Brittle Bone Disease (Osteogenesis Imperfecta)

INTRODUCTION

I decided to cover this Osteogenesis imperfecta after I heard it mentioned, in discussion, during my second student-teaching assignment. Its mention piqued my curiosity and incited me to learn more. As a student in this "Deaf Plus" class, I am interested to see how this disease interplays with deafness and how it affects a student's educational experience.

Brittle bone disease (Osteogenesis imperfecta (OI)), first defined by McKusick in 1956, is a disease that causes extremely fragile bones. It is a congenital disease - meaning that it is present during birth. It is often caused by a defect in the gene that produces type I collagen - an important building block of bone and the most abundant protein found in the body. This gene can be affected in many different ways. Osteogenesis imperfecta "results from mutation involving several genes responsible for synthesis or intercellular processing of type I collagen" (Pillion, Vernick, & Shapiro, 2011, p.1) The severity of OI, however, depends on the kind of gene defect (Pillion et al., 2011, p.1).

OI is considered an autosomal dominant disease - meaning if an individual has one copy of the affected or abnormal gene, they will have the disease [osteogenesis imperfecta]. In most cases, OI is inherited from a parent; however, there are some new cases that are the result of new genetic mutations. To paint a more clear picture of the frequency with which this disease is transferred, a person with OI has a 50% chance of passing on the affected gene and the disease to their children (Osteogenesis imperfecta, 2011).

SYMPTOMS

From the Ostegenesis Imperfecta Foundation (Osteogenesis Imperfecta Foundation, Inc., 2012):

**CAPTIONED PSA**

**SYMPTOMS**

Classic symptoms of OI include:

- Blue tint to the sclerae - the whites of the eyes. This abnormality results from altered light reflectance in the presence of abnormal scleral collagen (Pillion et al., 2011, p. 2).
- Multiple bone fractures
- Early hearing loss (deafness) - Disordered type 1 collagen in the ear involves each of the auditory structures - the hard and soft tissue, leading to early-onset hearing loss (Pillion et al., 2011, p. 2).
- Short stature
- Pes planus (flat feet)
- Increased joint mobility (hypermobility)
- Fractures following relatively minor injury
- Dentinogenesis imperfecta - a disorder of tooth development which can cause tooth discoloration (blue-grey or yellow-brown color) and translucent. It is the result of abnormal collagen in dental pulp which causes enamel breakage (Pillion et al., 2011; Genetics Home Reference, 2013).

![Figure 1. Photograph of children with Osteogenesis Imperfecta. This figure illustrates the different faces of children with OI.](image)

More severe types of OI can display themselves symptomatically with the following skeletal abnormalities:

- Bowed legs and arms (Osteogenesis imperfecta, 2011).
- Kyphosis - exaggerated outward curvature of the thoracic (relating to the thorax) region of the spine resulting in a rounded upper back (Osteogenesis imperfecta, 2011; Kyphosis, 2013).
- Scoliosis - "a lateral curvature of the spine" (Osteogenesis imperfecta, 2011; Scoliosis, 2013).

PROGNOSIS

There are four main types of OI and a patient's outlook depends on which type they have.
Type I (aka, mild OI) - the most common form. Persons with this type can expect to have a normal lifespan.
Type II - a severe form; usually leading to death in during the first year of life.
Type III - another severe form. Persons with type III are projected to have frequent fractures beginning early on in their life and may experience severe bone deformities. A significant number of patients become wheelchair bound and typically have an abridged life expectancy.
Type IV - a moderately severe form of OI and most similar to type I. Individuals with type IV usually require braces or crutches for ambulation (walking). Life expectancy is projected to be normal or close to normal.

"There are other types of OI, but they occur very infrequently and most are considered subtypes of the moderately severe form (type IV)" (Osteogenesis imperfecta, 2011).

COMPLICATIONS

The complications that are manifested with osteogenesis imperfecta weigh largely on the type of OI an individual has. However, most problems that emerge are directly related to weak bones and multiple fractures.

According to the osteogenesis imperfecta entry in the A.D.A.M. Medical encyclopedia (2011), some complications of OI may include:

- "Hearing loss (common in type I and type III)
- "Heart failure (type II)
- "Respiratory problems and pneumonias due to chest wall deformities
- "Spinal cord or brain stem problems
- "Permanent deformity" (Osteogenesis imperfecta, 2011).

PREVENTION

If there is personal/family history with OI, genetic counseling is recommended for couples considering pregnancy (Osteogenesis imperfecta, 2011). The role of a genetic counselor is to work with and a member of a health care team with the goal of providing support to families who themselves or whose members have birth defects or genetic disorder or who may be at risk for such.

The following is the information for the genetic counselors near and around the 14623 zip code:

<table>
<thead>
<tr>
<th>Genetic Counselor Company</th>
<th>City, State/Province</th>
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This information is pertinent to parents who children may have genetics which predispose their offspring to Osteogenesis Imperfecta or other related orthopedic impairments. It is consoling to know that if these services are needed by individuals in the Greater Rochester, NY areas, they are available.

DEAFNESS & OSTEOGENESIS IMPERFECTA
Hearing loss, as mentioned above can occur in Type 1 and type III and rarely in type IV of Osteogenesis Imperfecta. Hearing loss is most commonly reported in type I OI, but is a significant clinical, if not constant, feature in patients suffering from OI. From national surveys, the rate of hearing loss among patients with OI is much higher than the rate among the normal population (Approximately 12% of Americans have significant hearing loss). This means that the rate of hearing loss among patients with OI is roughly 34%-45.9% higher than the rate found amongst Americans without OI as a collective group. Patients with OI therefore suffer disproportionately from hearing loss than those without. The prevalence of hearing loss among patients with OI ranges from 46% to 57.9% (Pillion et al., 2011, p.2).

Conductive hearing loss typically is reported as the first sign of deafness in OI. Conductive hearing loss has frequently reported in younger patients with OI, while sensorineural hearing loss has been found more in older patients with OI (Pillion et al., 2011, p.3). Given this information, it is recommended that children with OI receive a formal audiological evaluation before they begin school. Testing is recommended to begin around the age of 3-4 and should be conducted again on a three-year cycle. If a student with OI demonstrates speech delay, articulation trouble, chronic ear infections, or whose parents have a suspicion of hearing, a formal audiological assessment should be conducted without consideration of the student's age. If borderline hearing is detected, yearly audiological screenings are recommended (Osteogenesis Imperfecta Foundation, Inc., 2005, p. 2).

EDUCATIONAL IMPACT

School-aged children with OI have a wide range of abilities, limitations and needs. This is due, in part, to the fact that the disease manifest itself symptomatically very differently. Some students with OI may utilize a wheelchair, crutches, walkers, while others may be able to walk independently (without aid) (Osteogenesis Imperfecta Foundation, 1998, p. 1). Given the differences in the individual experience of OI, it follows that educational impact because of OI will also vary on an individual basis.

INCLUSION OF STUDENTS WITH OI

It is recommended that teachers and educators meet prior to the beginning of the school to discuss any changes that need to be made in the student’s accommodations for the forthcoming school year.

ARCHITECTURAL BARRIERS

Physical barriers may present a challenge for students with limited mobility such as students with OI. Common barriers found on school campuses are as follows: "steps at school entrances or between floors; restrooms with narrow or heavy doors, high sinks, and/or stalls too narrow for a wheelchair; playgrounds that can only be reached via a flight of stairs or by walking up or down a hill; and hands-on work areas (such as in science labs or woodworking classes) that are inaccessible to a child who uses a wheelchair or is of short stature" (Osteogenesis Imperfecta Foundation, 1998, pp. 3-4).

MOBILITY ACCOMODATIONS

Students with OI can face mobility obstacles while in school. Those students that use wheelchairs may experience challenges while navigating crowded hallways and classrooms. And those who walk may have problems with stairs, walk slower than their peers, and/or may be at risk for falls while traversing their school campus. Some common mobility accommodations are (Osteogenesis Imperfecta Foundation, 1998, p. 4):

- Have the student tour the school, including the classrooms, restrooms, cafeteria, library, and the gym. This will reveal mobility barriers and allow the school to address the problems will in advance before school begins.
- Allow the student extra time to travel between classes. This may mean allowing the student to leave class several minutes early. (N.B. - it is important that allowing the student to leave early does not interfere with classroom instruction or the student's education).
- Provide the students with an elevator if the campus on which they are navigating is multilevel.
- Permit the student to choose a seat in the classroom that is easy to access.
- Provide an extra copy of books for the student to keep at home so that heavy loads of books do not have to be carried (Osteogenesis Imperfecta Foundation, 1998, p. 4).

ADVICE FOR PARENTS WITH INFANTS WITH OI

The following is a video (N.B. - this video does not have subtitles) chronically Angelo and Kim's experience of having a child with Osteogenesis imperfecta (Bobo & Grasta, 2012):

Handling Suggestions

- Movement should be slow, methodical, and gentle.
- Never pull, push, twist, apply pressure, bend or attempt to straighten arms or legs.
- Infants with OI should never be picked up by the armpits or around the ribcage because this can cause fractures.
- "The head and the trunk should be supported with one hand while the other supports the buttocks" (Osteogenesis Imperfecta Foundation, nd, pp.1-2).
- "Keep the fingers spread apart to provide a wider base of support and an even distribution of support pressure" (Osteogenesis Imperfecta Foundation, nd, pp.1-2).
- Apply support to the broadest possible area when diapering, feeding or dressing.
- When diapering the baby do not lift the baby by the ankles (this may result in fracturing). Slide the hand under the buttocks to lift the baby and replace the diaper.
- Infants that have fractures may be placed in casts/splints to reduce motion and to provide stabilization. These infants must not be put prone on the stomach due to the possibility of suffocation.
- Protecting the infant's arms, wrists, and fingers is critical when changing dressing or bedding.
- When dressing the infant, bring the garment over the limb; do not drag the limb (or extremities) through the garments because this pulling or twisting can cause fractures.
- Infants with OI need affection and touch from their care givers to develop appropriately (Osteogenesis Imperfecta Foundation, nd, pp.1-2).

Exercise and Physical Activity
• “A program of graduated physical activity is necessary during infancy” (Glorieux, 2007, p.1).
• Bath time is important because it allows children with OI to experience movement in a comfortable environment.
• “Carrying an infant with OI on a pillow is no longer recommended” (Glorieux, 2007, p.1).
• “Infants should be repositioned frequently during the day. Beneficial positions for an infant with OI include being held, carried, held on a caregiver’s shoulder, and side lying” (Glorieux, 2007, p.1).

Feeding

Feeding can be a intricate task for parents with a child that has OI. The most important thing to realize is that care must be taken as with all other activities done with the child with OI. Some infants with OI may be poor feeder because of weak sucking reflex. As a result, they may require small and frequent feedings. “The combination of small stature, feeding problems, and slow growth may be mistaken for failure to thrive” (Osteogenesis Imperfecta Foundation, nd, p.2).

Breast milk is an excellent source of nutrition for infants including those with OI. Some babies with OI, however, may not be able to breast feed because of their breathing difficulties (which may interfere with their ability to suck). Rapid respirations can predispose the infant with OI to aspiration - the accidental sucking in of fluid or food particles into the lungs). In such an instance, where an infants OI is so severe where breast feeding is not possible, the mother may opt to pump breast milk and feed the child from a bottle (Osteogenesis Imperfecta Foundation, nd, p.2).

Mothers have to exercise special care should while breast-feeding. “When feeding the infant, the mother should be especially careful to avoid having the baby positioned with arm behind the back or a leg pressed against the mother’s body in such a way as to put pressure on it at an abnormal angle” (Osteogenesis Imperfecta Foundation, nd, p.2).

Burping should be done gently so as to reduce the chance of fracture occurrence. It is recommended that the mother softly tap the infant, if needed with a padding covering the hand. “To pick up the infant for burping, lay the baby on his back while the caregiver bends over to pick up the infant. The caregiver's shoulder should very gently touch the baby at which point the infant is supported under the back and positioned on the shoulder as the care giver moves up and backwards. Gently rubbing the baby’s back while taking gentle steps may also be beneficial” (Osteogenesis Imperfecta Foundation, nd, p.2).

Bedding

Standard crib mattresses are most suitable for a baby with OI. Soft bedding and waterbeds should NEVER be used (Osteogenesis Imperfecta Foundation, nd, p.2).

Preventing Head Flattening

• Reduce pressure on the back of the head to prevent skull malformations. The following strategies are helpful:
  • Place gel pads under the infant’s head when they are on their back.
  • “Position the infant in a propped, side-lying position” (Glorieux, 2007, p. 1).
  • Frequently change the infant’s position throughout the course of the day.
  • “Carry the infant on your shoulder or in an approved sling carrier” (Glorieux, 2007, p. 1).
  • Avoid leaving the infant in a car seat for extended periods of time.
  • “Helmets have been used in some infants who have OI, but they are not universally recommended. In severely affected infants, the additional weight of a helmet may make the already challenging task of gaining head and neck control even more difficult” (Glorieux, 2007, p. 1).

Car Seats

Some preliminary information about car seats are that they are geared to the child's weight and their ability to sit up. All - not some - should be approved for safety and anchored correctly in the vehicle. Placing foam padding between the plastic shell and the padded cover is NOT safe; it will compress if a car accident were to occur. Should a softer surface be required, a much better option is to use a folded thin baby blanket. For children with OI, removable cotton slip covers are suggested because they are cooler and some children with OI sweat excessively. It is best practice to place the car seat in the back of the vehicle. In addition to this, many parents label the top edge of the car seat with their child's diagnosis, physician name and phone number, emergency contact numbers, and HANDLE WITH CARE instructions in the event of an accident (Osteogenesis Imperfecta Foundation, nd, p.3).

N.B. - “Infant car seats are designed for children under 20 pounds in weight. Features to look for include a well-padded harness and a head hugger support pillow. This type of U-shaped pillow is commercially available and is used to position the baby's head at midline. Small rolls or towels or other padding can be added to hold the child's hips in line. Some infant car seats can be fitted with a contoured foam insert” (Osteogenesis Imperfecta Foundation, nd, p.3)

References


