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EARLY OCULAR PHENOTYPE AND PROGRESSION OF COBALAMIN C DEFICIENCY: EXPERIENCE OF AN ITALIAN CENTRE.

Oral

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Purpose:

to report the ocular manifestations of cobalamin C (Cbl-C) deficiency, a rare autosomal recessive disease caused by mutations on the MMACHC gene.

Methods:

Clinical data from 8 children (4 females; median age at last available visit: 7,61 years; range: 1-11,32 years) with molecular diagnosis of Cbl-C deficit were retrospectively reviewed. Three of them were diagnosed at birth thanks to the extensive neonatal screening program, which became routinely performed since 2016. The others were diagnosed after the occurrence of hemolytic uremic syndrome. Color fundus photographs (CFP) and optical coherence tomography (OCT) images were collected and analyzed.

Results:

Nystagmus was present in 7 patients and 2 of them had high myopia (>6D). Visual acuity (available for 6 patients) was always ≤ 20/200 Snellen in the best eye. When available, CFP within the first month of age showed an altered foveal reflex, while a thinning of the outer retinal layers involving the fovea was already evident on OCT. At last visit (median follow-up: 2,8 years, range: 1-7,04 years), all children presented a central area of chorioretinal atrophy, which extended beyond the macula in 2 of them.

Conclusions:

Despite early diagnosis and treatment of Cbl-C deficit, ocular manifestations in these patients occur early and progress fast, confirming both developmental and degenerative natures of the phenotype. Visual prognosis is always poor and an early path of visual rehabilitation therapy and social support should always be considered.