76. EYELID TUMOR AND MICROCEPHALY IN HUNTER'S DISEASE

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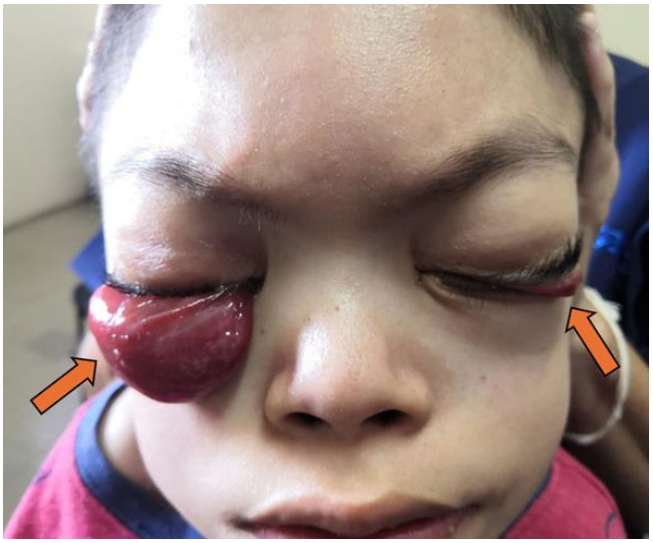
BACKGROUND: Hunter disease, or mucopolysaccharidosis type 2, is an X-linked inherited disease characterized by deficiency of the enzyme iduronate 2 sulfatase, which leads to accumulation of glycosaminoglycans. **THE CASE:** A 17-year-old male patient diagnosed with Hunter's disease at 15 months of age. He started his condition one year and three months earlier with edema in the right eye and inability to open it, which improved slightly with medical treatment. Eight months later, he came to our institution due to a tumor in both upper eyelids and microcephaly (Figure 1). Subsequently, surgical resection and external canthoplasty were performed. The surgical report showed pseudoepitheliomatous hyperplasia with no evidence of malignancy. Currently, the patient is stable with adequate healing and no visual sequelae. **CONCLUSION**: This condition usually manifests between two and four years of age, however, the most severe forms appear earlier. The most common ocular symptoms include glaucoma, retinopathy and hypertelorism, contrasting with the rare tumorization in both patient's eyelids. Although macrocephaly is common, the patient had microcephaly, a previously undocumented finding, therefore, to the best of our knowledge, it is the first reported case of a patient with microcephaly. Early identification and appropriate management are crucial to improve the quality of life and prognosis of these patients.

Figure: Protrusion of Both Upper Eyelids Due to Tumorization (Orange Arrows).



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