Achondroplasia

Achondroplasia is characterized as **disproportionate dwarfism** with short limbs, an average adult height of 125 cm. This disease is one of the most common bone dysplasias(incidence 1.5:10,000 live births).

Clinical picture	disproportionate dwarfism with short limbs, adult height on average 125 cm
Cause	enchondral ossification disorder of all bones (mutations in the *FGFR3* gene)
Diagnostics	prenatal: sonographic measurement of the length of long bones, skull atypia; diagnosis verification: targeted molecular genetic analysis of the *FGFR3 gene*
Investigation in the Czech Republic	list of workplaces
Incidence in the world	1/25000 live births (worldwide)
Classification and references	
ICD-10	Q77.4
MeSH ID	D000130
OMIM	100800
orphaned	ORPHA15
MedlinePlus	001577
Medscape	1258401

Etiopathogenesis

Achondroplasia is **an autosomal dominant** hereditary disease, but up to 90% of children are born on the basis of new mutací (the risk factor for new dominant mutations is mainly the older age of the father, especially over 40 years), homozygotes are mostly stillborn. There is a **defect in the enchondral ossification** of all bones (*FGFR3* gene, 4p16.3, perichondral (desmogenous and periosteal) ossification occurs normally. The most pronounced impairment is the growth of long bones. The epiphyses and joint surfaces have a normal shape, the width of the cortical bone is normal.

Clinical picture

Disproportional growth is typical the child is usually hypotonic after birth. **The trunk** is almost normal in length, but the limbs are very short (micromelia). Center of gravity of the body shifted cranially.

Another feature is a pear-shaped **skull** an enlarged, widely raised forehead, supraorbital arches and mandible, depression of the nasal cavity (the impression of a wider spacing of the eye sockets).

Proximal segments of the **limbs** are affected most (rhisomelic form, their growth plates are normally the most active), there is a disproportion of the length of the lower lef to the thigh and forearm to the arm. **The hands** are short, wide **fingers** of the same length and shape (microdactyly), a hand similar to a „trident“ (inability to pull the 4th finger to the 3rd when stretched). On the upper limbs, flexion contracture of the elbows appears, often with dislocation of the radial head. They also suffer from angular deformity of the lower limbs (mainly genu varum with varus tibia and a relatively longer fibula).

Hyperlordosis can be observed on the lumbar spine (often with stenosis of the spinal canal, preceded by thoracolumbar hyperkyphosis), in the sacral region*sacrum acutum* až *sacrum horisontale*.

Disabled people have normal, often even above-average **intelligence**.

Musculature developed normally, soft tissues of limbs folded into folds, internal organs are normally developed.

The average life expectancy is not reduced, some studied even indicate that it is higher, despite the fact that fatal injuries occur more often.

The unusally small **height** (in adulthood, men average 131 cm, women 124 cm) handicaps the disabled socially (e.g. they cannot reach the light switch, elevator buttons, sink tap, difficulty using public transport).Mobility varies widely. Some can be unusally agile (e.g. Andre Bouchet), but for many of them movement is difficult.

X-ray image

Distinct deformities. Long bones of the limbs shortened with a noticeable widening of the diameter and an increase in density. **Expansion of the metaphyses** of the tubular bones (caused by the growth cap), the epiphyses have a normal shape. V or U shaped growth plates, arcuate **curvature of the diaphyses** – most pronounced on lower leg

and forearms. Irrgular configuration of hip joints, flat acetabulum. Kidney *like aditus pelvis* (pelvis widened laterally and shortened anteroposteriorly). Reduced height of vertebral bodies. Cranial base shortened, *foramen magnum* reduced.

Prenatal diagnostics

Sonographic measurement of femur length, genetic examination (*FGFR3* gene).

Therapy

The effect of STH administration is individually variable.

Neurosurgical expansion of the *foramen magnum* in craniocervical spinal stenosis (risk of sleep apnea syndrome, sudden death, spasticity)

Treatment of thoracolumbar hyperkyphosis with a **corset**, treatment of spinal stenosis in adults.

Deformities of the diaphyses are corrected by **prolongation osteotomy** using the Ilizarov technique (the aim is to obtain a body height of around 150 cm) – gradual prolongation in 2-3 stages (simultaneous prolongation of both femurs or tibias / crossed prolongation of the femur and bilateral tibia). Risks: infection, joint and muscle contractures, circulatory complications.

Compensatory aids (e.g. bycicle adapted to height and proportions to facilitate movement)

Differential diagnostic

Lethal forms of dwarfism (thantophoric dwarfism), hypochondroplasia, mucupolysaccaridosis (Morquiuo syndrome).

Links

References

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