n=64) of the respondents indicated that they have easy access to a medical or scientific expert, and 25 of those participants indicated that even if they were assigned a medical expert, it was not likely that he or she would be knowledgeable in genetics. These numbers combined raise questions about how the participants from this study are expected to get assistance from the appropriate individual to assist them in understanding the genetic test result. Moreover, 5% (n=5) of the professionals that answered the question relating to VUS indicated that they believe that a VUS should be treated as the equivalent of either a positive or negative test result. If these professionals assume that they understand what a test result means, they may forgo requesting a scientific expert and proceed forward in the case with an incorrect understanding of the child's genetic test result. This misunderstanding could lead to deleterious consequences for the family represented by the professional.

Timing during an investigation that genetic testing for OI should be requested, if it is being considered

The majority of participants (89%, n=98) agreed that if genetic testing is going to be requested, the request should happen during an investigation, before charges are pressed.

However, not all participants agreed on this. According to information acquired from an interview conducted during Aim 1, waiting until after charges are pressed makes getting diagnostic testing much more complicated due to a variety of different reasons. For example, it can be difficult to get access to a child who is no longer local due to removal from the home, and it can be very hard to get anything to happen to someone who is not the defendant. Suggestions for changes in practice could help encourage professionals who are considering OI as a possible diagnosis to request the test at the most appropriate time during the investigation. The timing could vary for different cases but should take a number of factors into consideration such as where the child might go if removed from the home, and whether it will be possible to test the child after removal from the home or after a trial is underway.

Medical expert bias

Responses indicated that some participants believe that on occasion, medical experts (particularly child abuse pediatricians) are biased to believe that all bone fractures are a result of child physical abuse, and they do not always consider alternative explanations. It is unclear from participants' comments whether physician bias (perceived or real) could be due to lack of awareness of alternative explanations of child physical abuse, or whether they are aware of the existing conditions but firmly believe, as participants have indicated, all children's unexplained fractures are due to physical abuse. Either way, ethical questions are raised about a child abuse pediatrician's attitude towards these cases if not all possibilities are being appropriately considered.

Limitations:

There are many decisions that were made through the course of this research that limit the generalizability of the findings. For example, this research was likely affected by two

different types of selection bias. The first type of selection bias is non-response bias. Given that we have little information on the participants that did not respond to the survey, there is no robust way of ensuring that those that did not respond to the survey are not statistically significantly different from those that did respond to the survey. However, demographic data was obtained for the listservs that were used to recruit child welfare workers in order to attempt to make estimates about the non-responders. Given that only 3% (2050 out of 56,175) of the entire NASW listserv comprises professionals involved in child welfare, the response rate among child welfare workers that was obtained in this study (1.6%) using 3 different litservs that were all subsections of the total NASW listserv, the response rate seems to be an accurate reflection of the proportion of professionals that we wanted to participate. Of course, we have no accurate way of ensuring that the child welfare non-responders are different from the responding child welfare workers, so we cannot generalize these findings beyond the specific study population that chose to participate in the study.

Secondly, since potential participants made the choice themselves whether to participate in the study, voluntary sampling bias is also a likely factor in this research. For this reason, it is possible that the participants are an over-representation of those that have been involved in OI vs. NAI cases, and could over-represent those that had had particularly memorable experiences.

Thirdly, although the sample size was larger than the sample size calculation had indicated would be a sufficient size to yield 80% power for each of the group comparisons, a larger sample size would have resulted in more robust findings that are more generalizable, and would have allowed us to test more complex theories and interactions between variables. For this reason, a larger sample size with greater participant representation would have been useful.

Finally, there were some questions that would have been prudent to have asked in this survey that were not included. For example, this study did not ask about the number of years that each participant had been working in their current profession, which would have provided a better indication of about how long it might take a professional on average to learn about OI on the job. I also did not ask about when each participant had first heard of OI, which would have been a good indicator of whether awareness of OI has increased over time or not. Further research should be done to help answer these research questions.

Conclusions:

Child abuse cases are complex and involve numerous stakeholders with different areas of expertise. Findings indicate that professionals involved in NAI cases could benefit from an increase in education that raises awareness of OI as a potential alternative explanation to childhood fractures. Findings from this research also indicate that the medical professionals that are assigned to a case may not be the most appropriate person to help an attorney or child welfare worker understand a genetic test result, and some medical professionals introduce bias into an investigation. Many limitations of this study limit the generalizability of the findings, but based off of this specific study population, potential suggestions for educational or policy changes could be beneficial. The following chapter lays out a few potential suggestions for policy and educational changes, which were informed by the results of this limited study.

CHAPTER 5: POLICY RECOMMENDATIONS

ABSTRACT: Children with injuries suggestive of non-accidental injury (NAI) represent a health care problem that affects thousands of children each year. While NAI is common, it is not the only cause of unexplained fractures. Osteogenesis imperfecta (OI) is the most common cause of predisposition to bone fracture, and can result in injuries that are difficult to distinguish from NAI. Although genetic testing for OI can be done, previous research has indicated that the test is not always requested when OI cannot be ruled out, and the genetic test results are not always properly understood by professionals who may have to present the test results during a case. This article provides a list of suggested points to consider and potential suggestions to changes in policy, education, and practice that could be useful to professionals involved in OI vs. NAI cases.

Background:

Non-accidental injury (NAI), or child physical abuse, is a public health problem that affects thousands of people every year. There are over 119,000 cases of confirmed physical child abuse every year in the U.S. and it is likely that many more cases go unreported.[1] In 2004, in children less than three years old, 24%-40% of all fractures were suspected to be related to non-accidental injury.[24, 25, 26]Among children hospitalized each year for fractures (about 150,000), the percentage of those in which NAI could be a cause for fractures (and without traumatic brain injury) is highest among children less than one year of age (20.4%).[4]

While NAI is unfortunately ubiquitous worldwide, it is not the only cause of injuries suggestive of abuse. The inherited condition osteogenesis imperfecta (OI) is characterized by bone fragility and skeletal fractures. However, it may not be apparent that the child has a genetic condition if the case is mild and the child has no other accompanying features of OI, such as blue sclerae, bowing of the long bones, family history, or joint hypermobility. OI is a rare condition, with an incidence of about 1 in 10,000 live births, which corresponds to the birth of about 400 children each year with OI in the US.[48] It has been estimated that OI accounts for about 0.85% of hospitalized children with fractures and no head trauma who are evaluated for NAI. Many patients are referred for genetic testing for OI as a means to help rule out a genetic cause of unexplained fractures, but of course, OI and NAI are not mutually exclusive so a positive OI test result does not rule out NAI.[1]

Other genetic conditions exist that could also explain the fractures, such as genetic forms of rickets and hypophosphatasia. If the appropriate cause of injuries is not known and the process for determining the cause of the injuries is mishandled, it could lead to an emotionally traumatizing separation of children from their parents. Therefore, it is prudent to rule out OI or

other medical conditions in a time efficient manner as part of an evaluation of suspected child abuse.

Genetic testing for OI can be done to test for an alternative explanation for skeletal fractures; however, research has indicated that genetic test results are often written in a way that is difficult for non-scientists to understand.[57] Despite the fact that genetic test results can be difficult to understand, we know from previous research that in OI vs. NAI cases, genetic results are sometimes returned to providers (e.g., child welfare workers or attorneys) who have no formal training in genetics.[34] In addition, genetic testing could uncover variants of unknown significance (VUS) which introduce uncertainty into the diagnostic process. If an attorney or child welfare worker does not understand what a VUS result means, he or she may proceed as though the result is equivalent to a positive test result (confirmatory of OI) or a negative test result (free from OI) although neither of these conclusions are appropriate.[10]

Consequences of misusing genetic test results in court

In the US, after an allegation of child physical abuse is made, the professionals involved in the case often remove the child from his or her home before the investigation into the physical abuse allegation is complete in order to protect child safety. This lowers the risk that the child will be physically harmed further by any potential perpetrator at the home. Once removed, the child usually goes into temporary foster care or to stay with a relative while the investigation proceeds. Inappropriate separation can result in unnecessary trauma, both for the parents and the child, who has to endure the loss of familiar people, familiar settings, and their accustomed schedule. For their parents, they must endure the loss of their child, potentially the loss of other children, and both the embarrassment and cost of the abuse trial that ensues.

Importance of recommendations

Educational and policy changes could lead to improved utilization or timeliness of genetic testing for OI in the context of concern for NAI, and to ensure that professionals involved in suspected NAI cases are aware of the possibility of OI as a diagnosis.

Background research

In chapters 3 and 4 of this dissertation, preliminary data was collected that informed the policy suggestions outlined here. Qualitative research involving expert interviews and quantitative research involving an online survey was done to collect data regarding what the professionals already knew about OI vs. NAI cases and how they approach information from genetic test results during a case. The interview data is explained in chapter 3, and the survey data is explained in chapter 4. The information from these two aims was compiled and analyzed to identify the points to consider and develop suggestions.

Findings from previous research

1) Both the data collected from the expert interviews as well as the data collected from the survey revealed that attorneys and child welfare workers involved in child physical abuse cases do not have much, if any, training in genetics. Although subjects that participated in the research aims had moderate levels of awareness of OI, they indicated that their estimate of awareness of OI and its relationship to NAI among other professionals in their field is quite low. However, 90% of the participants that took the survey indicated that they believe professionals in their field would be interested in further education related to OI and genetic testing.

2) The study indicated that attorneys and child welfare workers, when given genetic test results, may not be able to understand them, and may not have easy access to a professional that is trained in genetics that can help them understand what conclusions can be drawn from the test results. According to participants, medical experts that are brought onto a case are often local physicians who may or may not be trained as a geneticist.

3) Data revealed that even if professionals involved in a case are inclined to have a child genetically tested for OI, there are numerous reasons why testing does not always get done, including lack of access to the child once a trial is underway, refusal to test by the parents, or fear that testing could lead to a more severe outcome for the parents.

Suggestions

While the following suggestions refer to OI in particular as it is the most common condition confused for NAI, these statements also apply to other conditions that result in a similar phenotype.

- Educational suggestions to consider
    - Education resulting in awareness about OI and its relationship to NAI should be offered as part of on-the-job training for any professional working with NAI cases
    - Education about OI could be offered as part of continuing education that attorneys involved in NAI cases are required to pursue
    - The role that OI plays in NAI cases could be covered in courses in social work and social welfare programs that relate to child physical abuse

- Policy suggestions to consider
    - Scientific or medical experts that are brought into a case to testify about a genetic test result should be formally trained as geneticists. The required background and training for these experts could be defined and standardized.
    - A physician should examine all children suspected of NAI, and if the child presents with fractures, the pediatrician should consider OI (e.g., ask about family history of fractures, look for other signs of OI, and potentially recommend genetic testing if OI cannot be excluded)
- Practice guidelines to consider
    - Child welfare workers and attorneys should have a list of medical professionals that are trained in genetics and aware of OI that they can refer to in the event that a child presents with unexplained fractures and it is unclear whether OI can be excluded
        - Although the most reliable way to rule out OI is with diagnostic testing, a diagnosis of OI becomes much less likely if the child presents with any other injuries that could not be due to OI, such as bite marks or burns. In the absence of any injuries other than skeletal fractures, a child welfare worker or attorney should consider having the child examined by a child abuse pediatrician who should do a physical examination and ask about social and medical history to determine whether diagnostic testing for OI might be appropriate.

    - Child abuse pediatricians or medical geneticists examining a child should have a checklist of factors to consider before ruling out OI as a possible cause.
        - The checklist should include physical features that sometimes accompany OI, social history, medical history, and potentially diagnostic testing

- Potential methods of information dissemination

The following four methods of information dissemination were the top four methods, in order, chosen by participants of the survey

    - Workplace seminar or training
    - Incorporated as part of a conference or workshop
    - Webinar
    - Emailed information

- A workplace seminar or training would consist of an in-person session that would be made available for professionals at an institution in order to engage in education surrounding OI or genetic testing.
- A session at a conference or workshop would be similar to a workplace seminar or training, however, it would be incorporated into a larger conference. These conferences could be nationwide or otherwise more largely accessible to a greater number of professionals.
- Webinars would be held entirely online, and could be archived for later viewing. Ideally, questions could be submitted electronically in real-time as the webinar was being recorded.

- Emailed information would consist of a few paragraphs of general information on OI vs NAI, as well as a list of resources for recipients to access for further information.

As a whole, participants that responded to the survey indicated that the following groups of professionals, in order, are the most crucial to receive further education on OI and genetic testing:

  - Child Protective Service (CPS) Workers
  - Child abuse pediatricians
  - Attorneys

Chapter 6: CONCLUSIONS

NAI represents a public health problem that affects thousands of children annually. Of those children that have been suspected of NAI, a subset of them have skeletal fractures. Of those children suspected of NAI and with skeletal fractures, a medical condition such as OI could be responsible for the fractures. At the moment, the number of children who are determined to have been abused when, in fact OI is the explanation for fractures, is unclear. The number is difficult to calculate due to the lack of an established and comprehensive child abuse registry, and the reliance on physicians to request genetic testing for OI when they deem it necessary.

Previous research has indicated that genetic test results are often written in a way that is difficult for non-scientists to understand.[57] In the context of an NAI case, if a genetic test result is requested, a non-scientist such as an attorney or a child welfare worker could be responsible for understanding and representing the test result in a court case. This study was designed to investigate how aware attorneys and child welfare workers are of OI, and whether they have any problems understanding and/or representing genetic test results.

The study was designed in three parts. The first aim of the study was a qualitative component involving expert interviews with child welfare workers and attorneys who are currently involved in child abuse cases. The goal of this aim was to gather a baseline of information about this topic from the two target professional groups that could be used to design a quantitative survey to survey a greater number of participants. The second aim of the study was the quantitative interview, which asked participants questions similar to those in the interview, but was designed in a way that could accommodate quantifiable responses, and would sample more people than the number of people that were interviewed. The third aim of the study was designed to be a reaction to the data that came out of the first two aims; a document containing

suggested policy or education changes that could be implemented to help address any of the issues that may have arisen in the first two aims.

Aim 1 consisted of expert interviews with 8 attorneys involved in child abuse cases and 6 child welfare workers. Each interview was recorded, transcribed, coded, and analyzed. Thematic content analysis indicated that the professionals involved in child abuse cases are not typically trained to consider potential causes of skeletal fracture other than NAI. This raises the question of whether genetic test results could occasionally be misunderstood. Furthermore, the medical experts that are brought into the case to help interpret the genetic test results are not always helpful in offering an accurate interpretation. The results of this study indicated that we need to confirm these findings with a quantitative evaluation among a larger population of the same professionals.

Aim 2 carried out the quantitative component of the study. Both child welfare workers and attorneys involved in child abuse cases were invited to participate. The data was organized using descriptive statistics. The results of this study supported the hypotheses that child welfare workers and attorneys involved in child abuse cases are not typically receiving training or background in genetics, but they do have moderate awareness of OI. . We also found that although nearly all participants who took the survey have rather low education in genetics, they rated their confidence in themselves to understand scientific literature rather highly, and they rated their colleague's abilities to interpret a genetic test result as much higher than their own. Overall, participants indicated that they were interested in learning more about OI and genetic testing and agreed that a policy or practice change could be beneficial to their field.

Aim 3 took synthesized the results of aims one and two and integrated the information into a list of suggestions for education, policy, and practice. The recommendations are for

professionals or professional groups to consider as potential changes to implement for cases in which NAI and OI are potential differential diagnoses. The recommendations are not sanctioned by any organization or government body, but rather have been suggested by the participants that were recruited from both of the first two aims of this study.

This dissertation research was designed as an inquiry into how attorneys and child welfare workers understand and use genetic test results for OI in NAI cases. The project was a small, exploratory study whose results are unable to be generalized to a larger population. Many limitations were identified and discussed. Future research on this topic is needed.

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Figure 1. Example scenario of the timing in which stakeholders get involved in OI vs. NAI cases

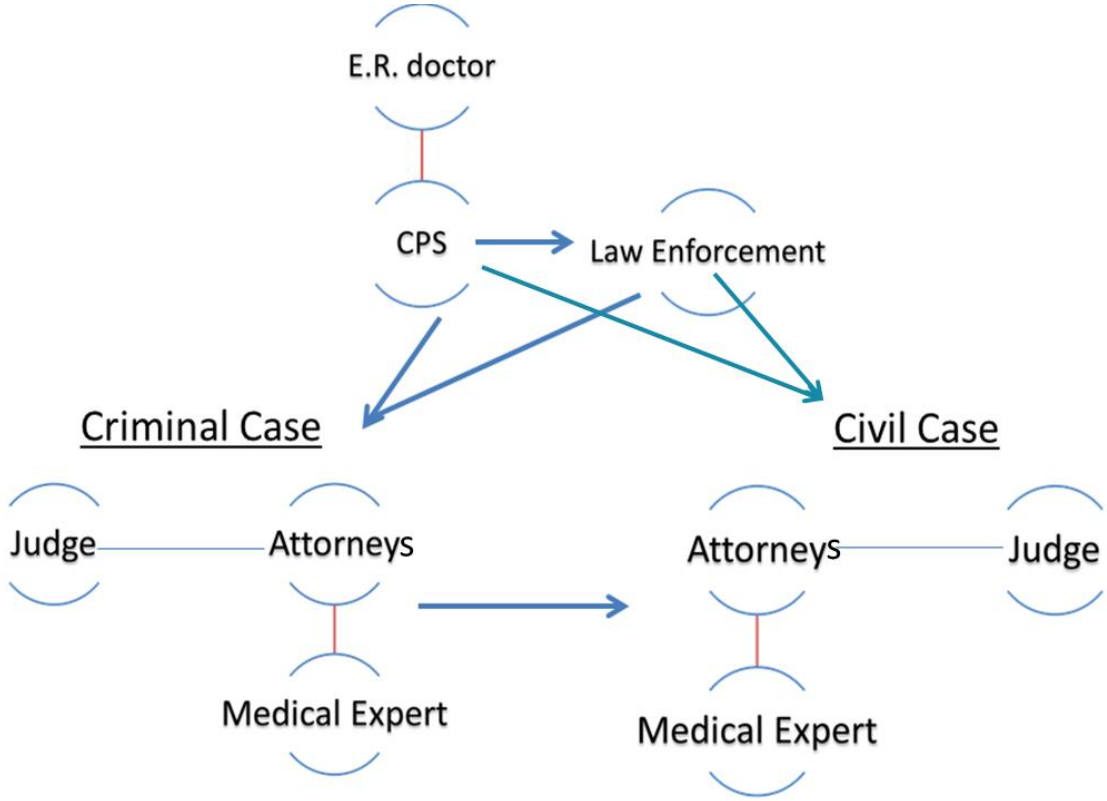

Figure 2. Example of a test result for COL1A1 and COL1A2 (the two main genes responsible for OI)

Sample Type: BLOOD

Referral Diagnosis: OSTEOGENESIS IMPERFECTA (OI)

Indications for Testing: MULTIPLE FRACTURES, FAILURE TO THRIVE

Molecular Test Results: *COL1A1* VARIANT OF UNKNOWN SIGNIFICANCE IDENTIFIED

Gene	Molecular Test Results	Number of Heterozygous Amplicons
COL1A1	**Variant of unknown clinical significance**	11
COL1A2	No abnormality	9

Heterozygous Variant of Unknown Significance Identified:

Gene	Exon	DNA Change	Protein	Type	Effect
COL1A1 GenBank: NM_000088.3 Chromosome 17	51	c.4228G>A	p.Val1410Ile	Substitution	Missense

Interpretation: We did **not** identify a disease-causing mutation in the *COL1A1* and *COL1A2* genes. We identified a variant of unknown significance (as described in the table) in one allele of *COL1A1*, the gene that encodes the proα1(I) chains of type I procollagen. We have not seen this variant before and it is not listed in the Collagen Mutations Database (http://www.le.ac.uk/ge/collagen/). It was also not identified among the about 13,000 alleles compiled in the Exome Variant Server (http://evs.gs.washington.edu/EVS/), and it is not reported in ClinVar (http://www.ncbi.nlm.nih.gov/clinvar/) or dbSNP.

The single base pair alteration results in substitution of valine (Val) by isoleucine (Ile) at protein position 1410, which is in the carboxyl-terminal propeptide of the proα1(I) chain. We think it is not likely to be of functional significance for several reasons: 1) valine at position 1410 is only moderately conserved (86 of 97 vertebrate species); 2) the substitution is a conservative change in that both valine and isoleucine are hydrophobic, branched-chain amino acids; 3) the site of the substitution does not affect a known functional domain; 4) based on the recently solved three-dimensional structure of the C-propeptide trimer of type III procollagen (Bourhis et al. 2012), valine at 1410 does not participate in interchain

interactions. Due to rarity, though, we cannot exclude the possibility that it could play a role in your patient's clinical picture.

A different amino acid substitution at this same site (p.Val1410Asp) was reported in one individual with a diagnosis of OI type III (Stephen 2014). However, the clinical information in that report is limited, no family studies were performed and other causes of severe OI were not investigated. In addition, it is important to note that the biochemical character of valine is similar to that of isoleucine but markedly different from aspartic acid.

We recommend targeted testing of parental DNA to determine if the variant occurred *de novo* or was inherited, and if inherited, whether it segregates with a bone phenotype. Please submit DNA or blood drawn in EDTA tubes (purple top), for each parent. **Parental studies will be provided free of charge only if clinical information is included on each parent for genotype-phenotype correlation.**

Reference:

Bourhis J-M et al. Structural basis of fibrillar collagen trimerization and related genetic disorders. Nat Struct Mol Biol 19:1031-1036, 2012

Stephen J, et al. Mutation Spectrum of *COL1A1* and *COL1A2* Genes in Indian Patients With Osteogenesis Imperfecta. Am J Med Genet Part A 164A:1482-1489, 2014.

Additional Information: The variant was confirmed by sequencing in both directions using a new amplification product. The identification of heterozygosity for polymorphic variants in one or more amplicons in each gene excludes a whole gene deletion.

Contact: Any questions or concerns about interpretation of the test results or additional testing can be directed to the laboratory genetic counselors (Dru Leistritz, MS, CGC or Sam Bailey, MS, CGC) at 206-543-5464.

Procedure: DNA was received or extracted from a submitted specimen. For the gene(s) below, the exon(s) and flanking intron sequences were amplified by PCR using the number of primer pairs noted in the table. The primers were designed based on publicly available gene reference sequences. The amplified fragments were sequenced by automated sequencing using the number of reactions shown in the table. Sequence data (ABI files) were analyzed by the most recent version of the Mutation Surveyor software (SoftGenetics). Identified gene sequence variants were annotated with A of the initiator methionine codon = 1 for the c. number, and initiator methionine = 1 for the p. number.

Gene	PCR Amplifications	Sequencing Reactions
COL1A1	17	26
COL1A2	23	35

Figure 3. OI diagnostic flow, as suggested by the European Molecular Genetics Quality Network.

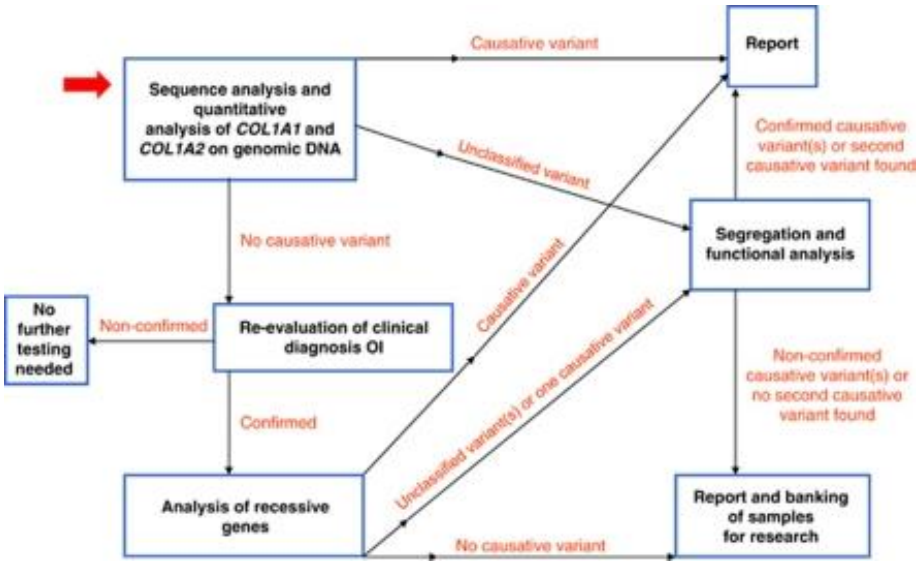

Figure 4. Breakdown of participants' responses to reasons that genetic testing for OI may not happen even if it could be helpful to a case. Percentages reflect the number of votes that each answer choice received out of the total number of votes cast. Percentages do not reflect the percentage of participants that selected each answer choice.

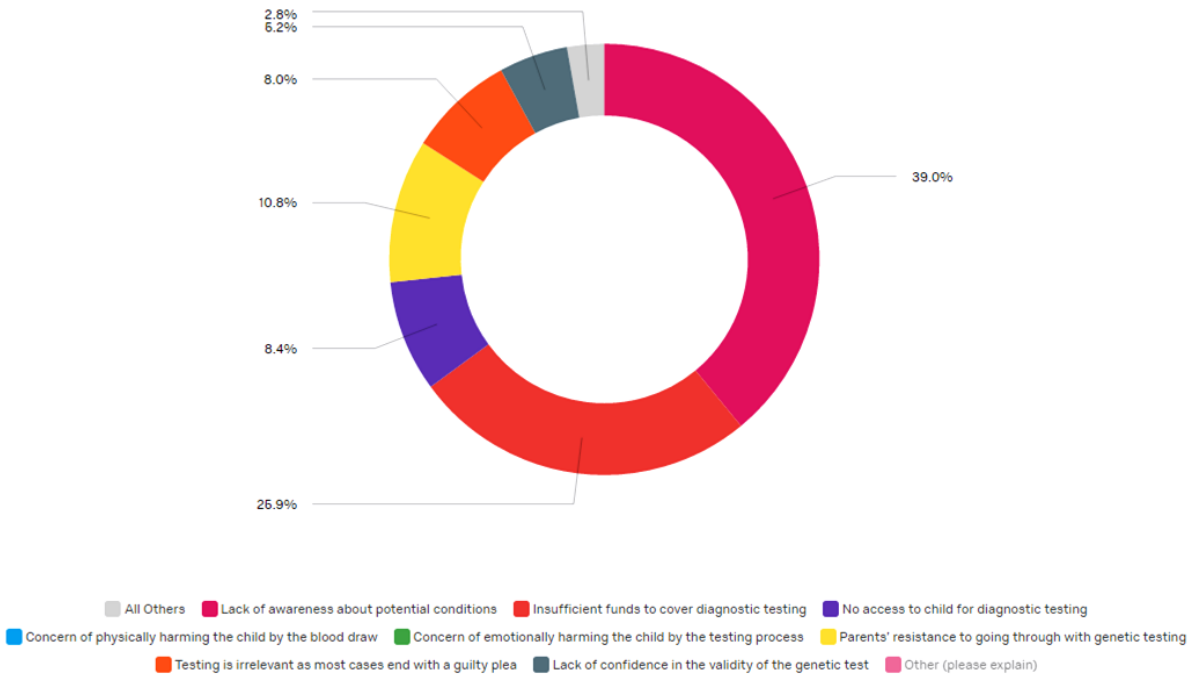

Table 1. Interview Participant Demographics

Attorneys

	Female	**Male**	**Total**
State/County	4	1	5
Private	1	2	3
Total	5	3	8

Child Welfare Workers

	Female	**Male**	**Total**
State/County	2	1	3
Hospital	0	1	1
University	1	1	2
Total	3	3	6

Table 2a: Sample size table for various estimates of the test value and the sample average, with increments set at 0.25, spanning sample values from 0-0.225

	0	0.025	0.05	0.075	0.1	0.125	0.15	0.175	0.2	0.225
0		1032	258	115	65	29	29	21	16	13
0.025			1032	258	115	65	29	29	21	16
0.05				1032	258	115	65	29	29	21
0.075					1032	258	115	65	29	29
0.1	65	115	258	1032		1032	258	115	65	29
0.125							1032	258	115	65
0.15								1032	258	115
0.175									1032	258
0.2										1032
0.225										

Table 2b: Sample size table for various estimates of the test value and the sample average, with increments set at 0.25, spanning sample values from 0.7-0.9

	0.7	0.725	0.75	0.775	0.8	0.825	0.85	0.875	0.9
0.7		1032	258	115	65	41	29	21	16
0.725			1032	258	115	65	41	29	21
0.75				1032	258	115	65	41	29
0.775					1032	258	115	65	41
0.8	65	115	258	1032		1032	258	115	65
0.825							1032	258	115
0.85								1032	258
0.9									1032

Table 3: Professional Demographics of Participants

Answer		Response	%
For child protective services		28	20%
For another child welfare services program		12	9%
In an office charged with the legal representation of parents		7	5%
In an office under contract with the state to provide services to children and families		13	9%
Volunteer CASA		2	1%
Attorney GAL		1	1%
Representing parents in dependency proceedings		46	33%
Representing parents in criminal proceedings		14	10%
Representing children		3	2%
Representing the state in dependency proceedings		1	1%
Representing the state in criminal proceedings		1	1%
Superior court Judge		0	0%
Other (please specify)		12	9%
Total		140	100%

Appendix 1. Qualitative Research Terminology Glossary

Code (n): tag or label for assigning units of meaning to the descriptive or inferential information compiled during a study. Code development is the initial step in analyzing interview data. Codes can come from existing theory (theory-driven), raw data (data-driven), or from specific research goals (structural)[1-2]

Coding (v): Assigning codes to sections of data that are connected to a specific context[1]

Codebook: A document that contains a set of codes, explains the coding process, acts as a guide for locating variables in the data set, and describes the meanings for each code used[3]

Content Analysis: any analysis of a body of communicated by objectively and systematically identifying key symbols and themes in order to ascertain the meaning[4]

Intercoder reliability: consistency in coding between multiple coders[3]

Subtle realism: A paradigm of inquiry that states that all research involves subjective perceptions and observations and concedes that different methods will produce different pictures of the participant(s) being studied[5]

[1] Miles M, Huberman A. 1994. Qualitative data analysis: An expanded sourcebook. 2nd ed. Thousand Oaks, CA: Sage.
[2] Ryan GW, Bernard HR. 2003. Techniques to identify themes. Field Methods 15:85-109.
[3] DeCuir-Gunby JT, Marshall PL, McCulloch AW. 2014. Developing and Using a Codebook for the Analysis of Interview Data: An Example from a Professional Development Research Project. Field Methods 23(2):136-155. Thousand Oaks, CA: Sage.
[4] Holsti OR. 1969. Content Analysis for the Social Sciences and Humanities. Reading, MA: Addison-Wesley.
[5] Hammersley M. 1992. What's wrong with ethnography? New York: Routledge.

Thematic analysis: An analysis technique emphasizing examining and recording patterns (or "themes") within data that are important to the description of a phenomenon and are associated to a specific research question[6-7]

Likert scale: A rating scale commonly used in questionnaires. The scale assumes that the strength/intensity of experience is linear on a continuum from strongly agree to strongly disagree. Usually consists of a five or seven point scale that allows subjects to rate how much they agree or disagree with a particular statement[8]

Purposive Sampling: A type of non-random sampling technique in which a researcher chooses specific people within the population to use for a particular study[9]

Snowball Sampling: A non-random sampling technique where existing study subjects suggest additional people to recruit into the study. This technique is often employed by researchers in situations where potential subjects are difficult to identify[10]

Mixed methods research: Collecting and analyzing both quantitative and qualitative data in the context of a single study[11]

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[6] Daly, Kellehear, Gliksman. 1997. The public health researcher: A methodological approach. Melbourne, Australia: Oxford University Press. pp. 611–618.
[7] Braun V, Clarke V. 2006. Using thematic analysis in psychology. Qualitative Research in Psychology 3 (2): 83.
[8] Likert, R. 1932. A Technique for the Measurement of Attitudes. Archives of Psychology, 140, 1–55.
[9] Tongco M, Dolores C. 2007. Purposive Sampling as a Tool for Informant Selection. Ethnobotany Research and Applications. 5: 147-158.
[10] Cohen N and Arieli T. 2011. Field research in conflict environments: Methodological challenges and snowball sampling. Journal of Peace Research**.** 48;4:423-435**.**
[11] Driscoll D, Appiah-Yeboah A, Salib P, Rupert D. 2007. Merging Qualitative and Quantitative Data in Mixed Methods Research: How To and Why Not. Ecological and Environmental Anthropology 3:1.

Appendix 2. Email that went out to potential participant listserv coordinators

To whom it may concern,

Hello, my name is Emily Youngblom, I'm a doctoral student at the University of Washington in the Institute of Public Health Genetics. My research focuses on a genetic condition called osteogenesis imperfecta (OI, or brittle bone disease) which can cause characteristics similar to those of child abuse, so it is sometimes difficult to distinguish the two. I would like to better understand what some of the problems are that result in signs of these medical conditions, such as broken bones, to be occasionally misattributed to physical abuse. I have written a survey that I am hoping that you might be able to distribute to your list of_____ so that I might better understand the situation. I will compile all the information I collect, which can then be used to inform policy or educational changes that could be made to help improve the system.

In order for me to understand where the specific gaps in knowledge are, I have designed a survey that I'm hoping that your office can help me distribute to your_____.

The survey is completely optional, completely anonymous, and participants can choose to skip any questions or exit at any time. The information provided will be highly valuable to me.

The link to the survey is here:

http://washington.co1.qualtrics.com/SE/?SID=SV_ePUT0wK9K8VXg1v

If you have any questions for me about this research, please feel free to contact me at eyoungb@uw.edu

Thanks so much for your consideration.

Emily Youngblom MPH

Institute of Public Health Genetics

University of Washington

Campus Box 357232

Seattle, WA 98105

206-616-0915

Appendix 3. Survey Questions

1 Thank you for your willingness to participate in this study. The survey is optional, anonymous, and you may choose to skip any questions or exit at any time. To thank you for your help, you have the option of entering in a drawing for one of five $20 Amazon gift cards. Although the email address that you provide for the optional drawing entry will be visible to researchers, it will not be associated with any of your answers, and does not have to be an email address associated with your work.

2 What state do you practice in?

- ❍ Alabama
- ❍ Alaska
- ❍ Arizona
- ❍ Arkansas
- ❍ California
- ❍ Colorado
- ❍ Connecticut
- ❍ Delaware
- ❍ Florida
- ❍ Georgia
- ❍ Hawaii
- ❍ Idaho
- ❍ Illinois
- ❍ Indiana
- ❍ Iowa
- ❍ Kansas
- ❍ Kentucky
- ❍ Louisiana
- ❍ Maine
- ❍ Maryland
- ❍ Massachusetts
- ❍ Michigan
- ❍ Minnesota
- ❍ Mississippi
- ❍ Missouri
- ❍ Montana
- ❍ Nebraska
- ❍ Nevada
- ❍ New Hampshire
- ❍ New Jersey
- ❍ New Mexico
- ❍ New York
- ❍ North Carolina
- ❍ North Dakota
- ❍ Ohio
- ❍ Oklahoma
- ❍ Oregon
- ❍ Pennsylvania

- ❍ Rhode Island
- ❍ South Carolina
- ❍ South Dakota
- ❍ Tennessee
- ❍ Texas
- ❍ Utah
- ❍ Vermont
- ❍ Virginia
- ❍ Washington
- ❍ West Virginia
- ❍ Wisconsin
- ❍ Wyoming
- ❍ Outside the U.S.

3 Please select the profession you most identify with

- ❍ For child protective services
- ❍ For another child welfare services program
- ❍ In an office charged with the legal representation of parents
- ❍ In an office charged with the legal representation of children
- ❍ In a guardian ad litem office
- ❍ In an office under contract with the state to provide services to children and families
- ❍ Volunteer CASA
- ❍ Attorney GAL
- ❍ Paid non-attorney GAL
- ❍ Representing parents in dependency proceedings
- ❍ Representing parents in criminal proceedings
- ❍ Representing children
- ❍ Representing the state in dependency proceedings
- ❍ Representing the state in criminal proceedings
- ❍ Commissioner
- ❍ Superior court Judge
- ❍ Other (please specify) _____

4 How many of each type of staff currently work at your local firm/department/institution?

_____ Attorneys
_____ CPS workers

5 To what extent have you ever had training in genetics?

- ❍ I've never had any training in genetics
- ❍ It was covered in a biology class I took as undergraduate in college
- ❍ I took a genetics class in college
- ❍ I've taken two or more genetics classes in college or later
- ❍ Other (please specify) _____

6 To what extent do you feel comfortable reading medical or scientific journal articles?

- ❍ I've never read a medical or scientific journal before
- ❍ I don't feel very comfortable reading medical or scientific journals. I would need help understanding most of it
- ❍ I feel fairly comfortable reading medical or scientific journals, although I would need help understanding some of it
- ❍ I have no problem reading medical or scientific journal articles

7 How did you first hear about osteogenesis imperfecta (OI)?

- ❍ The email that contained the link to this survey
- ❍ On the job
- ❍ Media (book, TV, radio, etc)
- ❍ From a friend
- ❍ At school/university
- ❍ At a work-related training/webinar
- ❍ Other (please specify) _____

8 Have you ever attended a work-related training in which OI was mentioned? If so, what was the training?

- ❍ No
- ❍ Yes _____

9 Please select which of the following statements apply to you (choose all that apply)

- ❑ I have little to no understanding of OI
- ❑ I am aware of OI and know that it can be confused for child abuse
- ❑ I can determine when it is appropriate to request diagnostic testing for OI
- ❑ I have no problem interpreting genetic test results for OI

10 Please select which ONE of the following statements you most agree with

- ❍ OI never plays a role in child abuse cases
- ❍ Fractures due to OI can be confused for fractures due to child abuse

11 Please indicate how strongly you agree or disagree with the following statement:Cases where alleged physical child abuse could be explained by OI are problematic and need to be addressed better. 1 = completely agree5 = completely disagree

_____ .

12 In your opinion, what percentage of professionals in your field do you expect to have heard of OI before?

_____ .

13 Who should know about OI? (Please select all that apply)

- ❑ Social Workers
- ❑ Anyone involved with CPS
- ❑ Law enforcement
- ❑ Attorneys
- ❑ Judges
- ❑ Guardians ad litum
- ❑ CASAs
- ❑ Pediatricians
- ❑ Child abuse pediatricians
- ❑ Emergency room doctors
- ❑ Other (please specify) _____

14 What should the following professionals know about OI?

	That it exists	That it can be confused for child abuse	Who to contact for expert advice	Which tests can be done to test for it	How to interpret the test results	Other
CPS workers	❑	❑	❑	❑	❑	❑
Law Enforcement	❑	❑	❑	❑	❑	❑
Attorneys	❑	❑	❑	❑	❑	❑
Judges	❑	❑	❑	❑	❑	❑
Guardians ad litum/CASAs	❑	❑	❑	❑	❑	❑
Pediatricians	❑	❑	❑	❑	❑	❑
Other (please specify)	❑	❑	❑	❑	❑	❑

15 Has the issue of OI ever been raised in a child abuse case that you were involved with?

❍ Yes
❍ No

If No Is Selected, Then Skip To End of Block

16 In 2-3 sentences, please describe the case(s).

17 The following four questions pertain to the case you just described. If you described more than one case, please choose ONE case on which to base your responses for the next four questions.In the previously described case, did the parent(s) lose custody of the child(ren)?

❍ Yes
❍ No

18 How long was/were the child(ren) separated from their parents?

- ❍ Never separated
- ❍ 0-3 months
- ❍ 3-6 months
- ❍ 6 months-1 year
- ❍ 1-3 years
- ❍ Over 3 years
- ❍ Permanently separated

19 Was genetic testing for OI done during the investigation?

- ❍ No
- ❍ Yes, on the child(ren)
- ❍ Yes, on the parent(s)
- ❍ Yes, on the child(ren) and the parent(s)

20 Would increased knowledge of OI have been beneficial during this case?

- ❍ Yes
- ❍ No

21 About how many cases, on average, do you think your local firm/department/institution (not just your own cases) handles ANNUALLY in which there is a question of OI?

_____ .

22 To the best of your knowledge, on average, what percentage of cases in which there is a question of OI vs. abuse go to court?

_____ .

23 Below is a list of reasons that preliminary research suggests play a role in hindering genetic testing from happening, even when there is a question of OI as it might be related to alleged child abuse. Please select which reasons you think are the most salient. You may choose up to THREE.

- ❑ Lack of awareness about potential conditions
- ❑ Insufficient funds to cover diagnostic testing
- ❑ No access to child for diagnostic testing
- ❑ Concern of physically harming the child by the blood draw
- ❑ Concern of emotionally harming the child by the testing process
- ❑ Parents' resistance to going through with genetic testing
- ❑ Testing is irrelevant as most cases end with a guilty plea
- ❑ Lack of confidence in the validity of the genetic test
- ❑ Other (please explain) _____

24 Sometimes cases end in an early guilty plea before a court hearing begins, in order to reduce the chance of a more detrimental outcome from the trial. What is the proportion of child abuse cases that end in an early plea? If you do not have an estimate, please leave this question blank.

_____ .

25 After an accusation of physical child abuse is made, in your community, how easy is it to remove a child from their home?

- ❑ Very easy. A child is often removed right away; workers err on the side of child safety
- ❑ Easy. Some evidence needs to be found, but child safety is the main priority
- ❑ Somewhat difficult. The investigation must find substantial evidence of abuse or risk to the child of future abuse before a child is removed
- ❑ Very difficult. The investigation must prove beyond a reasonable doubt that abuse happened or there is substantial risk to the child of future abuse before a child is removed
- ❑ Additional Comments: _____

26 Who would you turn to first if you decided that you wanted to get a child evaluated for OI?

- ❍ Pediatrician with a specialty practice in child abuse
- ❍ Radiologist
- ❍ Geneticist
- ❍ Scientific expert
- ❍ Local clinic/primary care doctor
- ❍ Local univeristy
- ❍ Office of public defense
- ❍ Attorney
- ❍ CPS Worker
- ❍ Law enforcement
- ❍ Internet
- ❍ Other (please specify) _____

27 Please indicate whether you agree or disagree with the following statements

	Agree	Disagree
Testing for OI should happen if the child has multiple unexplained fractures but no other injuries, and no confession of abuse from any of the child's caregivers	❍	❍
Testing for OI should happen if the child has multiple unexplained fractures, even if there is a confession of abuse	❍	❍
Testing for OI should happen only if the child has a family history of fractures	❍	❍
Children should only be evaluated for OI if it is requested by the parent, scientific expert, or court	❍	❍
Whether or not to test a child for OI depends primarily on the child's and child's family's social history	❍	❍
All children evaluated for abuse should also be evaluated for OI	❍	❍

28 Who has the primary responsibility for requesting testing for OI? (You may select more than one)

- ❑ CPS Workers
- ❑ Attorneys
- ❑ Judges
- ❑ Pediatricians or child abuse pediatricians
- ❑ Law enforcement
- ❑ Scientific experts
- ❑ Other (please specify) _____

29 At what point in the investigation/trial process should genetic testing for OI be requested?

- ❍ During the investigation, before charges are pressed
- ❍ After charges are pressed, before the trial
- ❍ During the trial
- ❍ Other (please explain) _____

30 What concerns, if any, do you have about the use of genetic testing? (please select all that apply)

- ❑ No concerns
- ❑ Harm/trauma to the child
- ❑ Potential breach of confidentiality
- ❑ Potential misinterpretation of the test results
- ❑ Skepticism about the accuracy of the test
- ❑ Potential genetic discrimination
- ❑ Fear that the test result will reveal something else that was not anticipated (e.g. health related condition)
- ❑ Fear of increasing the likelihood of losing the case
- ❑ Cost
- ❑ Fear of introducing ambiguity or confusion, especially with inconclusive test results

31 Does your institution/department/firm have easy access to a medical expert?

- ❍ Yes
- ❍ No
- ❍ I'm not sure
- ❍ Other _____

32 If a medical expert is acquired, what is the likelihood that the expert will be knowledgeable in genetics?

- ❍ A geneticist can be requested
- ❍ Not likely
- ❍ Likely
- ❍ Other _____

33 Where do these experts come from?

- ❑ Local physicians
- ❑ An assortment of medical experts that have testified in court before
- ❑ Anyone with expertise in the pertinent area may be selected
- ❑ I don't know
- ❑ Other (please specify) _____

34 What is it like working with a scientific or medical expert?

- ❍ The expert is usually unbiased and considers all possibilities of unexplained fractures equally _____
- ❍ The expert is usually biased towards injuries happening from child abuse _____
- ❍ The expert is usually biased towards injuries happening from accidents or medical causes _____

35 For the next four questions, please indicate how confident you are that the following statements are true. Scientific experts have the ability to interpret genetic test results.

- ❍ Very confident
- ❍ Somewhat confidence
- ❍ Neutral
- ❍ Not very confidence
- ❍ Not at all confident
- ❍ Other (please explain) _____

36 Attorneys have the ability to understand the implications of a genetic test.

- ❍ Very confident
- ❍ Somewhat confidence
- ❍ Neutral
- ❍ Not very confidence
- ❍ Not at all confident
- ❍ Other (please explain) _____

37 CPS workers have the ability to understand the implications of a genetic test.

- ❍ Very confident
- ❍ Somewhat confidence
- ❍ Neutral
- ❍ Not very confidence
- ❍ Not at all confident
- ❍ Other (please explain) _____

38 You could interpret a genetic test result on your own, without external help.

- ❍ Very confident
- ❍ Somewhat confidence
- ❍ Neutral
- ❍ Not very confidence
- ❍ Not at all confident
- ❍ Other (please explain) _____

39 How much confidence do you think the courts usually have in the validity of the genetic test?

- ❍ Very little to none
- ❍ A moderate amount
- ❍ Complete confidence
- ❍ It depends on the scientific expert's ability to explain the test
- ❍ It depends on the attorney's ability to explain the test
- ❍ It depends on the attorney and scientific expert's ability to explain the test
- ❍ Other (please explain) _____

40 Have you ever heard of a variant of unknown significance (VUS) in the context of genetic test results?

- ❍ Yes
- ❍ No

41 Please complete the following sentence to the best of your ability: A variant of unknown significance (VUS)...

- ❍ Should not be considered admissible evidence
- ❍ Should be treated as the equivalent of a negative test result
- ❍ Should be treated as the equivalent of a positive test result
- ❍ Needs to be followed up with DNA testing of the parents
- ❍ Is a mystery to me and I would need to seek help to understand what it means
- ❍ Other (please explain) _____

42 Which group(s) of professionals are most crucial to target for increasing education about OI? You may choose up to THREE.

- ❑ Attorneys
- ❑ CPS Workers
- ❑ The defendant
- ❑ Judges
- ❑ Law enforcement
- ❑ Pediatricians or child abuse pediatricians
- ❑ Other (please specify) _____

43 Which of the following methods do you think would be effective as a means to educate professionals in each field about OI?

	CPS Workers	Attorney	Judges	Pediatricians/Child Abuse Pediatricians	Law enforcement
Emailed information	❑	❑	❑	❑	❑
At a conference	❑	❑	❑	❑	❑
Via a webinar	❑	❑	❑	❑	❑
Using social media	❑	❑	❑	❑	❑
At a seminar or training	❑	❑	❑	❑	❑
Via a website	❑	❑	❑	❑	❑
Change in academic curriculum	❑	❑	❑	❑	❑
Other	❑	❑	❑	❑	❑

44 Do you believe that a policy change mandating genetic testing for OI under certain conditions within a child abuse investigation/case would be beneficial?

❍ Yes, a policy change like the following might be useful: _____
❍ No, because: _____

45 Do you think that practice guidelines for CPS workers or legal professionals would be beneficial to help these people explore alternative causes for unexplained fractures?

❍ Yes, something like the following might be helpful: _____
❍ No, because: _____

46 Do you think that most professionals involved in child abuse would be interested in learning more about OI?

❍ Yes _____
❍ No _____

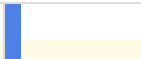
47 Do you have any additional comments about the issue of OI in relation to child abuse?

48 Thank you for your responses! If you would like to be considered for the drawing for one of five $20 Amazon gift cards, please enter your email address below. While the email address that you provide will be visible to researchers, it will not be associated with any of your answers, and does not have to be the email address associated with your work. The drawing will take place on June 15th, and winners will be notified by email.

Appendix 4: Survey Results

1. What state do you practice in?

#	Answer		Response	%
43	North Dakota		0	0%
46	Oregon		0	0%
40	New Mexico		0	0%
38	New Hampshire		0	0%
32	Minnesota		0	0%
33	Mississippi		0	0%
48	Rhode Island		0	0%
49	South Carolina		0	0%
59	Wyoming		0	0%
60	Outside the U.S.		0	0%
55	Virginia		0	0%
54	Vermont		0	0%
50	South Dakota		0	0%
28	Maine		0	0%
35	Montana		0	0%
24	Iowa		0	0%
23	Indiana		0	0%
21	Idaho		1	1%
36	Nebraska		1	1%
39	New Jersey		1	1%
25	Kansas		1	1%
17	Delaware		1	1%
16	Connecticut		1	1%
34	Missouri		1	1%
10	Alabama		1	1%
31	Michigan		1	1%
29	Maryland		1	1%
57	West Virginia		1	1%
15	Colorado		2	2%
11	Alaska		3	2%
12	Arizona		2	2%
52	Texas		2	2%
53	Utah		2	2%
58	Wisconsin		2	2%
47	Pennsylvania		2	2%
51	Tennessee		3	2%
42	North Carolina		2	2%
22	Illinois		2	2%
27	Louisiana		3	2%
26	Kentucky		2	2%
20	Hawaii		2	2%
37	Nevada		2	2%
45	Oklahoma		2	2%

30	Massachusetts		4	3%
13	Arkansas		4	3%
18	Florida		5	4%
19	Georgia		6	5%
14	California		7	6%
41	New York		7	6%
44	Ohio		11	9%
56	Washington		34	28%
	Total		122	100%

Statistic	Value
Min Value	10
Max Value	58
Mean	37.61
Variance	280.50
Standard Deviation	16.75
Total Responses	122

2. Please select the profession you most identify with

#	Answer		Response	%
7	For child protective services		28	20%
8	For another child welfare services program		12	9%
9	In an office charged with the legal representation of parents		7	5%
10	In an office charged with the legal representation of children		0	0%
11	In a guardian ad litem office		0	0%
12	In an office under contract with the state to provide services to children and families		13	9%
13	Volunteer CASA		2	1%
14	Attorney GAL		1	1%
15	Paid non-attorney GAL		0	0%
16	Representing parents in dependency proceedings		46	33%
17	Representing parents in criminal proceedings		14	10%
18	Representing children		3	2%
19	Representing the state in dependency		1	1%

	proceedings			
20	Representing the state in criminal proceedings		1	1%
21	Commissioner		0	0%
22	Superior court Judge		0	0%
23	Other (please specify)		12	9%
	Total		140	100%

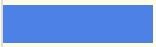
Other (please specify)
Clinical forensic medical specialist
social work advocate for parents
innocence network
Medical Social Worker
Medical Social Worker
Hospital Social Worker/Therapist
medical social worker
Hospital Social Worker
Family & Children's Ombudsman
Foster parent
therapist working with children in custody and their parents

Statistic	Value
Min Value	7
Max Value	23
Mean	13.53
Variance	24.41
Standard Deviation	4.94
Total Responses	140

3. How many of each type of staff currently work at your local firm/department/institution?

#	Answer	Min Value	Max Value	Average Value	Standard Deviation
1	Attorneys	0.00	200.00	9.57	28.33
2	CPS workers	0.00	1,200.00	20.52	104.33

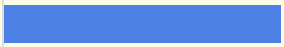
4. To what extent have you ever had training in genetics?

#	Answer		Response	%
1	I've never had any training in genetics		44	32%
2	It was covered in a biology class I took as undergraduate in college		69	50%
3	I took a genetics class in college		10	7%
4	I've taken two or more genetics classes in college or later		4	3%
7	Other (please specify)		12	9%
	Total		139	100%

Other (please specify)
Medical school and research
Trainings at conferences on several occasions
high school biology
just through legal cases re child abuse
reading about issues over last 30 years
took genetics class in college, parent of a child with a chromosome disorder
Ongoing through the hospital
Post Gruate courses
my oldest child has had extensive genetics testing due to developmental issues and I have been provided lots of research from Seattle Children's Hospital on the issues
training at work
Researched genetics in medical child abuse cases

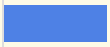
Statistic	Value
Min Value	1
Max Value	7
Mean	2.24
Variance	2.62
Standard Deviation	1.62
Total Responses	139

5. To what extent do you feel comfortable reading medical or scientific journal articles?

#	Answer		Response	%
4	I've never read a medical or scientific journal before		2	1%
5	I don't feel very comfortable reading medical or scientific journals. I would need help understanding most of it		25	18%
6	I feel fairly comfortable reading medical or scientific journals, although I would need help understanding some of it		82	58%
7	I have no problem reading medical or scientific journal articles		32	23%
	Total		141	100%

Statistic	Value
Min Value	4
Max Value	7
Mean	6.02
Variance	0.46
Standard Deviation	0.68
Total Responses	141

6. How did you first hear about osteogenesis imperfecta (OI)?

#	Answer		Response	%
1	The email that contained the link to this survey		27	22%
2	On the job		51	41%
3	Media (book, TV, radio, etc)		13	11%
4	From a friend		3	2%
5	At school/university		14	11%
6	At a work-related training/webinar		5	4%
7	Other (please specify)		10	8%
	Total		123	100%

Other (please specify)
have no idea whar osteogenesis imperfecta is
Case of brittle bones disease
often comes up in child abuse cases
a clients dependency case
Academic journal
this survey
Never
former client had it
Prior employment with Child Advocacy Center
during research on a case

Statistic	Value
Min Value	1
Max Value	7
Mean	2.85
Variance	3.44
Standard Deviation	1.86
Total Responses	123

7. Have you ever attended a work-related training in which OI was mentioned? If so, what was the training?

#	Answer		Response	%
1	No		85	70%
2	Yes		37	30%
	Total		122	100%

Yes
ABA training
Forensic Science CLE
SBS COCVENTION 2014
a training on child abuse "mimics"
Medical Child Abuse session at a conference
CJC - Dr. Sugar
have no idea whar osteogenesis imperfecta is
I don't recall
ABA parent attorney conference
Child abuse
handling severe child abuse cases
I have lectured on metabolic bone disease in infants, wrongly attributed to child abuse - OI is just one of many bone dysplasias
CLE Training; ABA Parent Rep Conference
Abuse training as an attorney representing the city before becoming a parent's attorney
joint contracter's conference, wenatchee
Children's Justice
presentation given by child abuse pediatrician.
Trainings on abuse by child abuse physicians
CORE training
Ongoing during team meetings at the hospital
Assessing non-accidental injuries
Nursing ceu
Medical Aspects of Child Abuse
Identifying non-accidental injuries
A learning didactic on OI
general training on physical abuse
genetic training at work
Marine Corps Community Services
NACDL Child Abuse Training
Don't remember the name

Statistic	Value
Min Value	1
Max Value	2
Mean	1.30
Variance	0.21
Standard Deviation	0.46
Total Responses	122

8. Please select which of the following statements apply to you (choose all that apply)

#	Answer		Response	%
1	I have little to no understanding of OI		33	27%
2	I am aware of OI and know that it can be confused for child abuse		82	67%
3	I can determine when it is appropriate to request diagnostic testing for OI		18	15%
4	I have no problem interpreting genetic test results for OI		3	2%

Statistic	Value
Min Value	1
Max Value	4
Total Responses	122

9. Please select which ONE of the following statements you most agree with

#	Answer		Response	%
1	OI never plays a role in child abuse cases		2	2%
2	Fractures due to OI can be confused for fractures due to child abuse		116	98%
	Total		118	100%

Statistic	Value
Min Value	1
Max Value	2
Mean	1.98
Variance	0.02
Standard Deviation	0.13
Total Responses	118

10. Please indicate how strongly you agree or disagree with the following statement:Cases where alleged physical child abuse could be explained by OI are problematic and need to be addressed better. 1 = completely agree5 = completely disagree

#	Answer	Min Value	Max Value	Average Value	Standard Deviation	Responses
1	.	1.00	5.00	2.42	1.54	106

11. In your opinion, what percentage of professionals in your field do you expect to have heard of OI before?

#	Answer	Min Value	Max Value	Average Value	Standard Deviation	Responses
3	.	0.00	100.00	42.41	28.39	118

12. Who should know about OI? (Please select all that apply)				
#	Answer		Response	%
11	Other (please specify)		21	17%
7	CASAs		98	80%
3	Law enforcement		104	85%
6	Guardians ad litum		106	86%
2	Anyone involved with CPS		108	88%
5	Judges		109	89%
4	Attorneys		113	92%
1	Social Workers		113	92%
10	Emergency room doctors		115	93%
8	Pediatricians		117	95%
9	Child abuse pediatricians		118	96%

Other (please specify)
Nurses
All of the above
Social Service Agencies
hospital interpreters
have no idea whar osteogenesis imperfecta is
parents
all of the above
parents
parents
Teachers (major referral source)
all of the above
OI is only one of the MBDs - almost too much emphasis put on it
Families with children of multiple births and premature babies
Physical, Occupational Therapists
Private Organization caseworkers
Nurses
teachers
Parents
PArents
hospital social workers
therapists and counselors who work with the cases and families

Statistic	Value
Min Value	1
Max Value	11
Total Responses	123

13. What should the following professionals know about OI?

#	Question	That it exists	That it can be confused for child abuse	Who to contact for expert advice	Which tests can be done to test for it	How to interpret the test results	Other	Total Responses
2	CPS workers	95	100	103	66	44	4	412
3	Law Enforcement	96	93	74	31	23	1	318
4	Attorneys	89	93	99	72	55	2	410
5	Judges	89	97	68	57	47	3	361
6	Guardians ad litum/CASAs	90	94	84	59	42	2	371
8	Pediatricians	90	96	94	96	96	3	475
10	Other (please specify)	8	7	7	6	5	1	34

Other (please specify)
have no idea whar osteogenesis imperfecta is
Again, not just OI -- all forms of metabolic bone disease
Teachers
Physical and Occupational Therapists
Private organization caseworkers
Nurses
teachers
ER doctors

Statistic	CPS workers	Law Enforcement	Attorneys	Judges	Guardians ad litum/CASAs	Pediatricians	Other (please specify)
Min Value	1	1	1	1	1	1	1
Max Value	6	6	6	6	6	6	6
Total Responses	120	114	114	112	112	113	10

14. Has the issue of OI ever been raised in a child abuse case that you were involved with?

#	Answer		Response	%
1	Yes		67	55%
2	No		54	45%
	Total		121	100%

Statistic	Value
Min Value	1
Max Value	2
Mean	1.45
Variance	0.25
Standard Deviation	0.50
Total Responses	121

15. In 2-3 sentences, please describe the case(s).
Text Response
Father accused of abuse due to fractures. Child protection team doc said not OI based on folk lore about OI. Case pled down to misdemeanor after excoriating Doctor in deposition.
in a child death, the child had many risk factors for OI but the prosecutor proceeded anyway; we represented on postconviction on newly discovered evidence and ineffective assistance
There was a situation where a child had multiple long bone fractures of varying ages. Some of these breaks occurred while under the care of the NICU. The child had multiple precursors for OI.
Multiple fracture case in infant.
Father charged with abuse of his child. Child had several broken bones.
Mother and father with no other risk factors for abuse (i.e., no drugs, stable relationship/employment) had premature infant who soon thereafter died from "child abuse."
My client was wrongfully convicted of killing his baby. It turns out she had vitamin D deficiency in addition to having been dropped from about 3 feet.
Many many cases of unexplained fractures in which the much more common metabolic bone disease infantile rickets was the underlying disorder. OI is so rare. 50-100 cases per year vs 3.4 million newborns per year
confidential
child had multiple fractures, parents adamant that no child abuse was involved
Child had broken bones, FTT, as well as Vitamin D deficiency.
Child with unexplained broken bone
2 cases in 2 different counties. In each case so called abused child returned to parents. Family traumatized by treatment of medical professionals. MD refused to acknowledge that causation is an opinion in fractures, not a certainty without corroboration.
2 medical experts from cal. universities said the 19 broken bones in 4 month baby were OI not abuse. She was found innocent of crime but lost her baby in dependency case
have no idea whar osteogenesis imperfecta is, so can not answer. Probably need training
Infant child presents with fracture to ribs. No bruising or obvious sign of injury except child is showing pain when picked up, but not laying down.
4-month old infant, multiple fractures, arms, legs and skull. Was admitted to ER by parents stating he wouldn't stop crying. ER tested for and ruled genetic abnormalities to explain injuries.
Child presented with several unexplained fractures.
The case involved an unexplained broken arm that both parents deny they were at fault. As a result, the CA pediatrician labeled it as unexplained blunt trauma resulting in the break of the arm which was more likely caused by abuse.
broken bone dependency case, parents accused of causing broken bone in child
Client in military had recently moved from another state. Had twins taken to children's hospital and CPS was called. Mother took medication during pregnancy that could have caused fragile bones.
child abuse case
When I was an AAG (attorney representing the state) I had two major/extended trials where this issue arose and was a main focus of the case.
Parents insisted that the child had OI after fractured leg on infant.
Parents accused of abuse. Child had issues with bone density due to vitamin deficiency. Child removed and lots of money spent to prove medical doctor's diagnosis was wrong.

In cases where a child abuse doctor had given an opinion of abuse, a pediatric orthopaedist and/or a radiologist opined that a fracture or fractures could be or were likely the result of OI, or that an OI test was needed.
baby with broken leg
On a case I was consulting on, if it had not been for the relatives pushing for testing, it would not have been done. Even after OI was confirmed, the child abuse pediatrician would not withdraw the abuse diagnosis.
Baby with multiple fractures at multiple times: once before being placed in foster care and several fractures received later on in foster care.
questionable fractures
infants with broken bones.
Child abuse case involving brittle bones
Parents claimed that it was an explanation, but the cases never got to trial/evidentiary hearing where experts could be examined.
child had known, diagnosed and subtyped OI, but families still referred to CPS for fractures. In another case parent has OI, but child not diagnosed, and referred after fracture
it is raised in almost every case, only confirmed genetically in one (UW lab, Peter Byers)
8 month old baby had a spiral fracture of the tibia. Doctor said mother's explanation inconsistent with injury and called in case. Neighbor was the translator at hospital, and genetic testing was not done.
a young mom said that her ex husband was criminally charged with child abuse for the broken bone of their infant son. as far as I know there were no tests run for genetics. she told me they ran in their family.
Triplets all with multiple rib fractures.
A mother claimed OI was in their family history and therefore it should explain her daughter's fractures. However, OI was ruled out and due to the length of time it took to receive the results, the child was discharged to mother per the request of DHS.
Child rolled off a bench at hospital while mother was changing her.
The child had been removed due to suspected child abuse and returned once the OI had been diagnosed. I provided services to help the family prices their anger and mistrust of the system and how tomove forward w their child ha I got OI.
The child had broken bones, was young and had a possible diagnosis of OI. His parents had previous interactions with CPS with older children.
OI is typically used by defense attorneys in all cases involving broken bones due to alleged child maltreatment
Child is a 5 year old white female with a history of repeated brusing and at least one fracture. Bruising was suspect as finger marks/handprint. Child reported that her stepfather plays rough and sometimes she gets hurt.. She said that sometimes when he is mad he spanks her. Family obtained a letter from their pediatrician that stated he felt child had OI. Case was dismissed in court.
Child with broken arm
Non-ambulatory infant with femur fractures, another non-ambulatory infant with multiple fractures of various bones. Both children were tested for OI
Child suspected to have OI. Placed in foster home which the caregivers are exteremly careful. biological parents are losing the child due to lack of understanding and support.
Child had multiple fractures of varying timeframes. Parents sought testing for OI and was

negative. Parents sought other medical explanations for injuries but fractures were ruled as probable child abuse
Don't remember the specifics
Humerus fracture of an infant. Explanation inconsistent with injury.
The child had multiple fractures and the parents/their attorneys wanted to rule OI out
A child with multiple fractures presented without a consistent history of how these fractures could have occurred. Our team came to the conclusion that the injuries were abusive in nature but the child's mother wanted OI testing. The child was tested and she did not have OI.
I reviewed a CPS investigaiton of an infant with broken leg in which the defence attorney for the parents raised this possibility as there was no clear perpetrator of abuse, medical child abuse experts were consulted and there was disagreement. as to whether it was OI or not. Case was settled.
young girl who had multi broken bones. child denied any abuse. parents from Portugal. children were removed multi times and the bone broke in foster care too. test finally done and showed OI
I ran family meetings and feel this condition was used a lot as an excuse for the child's injuries. Feel this condition is rare compared to the amount of avoide that takes place.
Fractured femur leg sprain claimed child was trying to climb out of crib and must've gotten caught
case happened years ago and can't rmember all the facts of the case
Unexplained broken bones in non-verbal child. Testing ordered & child did not have condition.
Mentioned in several cases involving multiple fractures and where lab tests show abnormalities in blood work.
The parents claimed that the child had OI and that is why the child had fractures and bruising. We discussed this repeatedly with the child's Ped. Dr. who determined that the child showed no signs of OI at all and no family history of OI. The doctor offered to do testing for OI but recommended testing the parents first before the child as the child had already been through many tests and many medical procedures and was traumatized by the hospital. One parent agreed to be tested and the other parent (who requested OI testing for the child) refused to be tested. There were other cases as well.

Statistic	Value
Total Responses	60

16. The following four questions pertain to the case you just described. If you described more than one case, please choose ONE case on which to base your responses for the next four questions.In the previously described case, did the parent(s) lose custody of the child(ren)?

#	Answer		Response	%
1	Yes		40	66%
2	No		21	34%
	Total		61	100%

Statistic	Value
Min Value	1
Max Value	2
Mean	1.34
Variance	0.23
Standard Deviation	0.48
Total Responses	61

17. How long was/were the child(ren) separated from their parents?

#	Answer		Response	%
1	Never separated		9	16%
2	0-3 months		4	7%
3	3-6 months		10	18%
4	6 months-1 year		12	21%
5	1-3 years		10	18%
6	Over 3 years		5	9%
7	Permanently separated		7	12%
	Total		57	100%

Statistic	Value
Min Value	1
Max Value	7
Mean	3.93
Variance	3.57
Standard Deviation	1.89
Total Responses	57

18. Was genetic testing for OI done during the investigation?

#	Answer		Response	%
1	No		28	47%
2	Yes, on the child(ren)		26	43%
3	Yes, on the parent(s)		1	2%
4	Yes, on the child(ren) and the parent(s)		5	8%
	Total		60	100%

Statistic	Value
Min Value	1
Max Value	4
Mean	1.72
Variance	0.75
Standard Deviation	0.87
Total Responses	60

19. Would increased knowledge of OI have been beneficial during this case?

#	Answer		Response	%
1	Yes		50	85%
2	No		9	15%
	Total		59	100%

Statistic	Value
Min Value	1
Max Value	2
Mean	1.15
Variance	0.13
Standard Deviation	0.36
Total Responses	59

20. About how many cases, on average, do you think your local firm/department/institution (not just your own cases) handles ANNUALLY in which there is a question of OI?

#	Answer	Min Value	Max Value	Average Value	Standard Deviation	Responses
1	.	0.00	50.00	9.27	12.82	98

21. To the best of your knowledge, on average, what percentage of cases in which there is a question of OI vs. abuse go to court?

#	Answer	Min Value	Max Value	Average Value	Standard Deviation	Responses
1	.	0.00	100.00	47.18	39.92	92

22. Below is a list of reasons that preliminary research suggests play a role in hindering genetic testing from happening, even when there is a question of OI as it might be related to alleged child abuse. Please select which reasons you think are the most salient. You may choose up to THREE.

#	Answer		Response	%
4	Concern of physically harming the child by the blood draw		5	5%
5	Concern of emotionally harming the child by the testing process		10	9%
8	Lack of confidence in the validity of the genetic test		20	18%
3	No access to child for diagnostic testing		23	21%
7	Testing is irrelevant as most cases end with a guilty plea		26	24%
6	Parents' resistance to going through with genetic testing		31	28%
9	Other (please explain)		32	29%
2	Insufficient funds to cover diagnostic testing		65	60%
1	Lack of awareness about potential conditions		99	91%

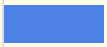
Other (please explain)
State and law enforcement have no interest in the truth.
CPS believes the parent(s) are guilty of abusing their child and don't seek alternative explanations
Overdelegation of diagnostic responsibility to "child abuse" peditricians who are underqualified to rule out this diagnosis.
Prosecutor, CPS and pediatricians aversion to believing it is an issue and preference to jump to the conclusion that it is abuse.
Lack of interest in discovering it and its potential impact on the prosecutor of child abuse
CASA or GAL meddling
Cases are resolved by agreement prior to trial, so no testing.
Doc for state are biased
have no idea whar osteogenesis imperfecta is, so can not answer. Probably need training
Belief that any injury is child abuse
fear of results
Court is inclined to follow medical doctor's opinion when abuse is suggested upon initial investigation and look no further.
Refusal to consider options other than abuse as the cause of the injury
Opposition from child abuse pediatricians to testing or lying about testing being negative
Don't know
The parents fail to maintain communication with counsel or the child safety workers and often lose their rights to contest for failing to appear at hearings
As I said, OI is just one of many possibilities - if excluded, used against the parents; if found, is ignored by prosecutors and CPS. So worth doing but it probably least likely cause of abnormal bone formations (Vit D, Ehlers Danlos etc. much more common)
Automatic dismissal of OI as a lofty diagnosis
Child abuse "expert" deciding it was child abuse instead of further
amount of time to get the results back - child not needing hospitalization that long
Transportation
Child abuse pediatricians rule OI out prior to any testing
Doctors determine that OI is not involved based on the child's lack of symptoms and that the abuse is caused by child abuse.

Statistic	Value
Min Value	1
Max Value	9
Total Responses	109

23. Sometimes cases end in an early guilty plea before a court hearing begins, in order to reduce the chance of a more detrimental outcome from the trial. What is the proportion of child abuse cases that end in an early plea? If you do not have an estimate, please leave this question blank.

#	Answer	Min Value	Max Value	Average Value	Standard Deviation	Responses
1	.	0.00	97.00	64.06	28.73	36

24. After an accusation of physical child abuse is made, in your community, how easy is it to remove a child from their home?

#	Answer		Response	%
1	Very easy. A child is often removed right away; workers err on the side of child safety		49	45%
2	Easy. Some evidence needs to be found, but child safety is the main priority		34	31%
3	Somewhat difficult. The investigation must find substantial evidence of abuse or risk to the child of future abuse before a child is removed		23	21%
4	Very difficult. The investigation must prove beyond a reasonable doubt that abuse happened or there is substantial risk to the child of future abuse before a child is removed		3	3%
5	Additional Comments:		4	4%

Additional Comments:
This is a terrifying outcome and more prevalent in communities with a CHILDRENS HOSPITAL and those nasty mean spirited child abuse pediatricians who do not understand that ALL ALLEGATIONS OF ABUSE devastate families.
The doctors say child abuse and the police can order that the parents not have any contact with the child even if the child is in the hospital and not physically in parent's custody. How devastating for a hospitalized child not to hear the sounds or feel the comforting touch of loving parents. Just because medical personnel are underqualified and the police are too deferential.
Too easy
Very easy if child abuse pediatrician diagnoses abuse which ends the investigation

Statistic	Value
Min Value	1
Max Value	5
Total Responses	109

25. Who would you turn to first if you decided that you wanted to get a child evaluated for OI?

#	Answer		Response	%
12	Local univeristy		1	1%
13	Internet		1	1%
10	Law enforcement		1	1%
7	Attorney		1	1%
14	Scientific expert		2	2%
8	CPS Worker		3	3%
3	Radiologist		5	5%
6	Office of public defense		7	6%
4	Geneticist		10	9%
5	Local clinic/primary care doctor		10	9%
11	Other (please specify)		11	10%
2	Pediatrician with a specialty practice in child abuse		57	52%
	Total		109	100%

Other (please specify)
Primary care doctor for my child; a pediatric radiologist for my clients
have no idea whar osteogenesis imperfecta is, so can not answer. Probably need training
nurse consultant
Pediatrician, but not one with a specialty in chid abuse. Those guys can be quacks and always find abuse.
Pediatric bone specialist--radiologist or orthopedist
Regional children's hospital that completes all investigation if physical Abuse it's standard and completed within in the 1st week of removal.
Judge. I would probably need a court order for testing.
CPS Supervisor/ Program manager
child welfare liaison housed at state children's hospital
my agencies medical consultant
Pediatrician or other specialist such as the Orthopaedist

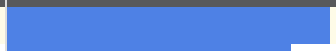
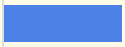
Statistic	Value
Min Value	2
Max Value	14
Mean	4.37
Variance	11.42
Standard Deviation	3.38
Total Responses	109

26. Please indicate whether you agree or disagree with the following statements					
#	Question	Agree	Disagree	Total Responses	Mean
1	Testing for OI should happen if the child has multiple unexplained fractures but no other injuries, and no confession of abuse from any of the child's caregivers	98	9	107	1.08
2	Testing for OI should happen if the child has multiple unexplained fractures, even if there is a confession of abuse	64	43	107	1.40
3	Testing for OI should happen only if the child has a family history of fractures	18	85	103	1.83
4	Children should only be evaluated for OI if it is requested by the parent, scientific expert, or court	34	70	104	1.67
5	Whether or not to test a	28	75	103	1.73

	child for OI depends primarily on the child's and child's family's social history				
6	All children evaluated for abuse should also be evaluated for OI	33	74	107	1.69

Statistic	Testing for OI should happen if the child has multiple unexplained fractures but no other injuries, and no confession of abuse from any of the child's caregivers	Testing for OI should happen if the child has multiple unexplained fractures, even if there is a confession of abuse	Testing for OI should happen only if the child has a family history of fractures	Children should only be evaluated for OI if it is requested by the parent, scientific expert, or court	Whether or not to test a child for OI depends primarily on the child's and child's family's social history	All children evaluated for abuse should also be evaluated for OI
Min Value	1	1	1	1	1	1
Max Value	2	2	2	2	2	2
Mean	1.08	1.40	1.83	1.67	1.73	1.69
Variance	0.08	0.24	0.15	0.22	0.20	0.22
Standard Deviation	0.28	0.49	0.38	0.47	0.45	0.46
Total Responses	107	107	103	104	103	107

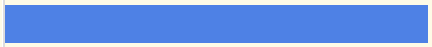
27. Who has the primary responsibility for requesting testing for OI? (You may select more than one)

#	Answer		Response	%
1	CPS Workers		74	68%
2	Attorneys		65	60%
3	Judges		37	34%
4	Pediatricians or child abuse pediatricians		91	83%
5	Law enforcement		27	25%
6	Scientific experts		27	25%
7	Other (please specify)		12	11%

Other (please specify)
Parents
Prosecutors
Note re selection of attorneys: I do not think attorneys _should_ have primary responsibility for requesting testing; but they become a person who has primary responsibility because of the failures by the child abuse pediatricians.
Everyone in the system should be responsible for ensuring that (1) an abused child is not given back to abuser and just as much (2) an innocent person is not wrongfully convicted and a family torn apart.
MDB testiing is routinely opposed by CPS and the judges tend to support CPS position
have no idea whar osteogenesis imperfecta is, so can not answer. Probably need training
parents
parents
teachers
mental health therapists?
parents
Parents

Statistic	Value
Min Value	1
Max Value	7
Total Responses	109

28. At what point in the investigation/trial process should genetic testing for OI be requested?

#	Answer		Response	%
1	During the investigation, before charges are pressed		97	89%
2	After charges are pressed, before the trial		8	7%
3	Other (please explain)		4	4%
5	During the trial		0	0%
	Total		109	100%

Other (please explain)
As soon as possible. Kidd need their parents.
Anytime it becomes relevant as a possible explanation for injury
at whatever point it is a consideration
Case Dependent

Statistic	Value
Min Value	1
Max Value	3
Mean	1.15
Variance	0.20
Standard Deviation	0.45
Total Responses	109

29. What concerns, if any, do you have about the use of genetic testing? (please select all that apply)

#	Answer		Response	%
1	No concerns		42	39%
2	Harm/trauma to the child		10	9%
3	Potential breach of confidentiality		8	7%
4	Potential misinterpretation of the test results		43	40%
5	Skepticism about the accuracy of the test		23	21%
6	Potential genetic discrimination		11	10%
7	Fear that the test result will reveal something else that wasnot anticipated (e.g. health related condition)		9	8%
8	Fear of increasing the likelihood of losing the case		20	19%
9	Cost		31	29%
10	Fear of introducing ambiguity or confusion, especially with inconclusive test results		39	36%

Statistic	Value
Min Value	1
Max Value	10
Total Responses	108

30. Does your institution/department/firm have easy access to a medical expert?

#	Answer		Response	%
1	Yes		64	59%
2	No		26	24%
3	I'm not sure		14	13%
4	Other		4	4%
	Total		108	100%

Other
I have some access, but it is not easy.
Funds are available to pay for experts, but the process of identifying the appropriate expert and lining up funding can be burdensome.
I would not describe the access as easy.
They are available outside of our community

Statistic	Value
Min Value	1
Max Value	4
Mean	1.61
Variance	0.73
Standard Deviation	0.85
Total Responses	108

31. If a medical expert is acquired, what is the likelihood that the expert will be knowledgeable in genetics?

#	Answer		Response	%
1	Not likely		25	24%
2	Likely		20	19%
3	A geneticist can be requested		51	49%
4	Other		8	8%
	Total		104	100%

Other
Not sure
unknown
Depends on the expert.
I have no way of knowing
Unsure
not sure
Unsure but certainly could make a referral if needed

Statistic	Value
Min Value	1
Max Value	4
Mean	2.40
Variance	0.88
Standard Deviation	0.94
Total Responses	104

32. Where do these experts come from?

#	Answer		Response	%
1	Local physicians		33	35%
2	An assortment of medical experts that have testified in court before		43	46%
3	Anyone with expertise in the pertinent area may be selected		44	47%
4	Other (please specify)		7	8%

Other (please specify)
Child abuse pediatricians
local children's hospital
Contracted MD
Oakland Children's Hospital I think
Children's Hospital of Pittsburgh
Contracted forensic medical center
the childrens hospital in the state

Statistic	Value
Min Value	1
Max Value	4
Total Responses	93

33. What is it like working with a scientific or medical expert?

#	Answer		Response	%
1	The expert is usually unbiased and considers all possibilities of unexplained fractures equally		35	81%
3	The expert is usually biased towards injuries happening from child abuse		8	19%
4	The expert is usually biased towards injuries happening from accidents or medical causes		0	0%
	Total		43	100%

The expert is usually unbiased and considers all possibilities of unexplained fractures equally	The expert is usually biased towards injuries happening from child abuse	The expert is usually biased towards injuries happening from accidents or medical causes

Statistic	Value
Min Value	1
Max Value	3
Mean	1.37
Variance	0.62
Standard Deviation	0.79
Total Responses	43

34. Attorneys have the ability to understand the implications of a genetic test.

#	Answer		Response	%
1	Very confident		9	8%
2	Somewhat confidence		37	34%
3	Neutral		17	16%
15	Not very confidence		30	28%
16	Not at all confident		14	13%
17	Other (please explain)		1	1%
	Total		108	100%

Other (please explain)
Some attorneys, but not all have sufficient knowledge to understand genetics with use of appropriate expert. Depends on back ground and training.

Statistic	Value
Min Value	1
Max Value	17
Mean	7.64
Variance	43.28
Standard Deviation	6.58
Total Responses	108

35. CPS workers have the ability to understand the implications of a genetic test.

#	Answer		Response	%
1	Very confident		6	6%
2	Somewhat confidence		22	20%
3	Neutral		13	12%
15	Not very confidence		28	26%
16	Not at all confident		38	35%
17	Other (please explain)		1	1%
	Total		108	100%

Other (please explain)
They do what the child abuse pediatician says, so if the CAP is a loon from SCH there will be no understanding.

Statistic	Value
Min Value	1
Max Value	17
Mean	10.50
Variance	43.19
Standard Deviation	6.57
Total Responses	108

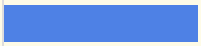
36. For the next four questions, please indicate how confident you are that the following statements are true.Scientific experts have the ability to interpret genetic test results.

#	Answer		Response	%
1	Very confident		31	29%
2	Somewhat confidence		44	41%
3	Neutral		19	18%
15	Not very confidence		5	5%
16	Not at all confident		2	2%
17	Other (please explain)		6	6%
	Total		107	100%

Other (please explain)
Maybe. "Scientific expert" is very broad and I am not sure what you mean.
Some experts, but not all experts have sufficient knowledge to testify about genetics. Depends on back ground and training.
lack knowledge
only if a geneticist
Unsure
Depends upon the expertise and credentials of the professional

Statistic	Value
Min Value	1
Max Value	17
Mean	3.60
Variance	22.30
Standard Deviation	4.72
Total Responses	107

37. You could interpret a genetic test result on your own, without external help.

#	Answer		Response	%
1	Very confident		1	1%
2	Somewhat confidence		10	9%
3	Neutral		17	16%
15	Not very confidence		33	31%
16	Not at all confident		44	41%
17	Other (please explain)		3	3%
	Total		108	100%

Other (please explain)
I always use an expert, I practice law and leave medicine to doctors and specialist, and in return they don't practice law.
I could not read the karotype on my own, but if given the genetics report with the report of the results, I could
no way I could

Statistic	Value
Min Value	1
Max Value	17
Mean	12.24
Variance	33.34
Standard Deviation	5.77
Total Responses	108

38. How much confidence do you think the courts usually have in the validity of the genetic test?

#	Answer		Response	%
1	Very little to none		2	2%
2	A moderate amount		20	19%
3	Complete confidence		12	11%
4	It depends on the scientific expert's ability to explain the test		29	27%
5	It depends on the attorney's ability to explain the test		4	4%
6	It depends on the attorney and scientific expert's ability to explain the test		37	35%
7	Other (please explain)		3	3%
	Total		107	100%

Other (please explain)
Can be hampered by the rhetoric of CAP
Depends on the judge
I don't know

Statistic	Value
Min Value	1
Max Value	7
Mean	4.27
Variance	2.65
Standard Deviation	1.63
Total Responses	107

39. Have you ever heard of a variant of unknown significance (VUS) in the context of genetic test results?

#	Answer		Response	%
1	Yes		18	17%
2	No		89	83%
	Total		107	100%

Statistic	Value
Min Value	1
Max Value	2
Mean	1.83
Variance	0.14
Standard Deviation	0.38
Total Responses	107

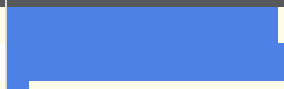
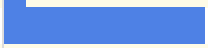
40. Please complete the following sentence to the best of your ability:A variant of unknown significance (VUS)...

#	Answer		Response	%
1	Should not be considered admissible evidence		3	3%
2	Should be treated as the equivalent of a positive test result		3	3%
3	Needs to be followed up with DNA testing of the parents		10	10%
4	Should be treated as the equivalent of a negative test result		2	2%
5	Is a mystery to me and I would need to seek help to understand what it means		78	77%
6	Other (please explain)		5	5%
	Total		101	100%

Other (please explain)
I need to research this to answer appropriately.
Depends on the situation.
I don't know what that is.
unsure
Should be explained to the court and used for purposes of argument by the attorneys (if I remeber correctly what it is)

Statistic	Value
Min Value	1
Max Value	6
Mean	4.62
Variance	1.08
Standard Deviation	1.04
Total Responses	101

41. Which group(s) of professionals are most crucial to target for increasing education about OI? You may choose up to THREE.

#	Answer		Response	%
1	Attorneys		61	57%
2	CPS Workers		89	83%
3	The defendent		5	5%
4	Judges		45	42%
5	Law enforcement		36	34%
6	Pediatricians or child abuse pediatricians		84	79%
7	Other (please specify)		2	2%

Other (please specify)
You email me at clappertonlaw#gmail.com a training then ask this question
Genetic specialists

Statistic	Value
Min Value	1
Max Value	7
Total Responses	107

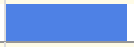
42. Which of the following methods do you think would be effective as a means to educate professionals in each field about OI?

#	Question	CPS Workers	Attorney	Judges	Pediatricians/Child Abuse Pediatricians	Law enforcement	Total Responses
1	Emailed information	44	44	40	41	39	208
2	At a conference	89	83	79	89	78	418
3	Via a webinar	54	65	55	51	47	272
4	Using social media	29	19	15	19	21	103
5	At a seminar or training	90	85	79	81	82	417
6	Via a website	37	46	35	38	36	192
8	Change in academic curriculum	53	27	25	62	40	207
9	Other	5	4	4	5	5	23

Emailed information	At a conference	Via a webinar	Using social media	At a seminar or training	Via a website	Change in academic curriculum	Other

Statistic	Emailed information	At a conference	Via a webinar	Using social media	At a seminar or training	Via a website	Change in academic curriculum	Other
Min Value	1	1	1	1	1	1	1	1
Max Value	5	5	5	5	5	5	5	5
Total Responses	58	96	69	32	96	53	72	6

43. Do you believe that a policy change mandating genetic testing for OI under certain conditions within a child abuse investigation/case would be beneficial?

#	Answer		Response	%
1	Yes, a policy change like the following might be useful:		76	75%
2	No, because:		26	25%
	Total		102	100%

Yes, a policy change like the following might be useful:	No, because:
Test when there are unexplained fractures of different ages.	Mandating testing, particularly genetic testing, would likely run the risk inadvertently being required to release confidential medical records to the courts, including any results of other conditions not expected to be found.
In cases of multiple fractures with no obvious cause	Would not mandate genetic testing, but perhaps a policy suggesting it
to increase awareness of the issue of OI as a possible affirmative defense/prevention of charges	it would likely be another unfunded mandate
Every fracture case gets it. You want to break up a family. Make sure there is REASON	defense attorneys need to make a call in individual cases
In CA there are certain mandatory tests on newborns, for example. Mandatory testing -- under certain circumstances -- might mitigate harm done to family's by the ignorance of other professionals.	There's a lot of things considered "abuse" so the policy would need to be more specific.
OI and other issues must be examined	there should be other indicia that such testing is needed.
OI testing with unexplained fractures.	The child safety workers in my state don't follow their own policies.
I emphasize "Might"	there are physician experts who can determine when OI testing is helpful and when it is not. In many of the cases I've seen, the radiologist, child abuse pediatrician, and others are able to determine that the child's bones appear normal on x-ray and would likely show some type of abnormality if they had OI. Thus, the testing is not necessary in every child abuse case.
all CPS shall receive yearly mandatory training regarding OI and other medical conditions that can be confused with Child Abuse	Our doctors are typically able to tell whether this is necessary or could be possible (a child having OI).
If it was well-informed and funded	it may unneccessarily delay cases where it is not a significant concern and abuse has occured.
For unexplained fractures, with a child abuse pediatrician diagnosing abuse, the agency should get second opinions from pediatric bone specialist and geneticist.	Genetic testing should be a personal choice by the child and/or parent.
If we are able to identify a cause for injuries and eliminate "child abuse" prior to the filing of a petition, that would reduce unnecessary filings against parents and caretakers.	most children that are abused come from low income homes and the abuse is most likely substantiated by the parents confessing.
Yes for all bone breaks, especially with no	I don't believe there is enough knowledge

other injuries	about this or confidence in the experts at this time to justify the resources. Right now, education is key.
if there is a family history or other medical factors indicating a possible link to cause other than abuse	It would waste time.
in cases with multiple fractures, it should be mandated to cover all bases	Too few cases would fit the conditions. Should be determined on a case by case basis.
When there is no clear perpetrator, no other injuries, and no history of injuries	every case is unique
depends on other factors too (ie cost)	policy changes rarely have real impact due to lack of training on said policy
	current policy is fine
	There are so few cases of OI and families hang onto the idea as a last resort when they cannot come to terms with the fact that someone hurt their child.
	I don't believe this would be a CPS policy. We follow the advice and recommendations of a physician. We typically don't tell physicialns how to do their jobs. Generally. IF there is a policy it probably should come from medicaid or insurance companies since there is a cost associated and needs to be considered allowable. Most parents will not have the means to pay for it otherwise and an agency wouldn't be responsible until after obtaining custody which is, of course, after proving the injury wasn't explained by something other than abuse.

Statistic	Value
Min Value	1
Max Value	2
Mean	1.25
Variance	0.19
Standard Deviation	0.44
Total Responses	102

44. Do you think that practice guidelines for CPS workers or legal professionals would be beneficial to help these people explore alternative causes for unexplained fractures?

#	Answer		Response	%
1	Yes, something like the following might be helpful:		91	91%
2	No, because:		9	9%
	Total		100	100%

Yes, something like the following might be helpful:	No, because:
Pursue testing for alternative causes of injuries.	Putting practice guidelines in place for attorneys could have extreme negative impacts on attorney-client privilege and relationships in the criminal context. Educating attorneys without requiring specific courses of action would best allow client's to maintain control over their cases while still allowing attorneys to effectively argue on their clients' behalf.
Testing is necessary rather than speculating about possible causes	You better start with educating defense attorneys educating the list you gave is hopeless
to clarify a situation where no abuse has occurred, however a child is injured.	They don't follow practice guidelines
Check list of mandated testing	I feel that medical professionals should decipher the results/make suggestions for the child's well being.
There is too much delegation of responsibility to medical professionals. Practice guidelines might be useful to encourage CPS workers and legal professionals know when a specialist might be needed or a second opinion obtained when reviewing medical records.	As a CPS worker, I rely on my medical staff to know this stuff.
Consideration of vit d defic/infantile rickets	workers aren't determining medical cuase - the doctors are. So if doctors should be explaining likely alternative causes maybe their practice guidelines should change.
same	
Have a go-to doctor that is aware of OI and can provide expert information on the case.	
manual	
a standard simply explained medical protocol would be beneficial for CPS and Attorney to follow along to see if all the steps were required to rule out OI in possible Child abuse cases	
But only if the child abuse pediatrician is willing to investigate medical causes. The agencies and courts give the CAP more power than they should have.	
One page sheet.	
Should the case involve unexplained fractures, the CPS worker should consult with Identified Expert on issue of OI to determine viability of that explaining the injuries	

having guidelines for parent defense attorneys to request testing at the initial removal hearing would be helpful, but the time for testing often exceeds the timeline to have a trial on the child abuse	
Explanation of the diagnosis and frequency as a valid defense as opposed to an unlikely, easily dismissed explanation	
however, it seems it could be used by some to argue abuse did not occur when it did. It seems that OI and abuse are not mutually exclusive.	
Increased education and trainings on medical conditions that can be confused for child abuse for mandated reporters.	
Training. What testing is used. Where to seek expert advise. Where to get testing done. Costs associated. Any funding sources available, i.e. covered by health insurance	
same as above	
Make sure you consult a child abuse pediatrician when a child has multiple unexplained fractures.	
checklist to go through, or a decision tree based on information gathered during investigation	

Statistic	Value
Min Value	1
Max Value	2
Mean	1.09
Variance	0.08
Standard Deviation	0.29
Total Responses	100

45. Do you think that most professionals involved in child abuse would be interested in learning more about OI?

#	Answer		Response	%
1	Yes		93	90%
2	No		10	10%
	Total		103	100%

Yes	No
Lawyers and CPS workers, yes; "child abuse" pediatricians are zealots and not neutral or impartial medical experts.	State workers are biased against marginal parents
attorneys	They should be very interested and willing to keep an open mind but I'm not sure that is actually the case.
If they can be convinced it is important to their work.	
Although I think some might be resistant to changing their views.	

Statistic	Value
Min Value	1
Max Value	2
Mean	1.10
Variance	0.09
Standard Deviation	0.30
Total Responses	103

46. Do you have any additional comments about the issue of OI in relation to child abuse?

Text Response
No, but if you need further questions answered, I am a Child Welfare Law Specialist in Georgia and handle a multitude of medical child abuse cases. I am also a 1996 UW Graduate!

Statistic	Value
Total Responses	1

47. Thank you for your responses! If you would like to be considered for the drawing for one of five $20 Amazon gift cards, please enter your email address below.While the email address that you provide will be visible to researchers, it will not be associated with any of your answers, and does not have to be the email address associated with your work. The drawing will take place on June 15th, and winners will be notified by email.

Statistic	Value
Total Responses	35