SKELETAL DYSPLASIA AND HOMOEOPATHY

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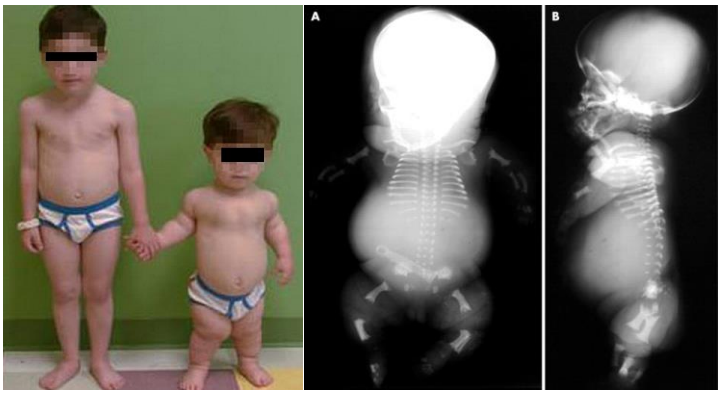
DEFINITION

Skeletal dysplasia is a group of conditions that cause abnormal bone and cartilage development resulting in short stature, unusual limb proportions, and several other symptoms and lead to various types of dwarfism (Psora/ Syphilis). The mutation causing skeletal dysplasia results from a single autosomal dominant gene, although the gene may have incomplete penetrance (Syphilis).



ETYMOLOGY

Chondro = cartilage; Achondro = without cartilage; Dysplasia = deformed; Osteo = bone

CAUSES

Skeletal dysplasia is a genetic condition and caused by a defect in a specific gene, called as genetic mutation which are passed down from parents to children. These mutations can prevent child's bones from growing normally. While skeletal dysplasia runs in families, it can be passed to the child even if parents do not have a known family history of it.

TYPES

Types of skeletal dysplasia are generally categorized by which parts of the skeleton are involved. The main types of skeletal dysplasia are achondrogenesis, achondradysplasia, chondrodysplasia, osteodysplasia and osteochondrodysplasia.

ACHONDROGENESIS

It is a disorder that causes failure in development of cartilage rendering the child to develop short limbs and a small body (Psora).

ACHONDROPLASIA

Achondroplasia or dyschondroplasia is a form of short-limbed dwarfism (Psora/ Syphilis). Achondroplasia is a common cause of dwarfism. It occurs as a sporadic mutation mainly in those with advanced paternal age or it may be inherited as an autosomal dominant genetic disorder (Syphilis). Achondroplastic dwarfs have short stature, with an average adult height of 131 cm for males and 123 cm for females. Most cases are due to a mutation in the fibroblast growth factor receptor 3 gene FGFR3. Unlike some forms of dwarfism, achondroplastic dwarfism is not curable with growth hormone. Normal parents may give birth to an achondroplastic baby due to mutation in this gene. A person with achondroplasia thus has a 50% chance of passing dwarfism to each of their offspring.

In normal development FGFR3 has a negative regulatory effect on bone growth. In achondroplasia, the mutated form of the receptor is constitutively active and this causes development of severely shortened bones. The effect is genetically dominant, with one mutant copy of the FGFR3 gene which is enough to cause achondroplasia.

If there are two copies of the mutant gene, it is invariably fatal i.e. recessive lethal before or shortly after birth, therefore known as a lethal allele. Elderly males are more prone to this mutation during spermatogenesis while females have some regulatory mechanism that prevents the mutation during oogenesis. There are two other syndromes with a genetic basis similar to achondroplasia-

HYPOCHONDROPLASIA

It is a condition that affects the conversion of cartilage into bone in the child's body and results in short arms and legs, as well as hands and feet that are short and broad (Psora).

THANATOPHORIC DYSPLASIA

It is a condition that causes the child to develop extremely short limbs, extra folds of skin on their arms and legs, and underdeveloped lungs (Psora/ Syphilis).

CHONDRODYSPLASIA

It is a hereditary skeletal disorder characterized by the formation of exostoses at the epiphyses and resulting in arrested development and deformity (Psora/ Syphilis/ Sycosis). It may be of following types-

CAMPOMELIC DYSPLASIA

It is an often fatal condition in newborns that causes dangerous bowing of the long bones in the child's legs and often their arms as well (Psora/ Syphilis/ Sycosis).

RHIZOMELIC CHONDRODYSPLASIA PUNCTATA

It is a rare, developmental brain disorder characterized by systemic shortening of the proximal bones (i.e. rhizomelia), seizures, recurrent respiratory tract infections, and congenital cataracts (Psora/ Syphilis).

TYPE 1 (RCDP1)

It is associated with PEX7 mutations. These are peroxisome biogenesis disorders where proper assembly of peroxisomes is impaired.

It is associated with significantly delayed development and severe intellectual disability (Psora/ Syphilis).

TYPE 2 (RCDP2)

It is associated with DHAPAT mutations (Syphilis).

TYPE 3 (RCDP3)

It is associated with AGPS mutations (Syphilis).

METAPHYSEAL CHONDRODYSPLASIA, SCHMID TYPE

It is also called as Japanese type spondylometaphyseal dysplasia, MCDS or Schmid metaphyseal dysostosis and is a very rare inherited disorder characterized by short stature with abnormally short arms and legs, called short-limbed dwarfism and bowed legs, called genu varum.

Other physical characteristics may include outward "flaring" of the bones of the lower rib cage, lumbar lordosis, pain in the legs, and/or hip deformities in which the thigh bone is angled toward the center of the body, called coxa vara. Such abnormalities of the legs and hips typically result in an unusual "waddling" gait. MCDS is transmitted as an autosomal dominant trait (Psora/ Syphilis).

MCKUSICK TYPE METAPHYSEAL CHONDRODYSPLASIA

It is also called as cartilage-hair hypoplasia, CHH or ESS and is a rare progressive inherited disorder characterized by unusually fine, sparse hair and short stature with abnormally short arms and legs, called short-limbed dwarfism.

Portions of the long bones of the arms and legs develop abnormally with unusual cartilage formations and subsequent abnormal bone formation at the large (bulbous) end portions (metaphyses) of these long bones, called metaphyseal chondrodysplasia. Patients may have cellular immunodeficiency and pancytopenia (Psora/ Syphilis/ Sycosis).

JANSEN TYPE METAPHYSEAL CHONDRODYSPLASIA

It is also called as Jansen disease, Jansen metaphyseal dysostosis or Murk Jansen type metaphyseal chondrodysplasia and is an extremely rare progressive disorder in which portions of the bones of the arms and legs develop abnormally with unusual cartilage formations and subsequent abnormal bone formation at the bulbous end portions or metaphyses of these long bones, called metaphyseal chondrodysplasia. As a result, affected individuals show unusually short arms and legs and short stature, called short-limbed dwarfism, apparent during early childhood (Psora/ Syphilis).

OSMED, HETEROZYGOUS

It is also called as ‘Oto-Spondylo-Megaepiphyseal Dysplasia, Autosomal Dominant’, ‘Oto-Spondylo-Megaepiphyseal Dysplasia, Heterozygous’, ‘Pierre-Robin Syndrome with Fetal Chondrodysplasia’, ‘Stickler Syndrome Type III’, ‘Weissenbacher-Zweymuller Syndrome’ or WZS and is a rare genetic disorder characterized by skeletal malformations resulting in shortening of the upper limbs and thighs and short stature, called rhizomelic dwarfism. Additional symptoms include distinctive facial features and delays in psychomotor development (Psora/ Syphilis).

OSTEODYSPLASIA

It is due to defective development of bones and may be of following types-

OSTEOSCLEROSIS

It is an elevation in bone density, which is normally detected on an X-ray. Localized osteosclerosis can be caused by injuries that compress the bone, osteoarthritis, and osteoma (Psora/ Syphilis/ Sycosis).

OSTEOGENESIS IMPERFECTA

It is a rare condition with brittle bones. Multiple genetic mutations in different genes for collagen result in this condition (Syphilis).

GAUCHER DISEASE

In this disease visible bony abnormalities due to the accumulated glucosylceramide develop.

A deformity of the distal femur in the shape of an Erlenmeyer flask is commonly found which is due to aseptic necrosis of the femur joint (Syphilis/ Sycosis).

MELNICK–NEEDLES SYNDROME

It is a genetic disorder of bone characterized by skeletal and cranio-facial abnormalities with a specific facial appearance (Psora/ Syphilis).

OSTEOCHONDRODYSPLASIA

Osteochondrodysplasia is a general term for a disorder of the development of bone and cartilage both.

The mutation causing osteochondrodysplasia results from a single autosomal dominant gene, although the gene may have incomplete penetrance (Psora/ Syphilis).

It may be of several types-

CLEIDOCRANIAL DYSOSTOSIS

It is a general skeletal condition named for the collarbone- cleido and cranium deformities. It presents with partly or completely missing collarbones, open fontanelles, underdeveloped bones and joints, failure in eruption of permanent teeth or the permanent teeth including supernumerary teeth, bossing or bulging of the forehead and hypertelorism (Psora/ Syphilis).

FIBROUS DYSPLASIA

It causes thinning, growths or lesions in the bones. It may cause visible deformities in bones (Syphilis).

LANGER-GIEDION SYNDROME

It is a very rare genetic disorder caused by a deletion of chromosomal material. It presents with mild to moderate learning difficulties, short stature, unique facial features, small head and skeletal abnormalities including bony growths projecting from the surfaces of bones (Syphilis).

MAFFUCCI SYNDROME

It is a sporadic disease characterized by the presence of multiple enchondromas associated with multiple simple or cavernous soft tissue hemangiomas and lymphangiomas. The enchondromas affect the extremities and their distribution is asymmetrical. This syndrome displays during childhood and puberty (Psora/ Syphilis/ Sycosis).

KASHIN–BECK DISEASE

It is an endemic disorder of the bones and joints of the hands and fingers, elbows, knees, and ankles of children and adolescents who slowly develop stiff deformed joints, shortened limb length and short stature due to necrosis of the growth plates of bones and of joint cartilage (Psora/ Syphilis).

CONRADI-HUNERMANN SYNDROME

It is also known as X-linked dominant chondrodysplasia punctate and is caused by defects in the EPB gene (Syphilis).

JEUNE SYNDROME

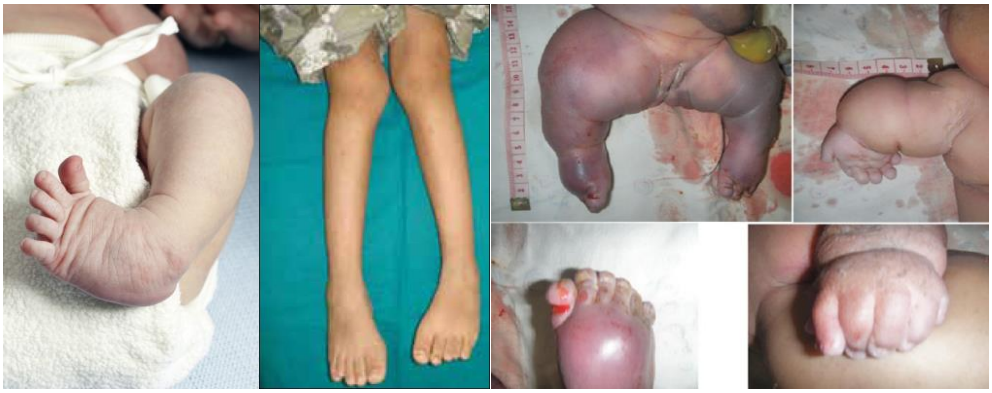
It is an extremely rare but severe recessive disorder that causes death in infancy or early childhood. It is also known as asphyxiating thoracic dystrophy (Syphilis).

Jeune syndrome is a ciliopathy caused by mutations in the IFT80 gene.

SIGNS AND SYMPTOMS

Specific symptoms of skeletal dysplasia vary depending on the disorder of skeletal dysplasia. Their arms, legs, trunk, or skull will likely develop with an unusual shape, size, or both. The common signs and symptoms of skeletal dysplasia are-

- Club feet
- Cognitive impairments or mental retardation
- Developmental delays
- Duplication of fingers or toes
- Fractured bones
- Joint pain
- Missing limbs
- Missing ribs
- Stubby fingers
- Disproportionate dwarfism
- Hydrocephalus
- Large head with prominent forehead frontal bossing
- Recurrent ear infections due to Eustachian tube blockages
- Short fingers and toes with trident hands
- Shortening of the proximal limbs, also called as rhizomelic shortening
- Sleep apnea both central and obstructive
- Small midface with a flattened nasal bridge
- Spinal kyphosis or lordosis
- Varus or valgus deformities



DIAGNOSIS

- Prenatal ultrasonography
- Skeletal X Ray
- Karyotype study

TREATMENT

Since it is a genetic disorder, there is no perfect treatment once the disorder is fully established. If deformities are not developed and there is rapid growth phase of the patient as in early childhood, the similimum Homoeopathic remedy can retard, suppress or even stop the abnormal growth process as well as enhance normal development, thereby minimizing deformities and promoting development of normal individual.

COMMON REMEDIES FOR SKELETAL DYSPLASIA

abrot. acon. agar. am-c. Am-f. ambr. anac. anan. ANG. ant-c. Ant-t. APIS apoc. ARG-MET. arg-n. Arn. ars-i. Ars. art-v. ASAF. aster. atro-s. aur-ar. AUR-M. aur-s. AUR. Bac. bad. BAR-C. Bar-f. bar-i. Bar-m. bar-p. bar-s. bell. Bism. borx. Bry. cadm-s. CALC-F. calc-i. CALC-P. CALC-S. calc-sil. CALC. canth. carb-ac. Carb-v. carbn-s. carc. caust. CENCH. Chin. chinin-s. choc. cic. cina Cinnb. clem. cocc. coff. colch. coloc. Con. conch. crot-c. crot-h. cupr-act. Cupr-f. cupr. cypr. cyt-l. daph. Dig. dros. Dulc. Ferr-i. Ferr. Fl-ac. Fl-pur. flav. galv. gels. graph. grat. GRIN. guaj. Hecla Hell. Hep. Hyos. ign. indg. Iod. iodof. Ip. ix. Kali-bi. kali-br. kali-c. Kali-f. Kali-i. kali-p. Lac-c. LACH. lap-a. Lith-f. LYC. mag-f. mag-m. Maland. Med. MERC-C. Merc-p. merc-pr-a. MERC. Mez. nat-c. Nat-f. nat-m. nat-sil. nep. NIT-AC. nux-m. nux-v. oeno. ol-j. OP. PH-AC. PHOS. Phyt. plat. Plb-act. plb-xyz. Plb. podo. psor. Puls. rhus-t. ruta sabin. samb. Sars. sec. sep. SIL. sol-ni. spig. squil. Staph. still. Stram. sulfa. SULPH. Syph. tarent. ther. thuj. thymul. Thyr. toxo-g. Tub. verat-v. verat. viol-t. zinc-br. zinc-m. Zinc.

SHORT REPERTORY OF SKELETAL DYSPLASIA

BACK - CURVATURE of spine acon. agar. ant-c. ASAF. aur. bar-c. Bar-m. bell. bry. CALC-F. calc-i. Calc-p. CALC-S. CALC. Carb-v. carbn-s. caust. cic. clem. coloc. Con. dros. dulc. ferr-i. hecla hep. ip. kali-c. lach. Lyc. MERC-C. Merc. mez. nat-c. nat-m. nux-v. op. PH-AC. Phos. plb. psor. Puls. rhus-t. ruta sabin. sep. SIL. staph. SULPH. syph. tarent. ther. thuj. tub.

BACK - EXOSTOSIS - Sacrum; on rhus-t.

CHEST – DEFORMED nat-m.

CHEST - EXOSTOSIS - Ribs; on calc-f. merc-c.

CHEST - EXOSTOSIS – Sternum merc-c.

CHEST – EXOSTOSIS Calc. Sil. Sulph.

EAR - EXOSTOSIS – Meatus calc-f. Hecla kali-i.

EAR – EXOSTOSIS puls.

EXTREMITIES - DEFORMED - Fingers – Joints kali-c. lyc. med.

EXTREMITIES - DEFORMED – Fingers ambr. anac. arg-met. Calc. caust. cina cocc. coff. colch. ferr. Graph. lyc. nux-v. phos. plat. RUTA sec. sil.

EXTREMITIES - DEFORMED - Hands - Back of hands plb.

EXTREMITIES - DEFORMED – Hands anac. caust. Lach. med. MERC. nux-v. sec.

EXTREMITIES - DEFORMED – Joints kali-c.

EXTREMITIES - DEFORMED - Upper limbs ant-c. lyc. sec.

EXTREMITIES - EXOSTOSIS - Feet – Heels conch.

EXTREMITIES - EXOSTOSIS – Feet hecla

EXTREMITIES - EXOSTOSIS – Fingers Calc-f. Hecla nat-sil. sil.

EXTREMITIES - EXOSTOSIS – Forearms Dulc.

EXTREMITIES - EXOSTOSIS – Joints sil.

EXTREMITIES - EXOSTOSIS - Knees – Patella calc-f.

EXTREMITIES - EXOSTOSIS - Legs - Bones – Tibia Ang. Aur-m. Aur. bad. calc-f. Calc-p. Cinnb. Dulc. Hecla merc-c. merc. NIT-AC. phos. Phyt. rhus-t. sars.

EXTREMITIES - EXOSTOSIS – Toes sil.

EXTREMITIES - EXOSTOSIS – Wrists choc. mag-m. ruta

EXTREMITIES – EXOSTOSIS aur-m. aur. CALC-F. calc. dulc. hecla mez. nat-sil. ph-ac. rhus-t. ruta SIL. staph. sulph. syph.

EXTREMITIES - KNEES; position of – inward bar-c. ix. lach. maland. nux-v. sep. staph.

EXTREMITIES - KNEES; position of – outward calc. nux-v. Ph-ac. Staph. sulph.

EYE - EXOSTOSIS – Orbits merc.

FACE - EXOSTOSIS - Jaws - dental origin; of hecla

FACE - EXOSTOSIS - Jaws – Lower ANG. CALC-F. Hecla Hep.

FACE - EXOSTOSIS – Jaws hecla

FACE - EXOSTOSIS - Malar bones – right Aur-m.

FACE - EXOSTOSIS - Malar bones aur.

FACE – EXOSTOSIS Aur-m. fl-ac. Hecla phyt.

GENERALS - DWARFISHNESS - children; in carc. med.

GENERALS - DWARFISHNESS - emaciated babies ol-j.

GENERALS – DWARFISHNESS ambr. aster. bac. BAR-C. bar-i. Bar-m. bar-p. bar-s. borx. CALC-P. Calc. Carbn-s. carc. Con. iod. lyc. mag-m. Med. merc-pr-a. merc. nat-m. nep. nux-m. Ol-j. ph-ac. sec. Sil. sulfa. SULPH. SYPH. thyr. Tub. zinc.

GENERALS - EXOSTOSIS - eruptions; after suppressed sulph.

GENERALS - EXOSTOSIS - injuries; after calc-f.

GENERALS - EXOSTOSIS – painful aur. daph. kali-i. merc. syph.

GENERALS - EXOSTOSIS – syphilitic fl-ac. hep. merc.

GENERALS – EXOSTOSIS am-c. Am-f. ang. Arg-met. AUR-M. AUR. Bar-f. Calc-f. Calc-p. calc. colch. crot-c. Cupr-f. daph. Dulc. Ferr-i. Fl-ac. Fl-pur. graph. hecla hep. Kali-bi. Kali-f. Kali-i. lap-a. Lith-f. mag-f. Maland. Merc-c. Merc-p. MERC. Mez. Nat-f. Nit-ac. ph-ac. PHOS. Plb-act. plb-xyz. Plb. Puls. rhus-t. Ruta Sars. SIL. staph. still. sulph. syph. zinc-m. Zinc.

GENERALS - GROWTH - complaints of growth process bar-c. calc-p. calc. ph-ac. phos. sil. Thyr.

GENERALS - HISTORY; personal - ear - inflammation - Internal; of recurrent calc. flav. Merc. psor. sil. thymul. tub.

GENERALS - SYPHILIS - accompanied by – exostosis Calc-f. fl-ac. Hecla merc-p. phos.

HEAD - EXOSTOSIS – painful Aur. carbn-s. Kali-i. MERC. syph.

HEAD - EXOSTOSIS – sensitive syph.

HEAD – EXOSTOSIS anan. ARG-MET. aur-m. AUR. CALC-F. Calc. carbn-s. cupr. Fl-ac. hecla Kali-i. merc-p. MERC. Mez. nit-ac. PHOS. Phyt. sars. sep. sil. still. syph.

HEAD – HYDROCEPHALUS abrot. acon. am-c. APIS apoc. arg-n. Arn. ars-i. Ars. art-v. atro-s. aur-ar. aur-s. Aur. Bac. bar-c. bell. Bism. Bry. cadm-s. calc-i. Calc-p. calc-sil. CALC. canth. carb-ac. caust. Chin. chinin-s. cina coloc. Con. crot-h. cupr-act. cupr. cypr. cyt-l. Dig. ferr-i. Ferr. galv. gels. grat. Hell. Hyos. ign. indg. Iod. iodof. Ip. kali-br. Kali-i. kali-p. lach. LYC. mag-m. Merc. Nat-m. nux-v. oeno. Op. ph-ac. Phos. plat. podo. Puls. rhus-t. samb. sep. SIL. sol-ni. spig. squil. Stram. Sulph. thuj. toxo-g. tub. verat-v. verat. viol-t. zinc-br. Zinc-m. zinc.

HEAD - SMALLER - left half retarded in growth fl-ac.

MOUTH - EXOSTOSIS at roof of mouth asaf. Aur.

NOSE – EXOSTOSIS Aur. merc. phos.

RESPIRATION - ARRESTED - sleep - during - agg. am-c. Ant-t. cadm-s. Carb-v. CENCH. dig. GRIN. guaj. Hep. Kali-c. Lac-c. LACH. lyc. OP. phos. samb. Sulph.

TEETH - DEFORMED – distorted sil. syph.

TEETH – DEFORMED sil. syph.

TEETH – DWARFED bac. staph. Syph.

URETHRA - FUNGOID growth calc. con. graph. lyc. thuj.

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Radar 10


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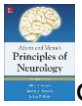
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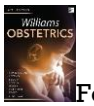
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