



RAPID ACCESS for HEALTHCARE PROVIDERS

For mothers and infants with a skeletal dysplasia diagnosis

Derived from *Best practice guidelines regarding prenatal evaluation and delivery of patients with skeletal dysplasia.*

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. Am J Obstet Gynecol. 2018 Dec; 219(6):545-562. PMID: 30048634.

1. Pregnancy and delivery in women with skeletal dysplasia

1. Preconception genetic counseling is recommended for individuals and/or partners who have skeletal dysplasia or are at increased chance of having a fetus with skeletal dysplasia.
2. Preconception medical evaluation is recommended to consider factors that may impact safety of pregnancy, mode of delivery, and anesthetic management (e.g., airway, cardiopulmonary status, and neuro-axial and musculoskeletal structure and function).
3. Standard recommendations for weight gain during pregnancy do not apply to women with skeletal dysplasia.
4. Women with skeletal dysplasia are not at a higher risk for preterm labor (24-37 weeks of gestation), but may need delivery prior to term.
5. Careful attention to fluid management in women with skeletal dysplasia is required to avoid fluid overload in the peripartum period.
6. The standard management of preterm labor may need to be modified for women with skeletal dysplasia.
7. Anatomical differences in women with skeletal dysplasia increase the risk of general and regional anesthesia during pregnancy and delivery, and require advanced planning when possible.
8. Management of delivery should be discussed early in pregnancy, including location, mode of delivery, anesthetic options, and perinatal care to optimize maternal and fetal outcomes.
9. Women with skeletal dysplasia characterized by a short trunk should be identified, as they are at higher risk during pregnancy for cardiopulmonary complications, maternal complications, and preterm delivery.
10. Vigilance is required for pregnant women with skeletal dysplasia, as increasing fundal height may adversely affect maternal cardiopulmonary and musculoskeletal status.
11. Pelvic anatomy in most women with skeletal dysplasia precludes vaginal delivery, and cesarean section is recommended.
12. A woman with skeletal dysplasia can have a cesarean section with a Pfannensteil skin incision and low transverse uterine incision.

2. Pregnancy and delivery of newborns with suspected skeletal dysplasia

1. Newborns with skeletal dysplasia may require immediate specialized medical management.
2. Pregnancies in which there is a suspected fetal skeletal dysplasia should be referred to appropriate high-risk maternal fetal medicine/perinatal medicine specialists for management or recommendations for management.
3. Use of instrumentation during delivery should be avoided if possible when fetal skeletal dysplasia is suspected, due to the increased risk of intracranial and cervical spine complications.
4. Caesarean section does not decrease the incidence of fractures in children with prenatal diagnosis of osteogenesis imperfecta.

3. Prenatal diagnosis of skeletal dysplasia

1. Prenatal diagnosis of a suspected fetal skeletal dysplasia is important for pregnancy management and counseling.
2. Pregnancies in which there is a suspected fetal skeletal dysplasia should be referred to appropriate centers with high-level ultrasound expertise and expert evaluation.
3. The finding of a femur length 5th percentile for gestational age during the 18-22-week ultrasound warrants further evaluation.
4. Fetal DNA obtained for genetic testing should be retained for further evaluation until a correct diagnosis is established.
5. Post-mortem evaluation is recommended for fetuses from pregnancies terminated due to suspected skeletal dysplasia.
6. As the modes of inheritance vary among skeletal dysplasias, establishing the correct diagnosis and giving individualized preconception genetic counseling are important.
7. Prenatal genomic tests need to be ordered and interpreted by a qualified expert and must be correlated with clinical findings.
8. Increased nuchal translucency at 10-14 weeks of gestation can be a sign of a severe skeletal dysplasia.
9. The most likely time in pregnancy to detect features of a skeletal dysplasia is 18-20 weeks of gestation.
10. Most of the severe skeletal dysplasias are detected at the routine ultrasound performed at 18-20 weeks of gestation.
11. Non-lethal skeletal dysplasia may not be evident by ultrasound until 28 weeks of gestation or after.
12. Some types of skeletal dysplasia may not be evident until birth.
13. Ultrasonography remains the main imaging modality to diagnose suspected prenatal skeletal dysplasias.
14. Plain radiographs do not help refine the diagnosis of suspected skeletal dysplasias.
15. Low-dose CT scanning can help refine the diagnosis of suspected skeletal dysplasias.
16. 3D ultrasonography can help refine the diagnosis of suspected prenatal skeletal dysplasias.
17. Prenatal MRI scanning can help refine the diagnosis of suspected skeletal dysplasias.
18. Determining predictive factors of lethality is important in the assessment of a prenatal suspected skeletal dysplasia.
19. Key predictors of lethality at the 18-20 weeks of gestation ultrasound are:
 - Chest to abdomen ratio <0.6
 - Femur length to abdominal circumference <0.16
 - Femur length to biparietal diameter
 - Micromelia 3 SD below the mean
 - Hydrops
 - Severely decreased mineralization of the axial skeleton

4. Considerations for newborns with skeletal dysplasia

1. All newborns with suspected skeletal dysplasia should be evaluated as soon as practical after delivery, regardless of previous diagnosis or prognosis.
2. If fetal skeletal dysplasia is suspected but a specific diagnosis is not known at birth, referral to a specialist center for diagnostic assessment and management is recommended.
3. If a specific skeletal dysplasia diagnosis is known at birth, appropriate management should be instituted.
4. Postnatal skeletal surveys are helpful in the diagnosis of skeletal dysplasia.