Endocrinologia | Casuística / Investigação

PAS-002 - (21SPP-11489) - LIFETIME IMPACT OF ACHONDROPLASIA IN EUROPE (LIAISE): FINDINGS FROM A MULTINATIONAL OBSERVATIONAL STUDY

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Introdução e Objectivos
Achondroplasia (ACH), the most common skeletal dysplasia, leads to significant multisystem complications. We report medical history data obtained from a multinational observational study (NCT03449368) designed to quantify the lifetime impact of Achondroplasia.

Metodologia

A retrospective cohort study of individuals with ACH aged ≥5 years in 6 European countries (Austria, Germany, Italy, Spain, Sweden, Denmark) assessed clinical burden and healthcare resource use data from medical records with a ≥5 year look-back period.

Resultados

Data were collected from 186 individuals (aged 5-84) (54.3% female; mean age 21.7 ± 17.3 years), including 40 (21.5%) who had experienced limb lengthening procedures. Most frequently reported medical complications were musculoskeletal and connective tissue disorders (12.0/100 patient-year [PY], 58.6%), nervous system disorders (8.7/100 PY, 61.3%), infections and infestations (6.2/100 PY, 25.8%), respiratory, thoracic and mediastinal disorders (5.8/100 PY, 41.9%), and ear and labyrinth disorders (5.4/100 PY, 43.5%). ENT and neurological issues were more common in children. Pain and spinal cord compression/stenosis increased as aged patients. Exploratory demonstrate demonstrate that in children, height was negatively correlated with ENT issues, spinal deformities, and infections, and height Z-score was negatively correlated with genu varum. Most frequent medications were analgesics (8.6/100 PY, 33.3%), antibacterials (7.3/100 PY, 37.1%), and anti-inflammatory/anti-rheumatic products (4.7/100 PY, 28.5%). 72.0% underwent at least one surgical procedure.

Conclusões
Significant clinical burden and healthcare utilization is associated with ACH.

Palavras-chave : achondroplasia, skeletal dysplasia