GTPIM102

FGFR3 Gene Related Skeletal Dysplasias

Patient Information Material

What are Skeletal dysplasias?

Skeletal dysplasias are a heterogeneous group of >200 disorders characterized by abnormalities of cartilage and bone growth, resulting in abnormal shape and size of the skeleton and disproportion of the long bones spine, and head (including dwarfism), duplication of fingers or toes etc. Skeletal dysplasia occurs in approximately one in every 4,000 births.

How are skeletal dysplasias inherited?

Skeletal dysplasias usually result from mutated genes. Some are sporadic (not inherited) or caused due to chromosomal defects. But many are inherited in autosomal dominant (with one copy of mutation), recessive (with two copies of mutation) or X-linked (with a mutation on chromosome X) manner.

What are FGFR3 gene related skeletal dysplasias?

A large spectrum of skeletal dysplasias are caused due to FGFR3 gene mutations. The gene plays an important role in cell growth, division, formation of blood vessels, wound healing, and embryo development and inherits in autosomal dominant manner. Some of the common FGFR3 related disorders are:

- Achondroplasia
- Hypochondroplasia
- Cruzon syndrome
- Craniosynostosis
- Muenke Syndrome
- Thanatropic dysplasia

What is Achondroplasia?

Achondroplasia is a form of short-limbed dwarfism with short arms and legs, restricted elbow motion, enlarged head (macrocephaly) with a prominent forehead. People with achondroplasia are generally of normal intelligence.

What is Hypochondroplasia?

Hypochondroplasia is a form of short-limbed dwarfism with short arms and legs, restricted elbow motion, enlarged head (macrocephaly) with a prominent forehead, lordosis and bowed legs. These signs are generally less pronounced than those seen with achondroplasia and may not be noticeable until early or middle childhood.

applying genetics to life

What is Cruzon syndrome?
Crouzon syndrome is a genetic disorder characterized by the premature fusion of skull bones, bulging eyes, vision problems.

What is Thanatropic dysplasia?
Thanatophoric dysplasia is a severe skeletal disorder characterized by extremely short limbs and folds of extra (redundant) skin on the arms and legs. Other features of this condition include narrow chest, short ribs, underdeveloped lungs, and an enlarged head.

What is Craniosynostosis?
Absence of "soft spot" (fontanelle) on the skull, presence of raised hard ridge, affected sutures, unusual head shape are characteristics of Craniosynostosis.

How are Skeletal dysplasias diagnosed confirmed in genetic lab?
A blood DNA test called “FGFR3 Skeletal Dysplasia panel” diagnoses the above skeletal dysplasias in affected individuals.

How are skeletal dysplasias diagnosed in pregnancy?
Skeletal dysplasia may be diagnosed by ultrasound (sonogram) examination around 20th week of gestation prior to birth. However, the exact type of dysplasia is difficult to diagnose until after birth. Prenatal genetic testing plays an important role in diagnosis. Amniocentesis and chorionic villus sampling procedures can help rule out Skeletal dysplasias in the fetus.

How are children with Skeletal dysplasia's treated?
Approximately 25-50% of affected fetuses with a skeletal dysplasia result in stillborn or neonatal demise. Although there is no cure for genetic skeletal dysplasias, dysplasias with less severe medical problems can be successfully managed with a team work of radiologist, geneticist, orthosurgeon and pediatrician. Surgery and bracing of the bones can greatly improve the quality of life for these children.

How can a Genetic Counseling help?
A genetic counselor is a supportive professional who works with individuals and families to review genetic testing and diagnoses, review inheritance patterns, help identify possible risk in the family, discuss the symptoms and features of genetic disorders and suggest appropriate reproductive options. Due to the complexity of skeletal dysplasia diagnosis and inheritance, counseling is essential in the interpretation of results and for providing accurate information to families.

GeneTech is a pioneer in medical genetics testing in the country and offers the most comprehensive testing and screening services for genetic disorders in India

GeneTech, # 6-3-1113/4, Behind B S Makhta, Greelands, Begumpet, Hyderabad, 500 016.
Tel: 040-23400932, 23406178 Website: www.genetech.co.in **Customer Care: 98480 41127**