PSEUDO-ACHONDROPLASIA

DESCRIPTION :

As its name suggests, pseudo-achondroplasia resembles achondroplasia, with which it has often been confused. Pseudo-achondroplasia, a bone dysplasia, affects the COMP gene. Affected individuals are smaller than average because of an abnormality in the production of proteins in the growth cartilage of long bones. The average adult height is 120 cm (3'11") in men and 116 cm (3'10") in women.
The diagnosis, which affects 1 in 30,000-60,000 births, is an autosomal dominant trait [see "Genetics"].

CHARACTERISTICS AND DIAGNOSIS :

Pseudo-achondroplasia causes disproportionate dwarfism. The limbs are short while the trunk and head are of average size. Unlike achondroplasia, the face is not affected. People with pseudo achondroplasia also have a waddling gait. The main structural characteristics are as follows:

- Short fingers, square phalanges and short nails;
- Hypermobility of the hands;
- Joint stiffness in the elbows and hips, accompanied by pain;
- Varus or valgus bowing in the legs;
- Lordosis (exaggerated hollow in the lower back) and/or kyphoscoliosis;
- Myopathy (muscle disease) in a minority of individuals.

The size at birth is average. The diagnosis is often made in the second year of life, when the parents report significant growth restriction and difficulties in learning to walk. The radiological diagnosis is based on the observation of epiphyseal and metaphyseal anomalies, marked by insufficient development of the areas at the ends of the long bones.

PRIMARY POSSIBLE COMPLICATIONS :

Hypotonia: Muscle weakness in children under 2 years of age causes delays in motor development (raising the head, sitting, standing, walking, etc.). The child is about 6 months behind the average individual for such milestones.
Muscle tone can be developed through physiotherapy exercises. Wearing a corset or brace may also be considered.

Hearing problems: Individuals with Pseudo-achondroplasia are more prone to ear infections than other children. Repetitive ear infections, sometimes even continuous, can lead to hearing

problems which, if left untreated, can cause delays in speech and language development.
An evaluation of the ears and eardrums should be done regularly from the time of diagnosis.
If necessary, a myringotomy (insertion of a ventilation tube into the eardrums) may be considered.

Varus or valgus (outward or inward bowing of the legs): It is important to monitor the alignment of the limbs and the development of chronic pain. If surgical correction is to be considered, it should be planned at the right age because of the risk of recurrence. Multi-level osteotomies (surgical sections of long bones) are often required (e.g. correction at the distal femoral and proximal tibial levels).

Early Osteoarthritis of the hip: Can occur as early as childhood. A regular radiological check-up should therefore be performed to monitor any progress of osteoarthritis.
The most common symptoms of osteoarthritis of the hip are localized pain in the groin, joint stiffness and loss of mobility.
More than half of people with pseudo-achondroplasia will need a joint replacement; an operation to restore the shape and mobility of the hip joint. The operation is often performed around the age of thirty or forty. It requires personalized prostheses.

Kyphoscoliosis (double deformation of the spine combining kyphosis and scoliosis): Usually begins in late childhood or adolescence and affects nearly half of all people with pseudo-achondroplasia.
A clinical examination should be performed every six months upon diagnosis. X-rays are required if kyphoscoliosis is suspected.
Surgical fusion is sometimes required to correct kyphoscoliosis.

Cervical spine instability: Present in a minority of individuals with pseudo-achondroplasia (approximately one in six). Instability should be monitored, as it can lead to spinal cord compression.
Symptoms to watch for include: Cervical pain, 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, pollakiuria and sometimes stress incontinence). If left untreated, compression can lead to paralysis and bladder control problems.
Surgery (rarely necessary): Cervical decompression with fusion.

TREATMENT:

The management of pseudo-achondroplasia is multidisciplinary (orthopedics, occupational therapy, physiotherapy, etc.) and preventive, and is essentially aimed at detecting and avoiding complications. Currently, there is no specific treatment for pseudo-achondroplasia.

Tests on growth hormones show only a very limited effect, which is logical, since this diagnosis 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 and managed [see the sheet "frequent interventions for people of short stature"]:

- Hypotonia;
- Hearing problems;
- Varus or valgus bowing of the legs;
- Osteoarthritis of the hip;
- Cyphoscoliosis;
- Cervical instability and the associated risk of 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 - explanatory sheet on pseudo-achondroplasia
https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=en&Expert=750

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

Association Québécoise des Personnes de Petite Taille

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