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Intracranial hypertension and papilloedema in a large cohort of pediatric alagille syndrome

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Abstract

Background: Ophthalmic abnormalities are amongst the 5 major criteria for diagnosing autosomal dominant Alagille Syndrome (AS). Embryotoxon, pseudo-papilledema and pigmentary retinopathy being most frequently part of the syndrome. Papilledema with or without intracranial hypertension (ICHT) are rarely described. We report 9 cases of bilateral papilloedema, 4 associated to ICHT at lumbar puncture (LP), all being diagnosed idiopathic ICHT in children with AS. Methods: We reviewed ophthalmologic examination data from 85 pediatric patients with clinically (n=48) and/or genetically (n=37) proven AS followed in two referring hospitals (UCL St Luc Brussels, n=75; HUG Geneva n=10). Patients were included if data were available before and after liver transplantation (LT). Results: 69 patients fulfilled inclusions criteria. 41 patients (41/69, 59%) of this group had LT of whom 2 developed true papilloedema before LT and 6 after LT (6/41, 14%). One papilloedema, but with normal neurologic exam and a rapid resolution was found in a non-transplanted child (1/28, 3%). Papilloedema resolved spontaneously in 4 patients (2 with pre-transplant onset) (4/69, 6%). In a fifth patient, severe papilloedema without documented ICHT (normal LP) resolved after switching tacrolimus to cyclosporine and adding acetazolamide. In only 4 remaining patients; the ICHT was revealed at LP. In all patients normal neurological exam and cerebral MRI were reported (4/69, 6%). ICHT was treated by steroids alone in one patient, and in the 3 others patients in combination with acetazolamide and a switch from tacrolimus to cyclosporine. Among these 3 patients, ventriculo-peritoneal derivation was ultimately required in 2 for severe progressive visual loss. We detected pseudo-papilloedema in 7 additional children (7/69, 10%); which was persistent and unchanged after LT in n=4. Conclusion: Besides pseudopapilloedema, true papilloedema with or without proven ICHT in a context of AS occurs in probably more patients than expected. A close follow-up of

ophthalmologic complications should be implemented for these patients before and/or after LT because the risk of severe and irreversible visual loss.

⬆ Drug terms

acetazolamide cyclosporine steroid tacrolimus

⬆ Disease terms

Alagille syndrome blindness complication **idiopathic intracranial hypertension** **papilledema**

⬆ Other terms

Brussels Capital Region child conference abstract diagnosis drug combination

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⬆ Additional information

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