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Basal carcinoma cell at paediatric age Gorlin-Goltz syndrome: clinical experience

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Introduction: The Gorlin-Goltz syndrome is an autosomal dominant inherited disease, associated with a PTCH gene mutation. Its presentation is polymorphous, being frequent the appearance of the clinical triad based on carcinomas basal cell, odontogenic cysts and skeletal abnormalities.
The estimated prevalence is 1: 150.000 with a ratio male/female 1: 1.
Skin lesions are a very frequent reason for consultation during paediatric age, being valued in most cases in Primary Care. Sometimes, patients would need the intervention of other specialists to deep in the given area.
Aim: To share a clinical experience of a syndrome with low appearance frequency. To evaluate the role of the specialist who develops its activity at Primary Care, with patients who requires a multidisciplinary intervention.
Material and Method: Clinical case: Two-year-old child arrives to consultation due to injuries at thorax, hands and feet of a month of evolution. Personal history: humeral fracture in 2011 and asthma.
The examination showed palmoplantar pits, thorax injuries difficult to typify macroscopically, hypermobile joints.
Results: Karyotype: PTCH1 gene mutation. Skin biopsy solid basal cell carcinoma. Orthopantomography: Anodontia of upper lateral incisors. Complementary tests within normal limits. Current monitory: Maxillofacial, Dermatology and Paediatrics Hospital Service.
Conclusion: Clinical and genetic confirmation of Gorlin-Goltz syndrome. The initial assessment in primary care was vital to the diagnosis orientation and early detection of the disease. Once confirmed the diagnosis, the family doctor has not been included in the multidisciplinary monitoring team. As an opportunity to improve, it is necessary to optimize the communication channels with other specialists, so these patients can continue receiving patient focused cares rather than just disease treatment.