133 Ocular manifestations of rare metabolic diseases
Centres of expertise for rare diseases

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Summary :
Although an accurate biochemical diagnosis is essential for the diagnosis of this group of diseases, systemic and ophthalmic findings are necessary to make us suspect their existence. The wide range of ocular findings shows how difficult it is to diagnose this group of diseases.

Text :
Abstract text
Introduction: Metabolic disorders are usually inherited autossomal recessive diseases in which there are an abnormal function of several enzymes in the cellular metabolic pathways, which are critical to normal cellular function, growth, and development. Ophthalmic manifestations sometimes are characteristic of the underlying metabolic disease and its detection is important for the correct diagnosis. Diseases associated with lipid metabolism have several types and subtypes with a very different clinical presentation.

Methods: The authors studied eleven patients with gangliosidosis, Krabbe disease and mannosidosis, who were referred to the Ophthalmology Department of Hospital São João. The possible existence of ocular manifestations was examined by means of biomicroscopic and fundoscopic examinations, OCT and electrophysiological examinations.

Results: These patients have a very wide spectrum of visual impairment and different degrees of neurological involvement. Even within the same disease there were differences in the clinical presentation. Fundoscopic examination ranges from normal to showing typical alterations with diffuse pallor. Electrophysiological alterations were detected in all patients. There was a case of congenital cataract.

Conclusions: This is a very rare group of diseases with distinct clinical presentation, usually associated with decreased visual acuity. Although an accurate biochemical diagnosis is essential for the diagnosis of this group of diseases, systemic and ophthalmic findings are necessary to make us suspect their existence. The wide range of ocular findings shows how difficult it is to diagnose this group of diseases.

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