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# SEPTO-OPTIC DYSPLASIA ASSOCIATED WITH MCARDLE DISEASE

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Introduction : Septo-optic dysplasia is a rare syndrome defined by any combination of optic nerve hypoplasia, pituitary hypoplasia and midline abnormalities of the brain such as agenesis of the corpus callosum and the septum pellucidum. McArdle disease is an uncommon entity. The diagnosis is establish by documentation of reduced phosphorylase activity in biopsied muscle. We described the association of this two rare entities in one girl.
Case report : A 13 month old girl of healthy non consaguineous parents was referred for evaluation because of left optic nerve hypoplasia. She was born weighting 3,6 Kg after an uncomplicated pregnancy. In the neonatal period she presented hypotonicity, jaundice and hypoglycaemia requiring treatment. At 13 month her weight was 9,3 Kg (P25-50) and height 77 cm (P75). Magnetic resonance imaging evaluation revealed left optic nerve hypoplasia and partial agenesis of septum pellucidum. The girl was identified to have central hypothyroidism and hipocortisolism. Insipidus diabetes was excluded. At four years old growth hormone failure was detected. At the age of twelve it was observed a persistent elevation of CPK ( 9360-20691 U/L). She was asymptomatic. Muscle biopsy was performed and McArdle disease was found.
Discusion : Septo-optic dysplasia´s genetic basis is unknown. Recently a missense mutation in the homebox gene Hesx1 was detected in one familial case. The gene of McArdle disease has been cloned and is located on chromosome 11. We ignore if it could exist some common genetic basis implicated in the pathogenesis of this two rare diseases