

PubMed

Format: Abstract

Full text links

JIMD Rep. 2017;35:17-22. doi: 10.1007/8904_2016_18. Epub 2016 Nov 18.



Intracranial Hypertension in Cystinosis Is a Challenge: Experience in a Children's Hospital.

Martín-Begué N¹, Alarcón S², Wolley-Dod C², Lara LE³, Madrid Á³, Cano P^{3,4}, Del Toro M^{3,5}, Ariceta G³.

Author information

Abstract

BACKGROUND: Cystinosis is a rare systemic lysosomal disease affecting mainly the kidney and eye. Ocular involvement in cystinosis is universal being the presence of cystine crystals in the cornea a diagnostic criterion and one of the earliest manifestations of the disease. Neuro-ophthalmologic manifestations are considered a rare and late complication in these patients. The aim of this article is to report the unexpectedly high incidence of intracranial hypertension in children with cystinosis at our centre.

METHODS: This study included eight children (0-16 years of age) with cystinosis seen at the paediatric ophthalmology department, Hospital Universitari Vall d'Hebron (Barcelona, Spain), a tertiary hospital, over the last 5 years.

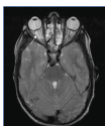
RESULTS: Three girls and five boys, mean age: 9.6 years (range: 5-14 years), were studied. During follow-up, 4 out of 8 developed papilledema and confirmed high cerebrospinal fluid (CSF) pressure. The only symptomatic child presented an Arnold-Chiari anomaly with enlarged ventricles, whereas the other three, all asymptomatic, were diagnosed by scheduled fundoscopy and had normal neuroimaging studies. All four patients had at least one known risk factor for developing intracranial hypertension: initiation of growth hormone therapy, tapering of corticosteroids, acute renal failure and Arnold-Chiari malformation. Two of them required a ventriculoperitoneal shunt.

CONCLUSIONS: Our results show that intracranial hypertension can occur more frequently than expected in patients with cystinosis. Furthermore, visual prognosis depends on early diagnosis and prompt treatment. A multidisciplinary approach is necessary, and we recommend fundoscopic examinations in all paediatric patients with cystinosis whether or not they present symptoms.

PMID: 27858370 PMCID: [PMC5585107](#) DOI: [10.1007/8904_2016_18](#)

[Free PMC Article](#)

Images from this publication. See all images (2) [Free text](#)



[LinkOut - more resources](#)