A RARE CAUSE OF HYPOPHOSPHATEMIC RICKETS; NON-LETHAL RAINE SYNDROME

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INTRODUCTION

Raine syndrome (RS) also known as lethal osteosclerotic bone dysplasia, is a rare autosomal recessive bone disorder. Most of patients with RS die within the first days or weeks of life due to pulmonary hypoplasia.

The causative gene *FAM20C* is located on chromosome 7p22.3. FAM20C is one of the genes that regulate phosphate production.

Here, we present a case of non-lethal RS with hypophosphatemic rickets and a new mutation in *FAM20C* gene.

CASE REPORT

A seven-year-old girl who had recurrent tooth extractions due to a dental disorder was referred to our clinic from the dental hospital. Her past history revealed that she had been operated for choanal atresia, cleft palate and atrial septal defect, and bowing of her legs after the age of one. According to his family history, our patient was the fifth child of consanguineous parents and his cousin had similar findings.

Physical examination at admission revealed a short stature girl (height sds -2.07) with facial dysmorphism (hypertelorism, exophthalmos, high palate, flattened nasal root, and anteverted ears), odontodysplasia, craniosynostosis, and o-bain deformity.

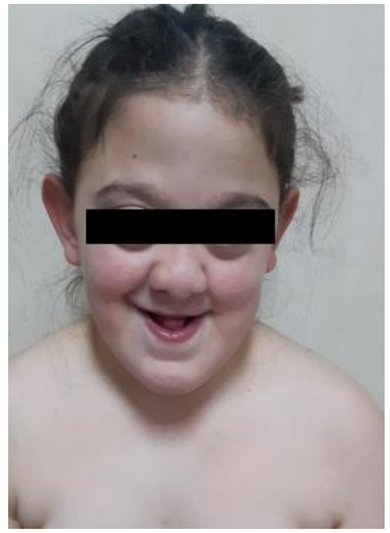


Figure: Facial appereance of the patient.

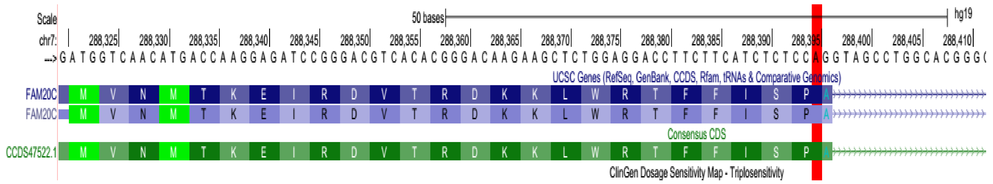
On laboratory; serum calcium level was 9.7mg/dL, phosphorus 2.7mg/dL, alkaline phosphatase 400U/L, parathormone 187ng/L, 25 (OH) D 24.2ng/mL. Tubular reabsorption of phosphate was 92%.

There was a minimal metaphysical fraying on x-ray of the knees, no apparent osteosclerosis was seen in the radiographs. Brain MRI showed a Chiari malformation.

Based on the clinical and laboratory findings, a diagnosis of hypophosphatemic rickets was made and oral phosphate and calcitriol treatments were started. Raine syndrome was considered due to her additional dysmorphic features.

GENETİC

WES analysis revealed a new mutation of FAM20C gene [NM_020223.3: c. 1071A>G;p. (pro557pro)] on exon 5.



CONCLUSIONS

The present case shows the patient with classical phenotypic features of Raine syndrome who had a new mutation of FAM20C gene.

The severity of symptoms varied significantly in RS. RS should be considered in the differential diagnosis of patients with hypophosphatemia and odontodysplasia. Little evidence of life expectancy and clinical outcome in non-lethal RS, however there are two reported patients with RS aged 61 and 72 years.

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