ACROMESOMELIC DYSPLASIA

DESCRIPTION :

Acromesomelic dysplasia is a group of rare skeletal conditions that affect the development of bone and cartilage. The average adult height is unknown but is estimated to be less than 120 cm (3'11"). Current knowledge suggests that this group has five types or classes:

- The Osebold-Remondini type, which has not yet been genetically mapped;
- The Maroteaux type, caused by mutations in the NPR2 gene;
- Du Pan type and Grebe dysplasia (including Hunter-Thompson type), which result from mutations in the GDF5 gene (previously known as CDMP1);
- Acromesomelic dysplasia with genital abnormalities, caused by mutations in the BMPR1B gene.

The different types of acromesomelic dysplasia have an autosomal recessive transmission pattern, except for the Osebold-Remondini type, which is transmitted by the autosomal dominant mode [see "Genetics"].
The exact prevalence of the skeletal disorder is unknown.

CHARACTERISTICS AND DIAGNOSIS :

Acromesomelic Dysplasia results in disproportionately short limb dwarfism. The main features of this type of dwarfismare as follows:

- Mesomelic shortening of limbs (affecting the forearms and legs below the knees);
- Short, stocky hands and feet;
- Kyphosis (hunchback) or lordosis (hollow in the lower back);
- Macrocephaly (larger head) and prominent forehead;
- Madelung's deformity (malformation of the wrists).

Acromesomelic Dysplasia is visible at birth, as newborns have abnormalities of the head, face, feet and hands. However, it is only in the first years of life that dwarfism appears, when the forearms, lower legs, hands and feet do not develop in proportion to the rest of the body.
X-rays confirm the abnormal development and premature fusion of the epiphyses (head) of the bones of the feet, hands, forearms and lower legs. These include a shortened ulna and radius (forearm bones), a dislocated or subluxated radial head (tip of the radius), short and malformed phalanges, and vertebral abnormalities.

MAIN POSSIBLE COMPLICATIONS:

There are no major medical or orthopedic complications in the evolution of this type of

dwarfism. However, the following are some of the problems that persons with a diagnosis of acromesomelic dysplasia may face:

- **Madelung's deformity** (wrist subluxation): Causes pain in the arms and wrists and limits mobility. Clinical and radiographic follow-up should be done regularly to monitor the potential progression of pain and loss of function.
  Splints can be used to reduce wrist discomfort.
  Surgery may be required to reduce pain and restore wrist function.

- **Premature Osteoarthritis**: Develops especially in the elbows and promotes stiffness, tenderness and pain.
  Clinical and radiological monitoring should be performed periodically from the time of diagnosis and in accordance with the onset of pain and functional limitations.

- **Kyphosis or lordosis**: In some cases, children affected by acromesomelic dysplasia may develop kyphosis (humpback) and/or lordosis (hollow in the lower back).
  Clinical monitoring should be done regularly, and X-rays should be taken as soon as any concern arises.

- **Anaesthesia**: Due to the small size of the patient, precautions must be taken when administering anaesthesia. Make sure that an experienced anesthetist is consulted and present (see the "anesthesia" sheet).

TREATMENT:

The management of acromesomelic dysplasia is predominantly concerned with functional restrictions, pain and aesthetics associated with the diagnosis. For this reason, several specialists such as pediatricians, orthopedists, surgeons and/or physiatrists must be called upon.

List of the main elements to be monitored and managed:

- Madelung deformity;
- Early Osteoarthritis;
- Kyphosis and/or lordosis.

Growth hormone treatment when tested on patients with the Maroteaux type, showed no improvement in growth rate and therefore is not recommended for acromesomelic dysplasia. Limb lengthening surgery, a controversial therapeutic practice, can be an option but the long-term side effects are not well documented.

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/

Little people UK – acromesomelic dysplasia fact sheet
https://littlepeopleuk.org/information-about-dwarfism-conditions/types-of-dwarfism/acromesomelic-dysplasia

Orphanet – fact sheet on acromesomelic dysplasia of the Maroteaux type https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=40

Orphanet – Fact sheet on Grebe type acromesomelic dysplasia
https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=2098

NORD – National Organisation for Rare Disorders
https://rarediseases.org/rare-diseases/acromesomelic-dysplasia/

GARD – Genetic and Rare Diseases Information Center
https://rarediseases.info.nih.gov/diseases/6/acromesomelic-dysplasia

Association Québécoise des Personnes de Petite Taille

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