HYPOCHONDROPLASIA

DESCRIPTION:

Hypochondroplasia, a bone dysplasia, is very similar to achondroplasia. Like achondroplasia, it affects the FGFR3 gene responsible for the linear growth of long bones and cartilage.
The final height of the adult varies between 118 and 165 cm (3'10" and 5'5"). The average adult size is 142 cm (4'8").
Prevalence is estimated to be about 1 in 30,000-33,000. The condition is an autosomal dominant trait (a child of an affected parent has a 1 in 2 chance of being impacted). However, in 90% of cases, it is a new genetic mutation/variant, meaning that the child is born to parents of average height [see "Genetics"].

CHARACTERISTICS AND DIAGNOSIS:

Though similar to achondroplasia, hypochondroplasia is less severe and causes fewer complications than its cousin. It is characterized by a disproportionately small size, to which certain physical characteristics are added:

- Moderate lumbar lordosis (hollow in the lower back);
- Micromelia (shortening of the limbs);
- Limited elbow extension;
- Macrocephaly (bulky head) present in approximately 50% of cases;
- Genu varum (bowed legs) present in 10-20% of cases.

At birth, hypochondroplastic babies are very often of average size and weight. Clinical manifestations usually appear during infancy. Diagnosis is therefore late (around 2-4 years of age) and is made when an inflection of the growth curve is observed. X-rays typically show a moderate narrowing of the inter-peduncular distances, shortening of the tubular bones with distal lengthening of the fibula and a short and enlarged femoral neck.
Hypochondroplasia does not exhibit facial dysmorphia, orthopedic malformations, or neurological problems. However, the association with an intellectual deficit has been reported in 10-12% of children with hypochondroplasia.

MAIN POSSIBLE COMPLICATIONS:

Developmental issues: There is an unexplained risk of increased learning disabilities in children with hypochondroplasia. Nearly half of the children are affected by this problem.

Hearing issues: Many infants and children with hypochondroplasia have middle ear

dysfunction. They are therefore more at risk of having hearing problems, which, if left untreated, can cause delays in speech and language development.
An eardrum and hearing assessment should be performed as early as 9 or 12 months of age, and then once a year during infancy.
If necessary, a myringotomy (insertion of a ventilation tube into the eardrums) may be performed.

Genu varum (bowed legs): It is important to monitor the alignment of the limbs and the presence of chronic pain. If necessary, a surgical procedure called an osteotomy (surgical section of the long bones) can be performed to restore the alignment of the legs.

Spinal deformities: Some people with hypochondroplasia may have lordosis (a hollow in the lower back) or scoliosis (a three-dimensional deformity). Therefore, the spine should be monitored clinically, and X-rays should be taken when in doubt. If necessary, a brace or corset may be recommended.

Hydrocephalus (excess cerebrospinal fluid in the brain): It is often benign. However, in less than 5% of cases, the placement of a shunt to drain the liquid out of the brain is necessary (surgical operation). Hydrocephalus must be monitored, with the signs of increased intracranial pressure recognized and addressed to avoid serious damage.
These signs include headaches, vomiting or nausea, visual disturbances, changes in alertness and consciousness (drowsiness, attention problems, apathy, etc.).

Epileptic convulsions: Between 5-10% of hypochondroplastic individuals have this type of seizure. Almost all people with this condition also have a structural abnormality of the brain called temporal lobe dysgenesis.
Children with episodes of sleep apnea or seizure like episodes should have both electroencephalography and magnetic resonance imaging of the brain.
If necessary, standard treatments for epilepsy can be used.

Spinal cord compression: It can occur but is much less common than in individuals with achondroplasia.
Symptoms to watch for include: Cervical pain and stiffness of the neck. The lower and upper limbs may be affected. Muscle weakness, numbness, decreased fine motor skills, gait and balance disorders, functional impotence, sphincter disorders (incontinence, dysuria and/or pollakiuria). If left untreated, compression can lead to paralysis and bladder control issues.
Surgery: Cervical or dorsolumbar decompression, with or without fusion.

TREATMENT:

The management of hypochondroplasia is multidisciplinary (surgery, physiotherapy, occupational therapy, orthopedics, etc.) and preventive, with the primary goal of detecting

and treating complications. Currently, there is no specific treatment for hypochondroplasia. Tests with growth hormone show only a very limited effect, which is logical, since this disorder results from an intrinsic abnormality of bone growth. Limb lengthening, a controversial treatment, is chosen by a small minority of affected individuals.

List of the main elements to be monitored or managed [see the sheet "frequent interventions for people of short stature"]:

- Developmental delay;
- Hearing problems;
- Risk of hydrocephalus;
- Leg (genu varum) and back deformities;
- Epileptic convulsions;
- Spinal cord compression.

RESOURCES:

Association québécoise des personnes de petite taille
https://www.aqppt.org/

Little People of Ontario
https://littlepeopleofontario.com/

Regroupement québécois des maladies orphelines - Centre iRARE
https://rqmo.org/centre-dinformation-et-de-ressources-en-maladies-rares/

Orphanet
https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=FR&Expert=429

Little people of America
https://www.lpaonline.org/assets/documents/NH%20Hypochondroplasia1.pdf

NORD – National Organization for Rare Disorders
https://rarediseases.org/rare-diseases/hypochondroplasia/

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