Information for you

Before, during and after pregnancy
For mothers and infants with a skeletal dysplasia diagnosis

This is a quick summary of our medical article, with important questions and answers about your pregnancy. To see the full article, visit http://www.skeletaldysplasia.org/physician-resources/.*

Prenatal diagnosis of a skeletal dysplasia

QUESTIONS	ANSWERS
If my partner and I have a skeletal dysplasia diagnosis, can we pass one or both of our conditions to our fetus?	The risk of inheriting a skeletal dysplasia from a parent varies according to each parent's diagnosis. *Before* or *early in* pregnancy, a pregnancy specialist (e.g., Maternal-Fetal-Medicine physician [MFM]) and a genetic counselor can explain your specific risk and give you testing options.
Do you know what to look for to make a fetal dysplasia diagnosis during pregnancy?	An MFM physician is familiar with specific measurements and physical features as shown by ultrasound to recognize a skeletal dysplasia in a fetus. If fetal ultrasound results are abnormal, you should consult an MFM physician.
If prenatal ultrasound shows a possible skeletal dysplasia, must I get prenatal genetic testing to know for sure?	It's your choice. Many options are available for testing during pregnancy, or the child can be tested after birth. See below for guidelines for healthcare providers preparing for the birth of an infant with a skeletal dysplasia.
Is it possible to determine if a fetus with a skeletal dysplasia will survive after delivery?	There are important measurements and physical features seen in ultrasound that, when combined, can greatly increase the accuracy of predicting how severe the dysplasia-related complications may be.

Guidance during pregnancy:

- Preconception (before pregnancy) genetic counseling is recommended for parents who have skeletal dysplasia to help better understand the risks of passing it on.
- There are over 250 different skeletal dysplasia diagnoses with all modes of inheritance, and genetic testing is available for nearly all of them.
- Fetal DNA obtained for genetic testing should be retained for further evaluation until a correct diagnosis is reached.
- Prenatal genomic test results need to be ordered and interpreted by a qualified expert and must be correlated with clinical findings on the fetus.
- Ultrasound is the main means of imaging to diagnose suspected skeletal dysplasias during pregnancy.
- There are several specific measurements and assessments of a fetus during prenatal ultrasound that strongly suggest the likelihood of a skeletal dysplasia and how severe it will be.
- 3D ultrasound, MRI and dose-minimizing CT may be used during pregnancy to assess for signs of skeletal dysplasia.
- If a pregnancy is terminated due to suspected skeletal dysplasia, it is recommended that the fetus be evaluated afterward; this may direct further genetic testing, which may give helpful insights for future pregnancies.

Delivery of a fetus with a skeletal dysplasia

QUESTIONS	ANSWERS
Who will be in the delivery room?	Healthcare providers for the mother and the baby are needed in the operating room (where the cesarean section will occur). A tertiary specialty care hospital has specialists on site to provide immediate care, if needed.
How does the delivery hospital manage critically ill infants at delivery?	Not all delivery sites have intensive care services available for mother and baby. The baby may need to be transferred to a different hospital, but the transfer may be complicated and delayed, also making it difficult to see your baby.

Guidance during pregnancy and delivery of a fetus with a skeletal dysplasia:

- Diagnosis of a suspected fetal skeletal dysplasia is important for pregnancy management and counseling.
- Pregnancies in which there is a suspected fetal skeletal dysplasia should be referred to appropriate centers with high-level ultrasound expertise and Maternal-Fetal-Medicine (MFM) experts to diagnose the fetus and manage the pregnancy and delivery.
- Counseling of a couple with a suspected skeletal dysplasia should include discussion of probable medical needs as well as possible new treatments and therapies.
- For a fetus with osteogenesis imperfecta (a brittle bone disease) specifically, cesarean section delivery does not reduce fractures.
- For all fetuses with a skeletal dysplasia, use of instruments such as forceps should be avoided during delivery due to increased risk of complications for the intracranial bones (skull and face) and the cervical spine (neck and upper part of the spine).
- All fetuses with suspected skeletal dysplasia should be delivered in a tertiary center with appropriate pediatric care and neonatal intensive care unit facilities.

Pregnancy and delivery for a woman with a skeletal dysplasia

QUESTIONS	ANSWERS
What special concerns do you have about me during pregnancy due to my skeletal dysplasia?	A pregnancy healthcare provider who is prepared to care for a woman with a skeletal dysplasia will evaluate your lungs, heart and neurologic system to determine if you will safely be able to carry a fetus through pregnancy and delivery. Ideally this evaluation occurs *before* or *early in* pregnancy.
What special concerns do you have about me during delivery due to my skeletal dysplasia?	A pregnancy healthcare provider managing a woman with a skeletal dysplasia will be prepared for a cesarean section delivery. This includes involving early in your care the anesthesiologist who will provide anesthesia for your delivery (whether general or regional anesthesia).
Are there specialists available at my planned delivery hospital to manage complications that may arise for me and my baby?	Ask what type of specialists are available to care for you and your baby after delivery, including when delivery occurs at night or on the weekends.
Will I have a premature delivery due to my skeletal dysplasia?	No, not necessarily. You will need to have more frequent visits with your pregnancy healthcare provider to ensure your lung, heart and neurologic function are not being compromised by the size of the fetus and/or compression on your organs. If there is concern in any of these areas, an earlier delivery may be needed.

Guidance during pregnancy and delivery for women with skeletal dysplasia:

- Preconception genetic counseling is recommended for parents who have skeletal dysplasia to help better understand the risks of passing it on.
- Preconception (or as early as possible in pregnancy) medical evaluation of a woman with a skeletal dysplasia is recommended to identify medical issues that may influence the safety of her pregnancy, the delivery method and the anesthesia for delivery.
    - The mother's evaluation must include her airway, heart, lungs, spinal cord and skeleton.
    - Women with a short trunk skeletal dysplasia should be identified, as they are at higher risk of heart and lung issues in pregnancy.
- Details of the delivery should be discussed early in pregnancy, including *location*, mode of *delivery* (vaginal or cesarean section), *anesthesia* and *perinatal* care (from the time you get pregnant until up to a year after delivery) to optimize maternal and fetal outcomes.
    - Newborns with skeletal dysplasia may require immediate specialized medical management.
    - Vaginal delivery is not possible for most women with skeletal dysplasia women; cesarean delivery is recommended.
    - The body structure of women with skeletal dysplasia increases the risk of general and regional anesthesia during delivery; advanced planning is needed.
- Standard recommendations for pregnancy weight gain are not applicable to women with skeletal dysplasia.
- Women with skeletal dysplasia are not at a higher risk for preterm labor (24–37 weeks of gestation), but may need early delivery due to lung function, neurologic problems or other issues.

Newborns with skeletal dysplasia

QUESTIONS	ANSWERS
After delivery, how will my baby be evaluated?	Physical examination, measurements of length, weight and head circumference; X-rays; possibly ophthalmology (eye) exam, hearing test, echocardiogram (to assess the heart structure); genetic testing. Ask how and when these evaluations will be performed and make sure that a specialist familiar with skeletal dysplasias interprets them.
Who will evaluate my baby for a skeletal dysplasia diagnosis?	Pediatrician, neonatologist, clinical geneticist, nurse practitioner, orthopedists, neurosurgeon and/or other specialist(s); via telemedicine or in-person. Ideally, a healthcare provider will have some experience with skeletal dysplasias or consult with a specialist who does. Ask at your birthing hospital about their experience in recognizing and managing skeletal dysplasias.
How does the hospital arrange for a specialist to examine my baby, if needed?	In most large hospitals (particularly teaching hospitals), specialists are available to evaluate your baby immediately. In smaller hospitals, they may not be available. The local hospital personnel may consult specialists by telemedicine or request evaluation before discharge. Specialists may evaluate your baby as an outpatient. This varies widely; ask about the practice at your birthing hospital. If you and the healthcare providers are not comfortable caring for an infant with a skeletal dysplasia, seek out a different birthing hospital.
Who will manage medical issues that may occur related to the dysplasia diagnosis?	This also varies among hospitals based on availability of specialists. Neonatologists or neonatal nurse practitioners manage common issues related to prematurity, breathing difficulties, infections and other newborn issues. Dysplasia-specific problems in the newborn period may require a pulmonologist, neurosurgeon or other specialist.
Will my baby be evaluated while in the hospital or after discharge at a clinic visit?	Assessment may occur in the delivery room shortly after birth; in the nursery in the first day or two of life; or in an outpatient clinic after discharge. If a diagnosis hasn't been confirmed prenatally, a healthcare provider should examine your baby as soon as possible after delivery. Different dysplasias have different potential complications. X-rays and genetic testing should be readily available to work toward a diagnosis.

Guidance for a newborn with a skeletal dysplasia:

- All newborns with suspected skeletal dysplasia should be evaluated as soon as is practical after delivery.
- If a specific diagnosis *is not* known at birth, the newborn should be referred to a specialist center for diagnostic assessment and management.
- If a skeletal dysplasia diagnosis *is* known at birth, then appropriate management specific to that dysplasia should be provided.
- X-rays are helpful to reach a diagnosis of a skeletal dysplasia.
- Genetic testing is available to diagnose nearly all short stature skeletal dysplasias.
- All fetuses with prenatal suspected skeletal dysplasia should be delivered in a tertiary specialty center with appropriate pediatric care and neonatal intensive care unit facilities.

*** Citation:** Savarirayan R, Rossiter JP, Hoover-Fong JE, Irving M, Bompadre V, Goldberg MJ, Bober MB, Cho TJ, Kamps SE, Mackenzie WG, Raggio C, Spencer SS, White KK; Skeletal Dysplasia Management Consortium. Best practice guidelines regarding prenatal evaluation and delivery of patients with skeletal dysplasia. Am J Obstet Gynecol. 2018 Dec; 219(6):545-562. PMID: 30048634.