P2-93 CHARACTERIZATION OF PATIENTS WITH ACHONDROPLASIA IN A PEDIATRIC CLINIC OF CALI, COLOMBIA

Liliana Mejía De Beldjenna Endocrinóloga pediatra.
Fundación Clínica Infantil Club Noel . UNILIBRE GRIMPED.Cali Colombia

INTRODUCTION

Achondroplasia is the most common of the skeletal dysplasias and short stature with severe anatomic disproportion. Bone endochondrial growth is affected.
It is an autosomal dominant monogenic disease with complete penetrance.
Incidence is 1/25000 to 1/40000 of live births. It is caused by a mutation of the gene of fibroblast growth factor (FGFR3) located on the shorth arm of chromosome 4.
DIAGNOSIS; Is clinical and patients present with asymmetrical short stature, poor development of the solid middle phase, flattening of the middle phase,,flat nasal bridge, small thorax, thin ribs , elbow limitation, hypotonic trunk with macrocranium, flat chest, prominent abdomen, thoracolumbar kyphosis, lumbar hyperlordosis, articular hypermobility and bowing of the middle segment of the leg.
Males reach 131±5.6 cm and women 124±5.9 cm. There are many complications and reduced life span requiring a multidisciplinary management

AIM

To characterize patients with achondroplasia seen in the Pediatric Endocrinology Clinic of Club Noel of Cali, Colombia between June of 2015 and June of 2020, Study an observational descriptive type of number of cases.

METHOD

Review of clinical charts during the established period

RESULTS

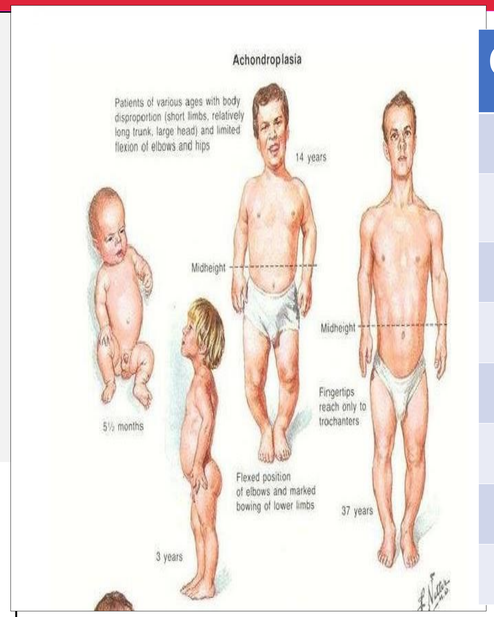
We found 8 patients with equal sex distribution.

75% of cases were diagnosed in utero and 87.5% were born full term. Range 29 to 40 weeks gestacional age . Average weight at birth of 3.388 kg and height of 47 cm Hypothyroidism and hypoacusia were not present in any of the patients.

In 37.5% lower limb surgery was required. 25% underwent bone lengthening as a low size management option.

MUTATION of FGFR3 gene was confirmed in 4 patients c 1138** y c1144. pGlic >Arg . Mean final growth was 127 cm,:

the male with 138 cm and 122 cm the female..



CLINICAL FEATURINGS n= 9	%
Macrocranium	100
Hydrocephalus	25
Middle phase hypoplasia	100
Trident hand	85
Lumbar hyperlordosis	100
Disproportion of segments	100
Rhizomelia	100
Short stature	100

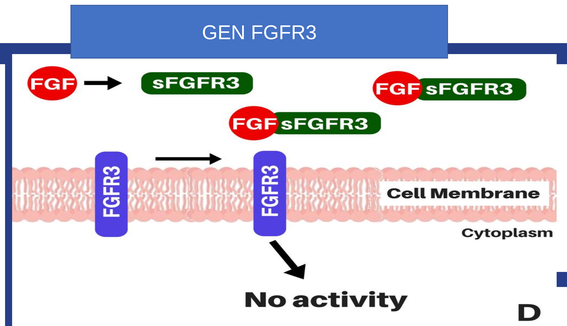
CONCLUSIONS

Patients with achondroplasia consult for asymmetrical short stature with bone and neurologic abnormalities requiring a multidisciplinary approach to improve the quality of life.

This work contributes to our national and regional statistics

REFERENCES

1. Mejia de Beldjenna Liliana. “Medical and Orthopedic Management with Growth Hormone and Bone Lengthening in a Patient with Achondroplasia”. EC Orthopaedics 3.1 (2016): 229-232.
2. Hoover-Fong, J.E., Alade, A.Y., Hashmi, S.S. et al. Achondroplasia Natural History Study (CLARITY): a multicenter retrospective cohort study of achondroplasia in the United States. Genet Med (2021).



CONTACT INFORMATION

Liliana Mejia de Beldjenna
.lmameza2@yahoo.com. Endocrinology paediatric Foundation Clinic Club Noel xt.