ACHONDROPLASIA

DESCRIPTION :

Achondroplasia, a bone dysplasia, is the most common form of dwarfism. It results from mutations in the FGFR3 gene, which affects the linear growth of long bones. Impacted individuals remain short in stature because of damage to their growth plates (the part of the bone associated with the growth or lengthening of long bones). In adulthood, achondroplastic individuals range in height from 115 to 145 cm (3'8" to 4'9").

Achondroplasia affects about one in 25,000 births and is an autosomal dominant trait (i.e., a child of a parent with achondroplasia has a 1 in 2 chance of inheriting achondroplasia). However, in 80% of cases, it is a spontaneous mutation, meaning that the child is born to parents of average height [see "Genetics"].

CHARACTERISTICS AND DIAGNOSIS :

Achondroplasia is a type of disproportionate dwarfism. The condition is often detected at birth because of the disparity between the (shortened) limbs and the trunk. In addition, there may be other identifiable physical characteristics:

- Macrocephaly (larger head);
- Protruding forehead;
- Depressed nose at the bridge;
- Trident hands (wider spacing between middle and ring fingers);
- Kyphosis (humpback) and/or lordosis (exaggerated hollow in the lower back);
- Genu varum (bowed legs);
- Joint hypermobility;
- Shorter thorax.

Diagnosis is based on the presence of clinical and radiological features. X-rays show rhizomelia (shortening of the upper limb sections), generalized metaphyseal irregularities (growth plates located under the epiphysis), reduced interpedicular distance between the lower lumbar vertebrae and an abnormal pelvis.

Molecular genetic testing confirms the diagnosis by demonstrating the FGFR3 mutation.
At birth, babies are only slightly smaller than average (47 cm instead of 50 cm, or 18.5 in. instead of 19.7 in.).

MAIN POSSIBLE COMPLICATIONS :

Genu varum (bowed legs): It is important to be aware of and to watch for limb dysplasias and chronic pain. If necessary, a surgical operation, an osteotomy (a surgical cutting and realigning of the long bones), may be performed to restore the alignment of the legs.

Hypotonia: Muscle weakness in children under 2 years of age can delay motor development (e.g., lifting the head, sitting, standing or walking) Development trails from between 6 to 12 months behind the average; therefore, the developmental assessment should be compared to achondroplasia-specific standards.
Muscle tone can be stimulated through physiotherapy exercises. Wearing a back brace may also be considered if the spine shows curvature.

Hearing problems: Most infants and children have some irregularity of the middle ear, which promotes the development of ear infections. Typically characterized by sudden and severe pain in the ear, as well as fever and hearing loss. Additionally, about 1 in 3 ear infections can also go unnoticed when the only symptom is a feeling of blockage in the ear, accompanied by hearing loss.
To prevent hearing issues, 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 (the insertion of a ventilation tube into the eardrum) may be performed.
If left untreated, hearing problems can cause delays in speech and language development., This can manifest in nearly one in four children with achondroplasia.

Hydrocephalus (excess cerebrospinal fluid in the brain): Occurs in the first few months of life and often resolves on its own by age 2 or 3. However, in 3-5% of cases, a shunt to drain the fluid out of the brain is necessary (surgical operation). Hydrocephalus must be monitored, and the signs of increased intracranial pressure must be recognized and addressed to avoid serious complications. These signs include headaches, vomiting or nausea, visual disturbances, and issues with alertness and consciousness (drowsiness, attention problems, apathy, etc.).

Obstructive Apnea: Common in achondroplastic children aged 2 to 10 years. If left untreated, obstructive apnea can lead to chronic respiratory failure as well as lung and heart problems.
The main signs of obstructive apnea are loud snoring, neck hyperextension, apneic pauses, compensatory sighs, new onset of enuresis (bed-wetting), irritability, memory or concentration problems, and drowsiness during the day.
Weight loss, removal of tonsils and/or adenoids, and/or the use of ventilators (CPAP or BPBV) are the most recommended techniques for treating obstructive sleep apnea [see the "Sleep Apnea" fact sheet].

Occipital Hole Stenosis (small area between the neck and skull): Narrowing of the occipital hole, which is common in infants with achondroplasia, can have serious consequences if it compresses the spinal cord. When compression occurs, the spinal cord and the medulla

oblongata (which regulates breathing and heart rate, among other important bodily functions) are affected. Neurological damage and breathing difficulties are possible (including central sleep apnea), and can lead to sudden infant death in 2-7% of cases.

Because of these risks, the presence of possible compression should be ruled out from the first days of life of the newborn with achondroplasia. (This work-up can include an MRI looking for a narrowing of the occipital hole and/or an increase in the volume of the brain ventricles, a test to measure the brain's reactions to nerve fibre stimulation, polysomnography to diagnose sleep apnea, etc.). If the tests show nothing abnormal, it is suggested that they be repeated at regular intervals during the first years of life.
If the tests confirm compression, an operation must be performed by a neurosurgeon to enlarge the occipital hole.

Cervical (neck) or dorsolumbar (back) spinal cord compression: Acute or chronic spinal cord injuries can occur and cause neurological disorders if left untreated.
Symptoms to watch for are neck or back pain as well as stiffness in the neck. The lower and upper limbs may be affected. Muscle weakness, numbness, decreased fine motor skills, gait and balance problems, functional impotence, sphincter disorders (incontinence, dysuria, polycythemia, and sometimes stress incontinence) can be experienced. These disorders can often appear towards the end of adolescence. If left untreated, compression can lead to paralysis and problems with bladder and bowel control.
Surgery: Cervical (neck) or dorsolumbar (lower back) decompression, with or without fusion.

Anesthesia: The risk of complications is increased due to spinal, neurological and sleep apnea problems resulting from achondroplasia.
Therefore, in the event of an operation, it is important to ensure that an experienced anesthetist is consulted and present. Finally, before implementing any invasive technique, it is necessary to have the patient's lung function evaluated [see the "anesthesia" section].

TREATMENT:

The management of achondroplasia is multidisciplinary (surgery, orthopedics, occupational therapy, physiotherapy, speech therapy, etc.) and preventive in its nature. It is essentially geared towards the prevention and detection of complications. Currently, there is no specific treatment for achondroplasia. Growth hormone tests do not show any long-term benefits. Limb lengthening, a controversial treatment, is chosen by a small minority of individuals. Currently in trials, there are some new interventions specifically targeting the affected metabolic pathways, being evaluated.

List of the main elements to be monitored as a function of age [see the sheet "frequent interventions in short people"]:

- Infant (1 month to 1 year): Risks of hydrocephalus, compression of the occipital hole, hypotonia, kyphosis, sleep apnea, otitis and hearing acuity;
- Early childhood (1 to 5 years): kyphosis, ear and hearing impairment, hip hyperflexion and/or genu varum, gastroesophageal reflux, sleep apnea;
- Childhood (5 to 13 years) and adulthood: Weight control, cervical or dorsolumbar spinal cord compression, sleep apnea.

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 - information resource on achondroplasia
https://www.orpha.net/consor/cgi-bin/Disease_Search.php?lng=EN&data_id=148

Little people of America - fact sheet on pseudo-achondroplasia
https://www.lpaonline.org/assets/documents/NH%20achondroplasia.pdf

OMIM – fact sheets on achondroplasia
https://www.omim.org/entry/100800

Association Québécoise des Personnes de Petite Taille

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