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Clinical Image

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Osteogenesis imperfecta XV: An ultra-rare genetic disease with neurological anomalies

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Clinical image description

A six-year-old Brazilian boy was born to non-consanguineous and healthy parents at 40 weeks of gestation via cesarean delivery. The family history is unremarkable. He had diagnosis of hydrocephalus since prenatal period and the treatment was the placement of a shunt at the first week of life. Because he had many bony fractures in 50 days of life, he became bisphosphonate therapy. The phenotype of osteogenesis imperfecta has been characterized by multiple fractures but the neurological abnormalities are unusual [1]. Intravenous bisphosphonate therapy is still the most widely used drug treatment approach [1,2].

We found significant neurological malformations similar it was seen in the literature [3]. Brain MRI revealed alterations in the supratentorial parenchyma with a diffuse alteration of the sulcation pattern, corpus callosum of difficult characterization, focal communication between the white matter of the cerebral hemispheres through the midline, reduction of the dimensions of the lateral ventricles, alteration of the signal in T2/FLAR in the supratentorial white matter in the periventricular regions with reduction of the parenchymal mantle, volumetric reduction of the brain stem, notably of the pons and signs that appear fusion of the thalamus with important reduction of the dimensions of the cerebellum. There is only a small remnant of the superior aspect of the cerebellar parenchyma. Only one artery was characterized in the topography of the anterior cerebral arteries, malformation of the basal ganglia, diffuse thinning of the optic nerves, and there was also a reduction in the thickness of the optic chiasm (Figures 1 and 2).

The clinical exome sequencing revealed homozygosis of pathologic variant mutation in exon 4 of the WNT1 gene. Homozygous or compound heterozygous mutations in WNT1 were recently described as a novel cause for severe autosomal-recessive OI [1-3]. Genetic counseling was made for the family. The child remains in clinical follow-up with severe neurological disabilities.

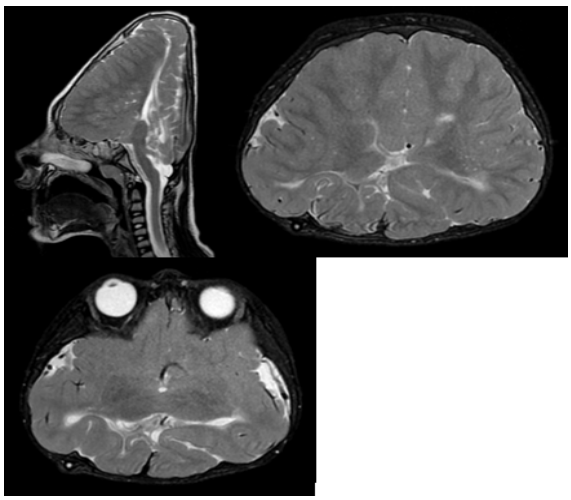

Figure 1: Only one artery was characterized in the topography of the anterior cerebral arteries, malformation of the basal ganglia.

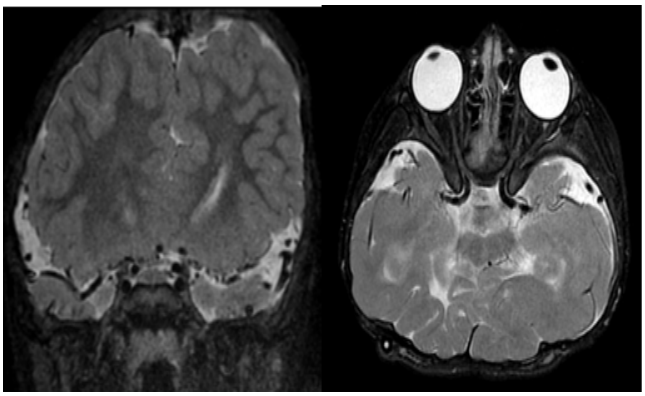

Figure 2: Diffuse thinning of the optic nerves, and there was also a reduction in the thickness of the optic chiasm.

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