Introduction

Raine Syndrome is an autosomal recessive disorder associated to mutation of the FAM20C gene in chromosome 7, whose DMP4 protein is thought to play an important role in dentin mineralization. This congenital osteosclerotic bone dysplasia is characterized by generalized osteosclerosis, facial dysmorphism, chonagal atresia or stenosis, intracranial calcifications, and thoracic hypoplasia. Since first reported by Raine et al. in 1989, only 23 cases of this syndrome have been reported.

Case Report

We report on two siblings, born 8 years apart to a Guatemalan consanguineous family, who presented with congenital skeletal malformations corresponding to Raine Syndrome. Assessment of the first patient upon birth revealed a cleft palate, hypoplasia of the orbital bone and exophthalmos, hypoplasia of the nose and a flat nasal bridge, as well as dysplastic ribs, and a dysplastic skull. The second patient also presented with the aforementioned anatomy, in addition to chonagal stenosia, a hypoplastic thorax, and a dysmorphic skull. Radiological findings for the second sibling include intracranial calcifications, in utero fractures scattered throughout the patient's body, and generalized osteosclerosis. DNA microarray testing showed significant homozygosity of area corresponding to the FAM20C locus on chromosome 7. Further analysis revealed a homozygous mutation in exon 5 of the FAM20C gene.

Discussion

Many of the described facial and radiological anomalies described in Raine Syndrome may often be erroneously attributed to other clinical pathologies that share similar findings. Osteopetrosis and Osteogenesis imperfecta are among diseases that come into differential diagnosis. It is thus important for clinicians to expand their repertoire of possible entities that may contribute to the described congenital skeletal malformations by including Raine Syndromes.

References