

Skeletal Dysplasias

What are skeletal dysplasias?

Skeletal dysplasias are a class of conditions that encompass more than 400 disorders and affect approximately 1 in every 5,000 babies. Individuals with skeletal dysplasias can have a variety of findings which may include shortened arms and/or legs, smaller chest size, and an abnormal head size and shape. Skeletal dysplasias can be inherited, can occur due to random genetic changes, or can be due to disruptions in normal skeletal development.

Symptoms of skeletal dysplasia vary depending on the specific disorder your baby has but can include:

- » Short arms and legs
- » Fragile bones that may fracture
- » Duplication of fingers or toes
- » Missing limbs
- » Joint issues
- » Developmental delays
- » Bowed legs or arms
- » Short, broad hands and feet
- » Club feet
- » Missing ribs
- » A curvature of the spine known as “scoliosis”
- » Cognitive impairment

How are skeletal dysplasias diagnosed?

Skeletal dysplasias are often suspected before birth. Bone abnormalities and other differences listed above may be visible on a prenatal ultrasound. Once a skeletal dysplasia is suspected, genetic testing is typically offered to try to determine which exact condition a baby may have. In some cases, a low-dose fetal CT scan or X-ray may also be recommended to help make a definitive diagnosis.

Because of the overlapping features between various skeletal dysplasias, it can often be challenging to make a specific diagnosis prenatally. In some cases, a combination of ultrasound evaluation, genetic testing, and other assessments can allow for such a diagnosis to be made before a baby is born. In other cases, the medical team may need to evaluate a baby after birth to make a specific diagnosis.

It is important that the medical team caring for a baby suspected to have a skeletal dysplasia include experts in genetics, neonatology, orthopedics and maternal-fetal medicine. The specialists at the SSM Health Cardinal Glennon St. Louis Fetal Care Institute will monitor the baby throughout the pregnancy and assist families with planning for delivery and arranging for postnatal care.

A Cardinal Glennon St. Louis Fetal Care Institute nurse is available 24 hours a day, seven days a week to discuss referrals with physicians and potential families by calling 314-268-4037, option 2.

Phone 314-268-4037, option 2
Toll-free 1-877-SSM-FETL (776-3385)
Web stlouisfetalcare.com
Email fetalcare@ssmhealth.com
Facebook facebook.com/fetaldocs

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How will a skeletal dysplasia affect my baby after delivery?

Because of the complex nature of skeletal dysplasias, babies often face medical challenges during delivery and in the newborn period. This is why it is important for them to be delivered at a hospital that can meet their unique medical needs. Every baby's situation is different, and the Cardinal Glennon St. Louis Fetal Care Institute works with each family to prepare for their baby's delivery and care after birth.

The delivery and care plan can include medical interventions or be restricted to comfort measures, depending on the baby's diagnosis and prognosis and the desires of the family.

How will a skeletal dysplasia affect my baby into childhood?

Children with skeletal dysplasias benefit from the care of a multi-disciplinary medical team, which may include providers from:

- » Genetics
- » Orthopedic Surgery
- » Neurosurgery
- » Ophthalmology
- » Occupational Therapy
- » Neonatology
- » Endocrinology
- » Otolaryngology (ENT)
- » Physical Therapy

The type of skeletal dysplasia and the associated abnormalities a baby has will affect a child's long-term prognosis. Children who do not have life-threatening or severe forms of skeletal dysplasia can usually lead relatively typical lives.

We understand that Skeletal Dysplasias can be a scary diagnosis. That's why we're available to help 24 hours a day, 7 days a week. For more information or to schedule an appointment, call us at 314-268-4037 or toll free at 877-SSM-FETL (877-776-3385).

While we can't change the diagnosis, we can provide you with expert care and support, helping your baby get the most out of treatment and life.
