



GENERAL INFORMATION ABOUT SKELETAL DYSPLASIAS and WHO WROTE THESE CHECKLISTS?

What does “skeletal dysplasias” mean?

“Skeletal dysplasias” is a medical term that includes many different genetic conditions affecting the growth and development of the skeleton. A woman with a short-stature skeletal dysplasia may have an increased risk of complications during a pregnancy because of her diagnosis. A fetus with a suspected skeletal dysplasia diagnosis—regardless of the mother’s stature—also has increased risk of complications at delivery.

Short-stature skeletal dysplasia diagnoses are relatively rare. It is likely your healthcare provider will have limited experience taking care of pregnant women or fetuses with a short-stature dysplasia. It is essential for them to be aware of the risks associated with short-stature dysplasias to make sure the appropriate resources and facilities are available to you throughout pregnancy and delivery.

Who wrote these checklists?

The Skeletal Dysplasia Management Consortium, an international group of physicians with particular experience caring for people with skeletal dysplasias, has published best-practice guidelines to help manage several medical issues specific to this patient population. These publications cover prenatal/pregnancy care, perioperative (during a surgery) management, the spine, craniofacial (bones of the skull and face) issues, the management of the foramen magnum (the large hole at the bottom of the skull that connects with the spine) in achondroplasia and type 2 collagen disorders (e.g. SEDC, Kneist). From these publications, we have created shorter Healthcare Provider Checklists and separate, easy-to-use Patient Checklists for you.

We hope you will use the Patient Checklists to guide your discussions with your healthcare professionals. Please share the original articles and the Healthcare Provider Checklists with your healthcare professionals. All the publications and Checklists can be accessed at the Skeletal Dysplasia Management Consortium website: <http://www.skeletaldysplasia.org>