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Case report of juvenile hyaline fibromatosis

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Introduction: Juvenile hyaline fibromatosis is classified as a rare disease, with an autosomal, recessive pattern of inheritance, characterized by the deposition of hyaline matter in the tissues.

Case report: A 33-year-old patient who began to suffer from joint pain, flexion joint stiffness, in particular elbows and knees, from approximately 6 months of age, as well as the progressive appearance of movable subcutaneous fibrous tumors present in both ears. In addition, a slight pseudostatural hypodevelopment and severe hyperplasia and gingival hypertrophy. Radiological studies showed a bone age consistent with the chronological, showing slight osteoporosis, digital tumors without osteolysis.

Conclusion: Juvenile Hyaline Fibromatosis is one of the less frequent rare diseases. The case is the only one reported in the records of Medical Genetics of Pinar del Río known to date. The multidisciplinarity of key specialties such as Pediatrics, Oncology and Genetics is evident.

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