Osteogenesis Imperfecta (OI)

Patient Information Series – What you should know, what you should ask.

What is Osteogenesis Imperfecta (OI)?

OI, also known as “brittle bone disease,” is a group of rare connective tissue disorders that leads to fragile bones. The condition can vary greatly among affected individuals and is associated with frequent bone fractures, bone deformities, stunted growth, and in severe cases, can lead to death in utero or shortly after birth. The different types of OI are classified based on the severity of the disease.

How does OI happen?

OI is often the result of a genetic mutation that impacts the structure or function of type 1 collagen, a critical component of bone. There have been many different genes associated with OI. It is a rare condition that can happen about 1 in 10,000 to 20,000 births. While sometimes the mutations are passed along in certain families, many of the mutations are a result of new genetic accidents and not due to factors that can be controlled.

When is it detected?

OI may be detected as early as 14 weeks gestation but most is most commonly first noted at the time of routine anatomy fetal ultrasound at about 20 weeks. The most common finding that is first noticed is a short thigh bone as well as bowing of the bones, demineralized bones, and even fractures. Unfortunately, OI that is detected while the baby is still in the uterus tends the more severe forms of the condition. Mild forms of OI may not be detected until childhood or later in life.

Should I have more tests done?

Many women will choose to have more tests done to know more about the condition of the baby. The tests available depend on where you are. Tests to ask about include:

- **Fetal ultrasound** - typically done in all pregnancies and is part of routine prenatal care to look at the full anatomy of the fetus. This uses “echo” of ultrasound waves to identify structures without posing any risks of radiation to the mother or fetus.
- **Amniocentesis** – A needle is inserted into the amniotic sac to remove a small amount of the fluid surrounding the fetus. This fluid contains the genetic material for the fetus and may be tested to see if there are genetic mutations associated with OI.
- If available, an **MRI scan** can sometimes be done to provide information on the condition of the baby. This scan uses strong magnetic fields and radio waves to create detailed images of the inside of the body. It does not use radiation.

What are the things to watch for during the pregnancy?

OI diagnosed during the time of pregnancy is typically associated with the more severe forms of the disease. There may be impaired overall growth of the fetus, as well as underdevelopment of the lungs. You will continue to have frequent fetal ultrasounds throughout your pregnancy to determine the severity of the disease. Based on the baby's growth, the number of fractures, and other findings, there may be a possibility that OI is lethal for the fetus.

What does it mean for my baby after it is born?

The most severe form of OI is lethal during pregnancy or shortly after delivery. The second most severe form may be associated with severe disability of the surviving baby. You will meet with high-risk pregnancy specialists and baby doctors to better discuss your individual risks and develop the best plan for your baby, you, and your family.

Will it happen again?

The risk of OI happening again depends on whether or not it is associated with a familial gene. Genetic consultation may assist in determining the risk of occurring again in the future and is recommended for those with family history of OI or OI diagnosis in pregnancy.

What other questions should I ask?

- Does this look like severe OI?
- Is my baby likely to survive?
- How is my baby likely to fare after it is born?
- How big do the lungs seem to be?
- How often will I have ultrasound examinations done?
- Where should I deliver?
- Where will the baby receive the best care after it is born?
- Can I meet in advance the team of doctors that will be assisting my baby when it is born?

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