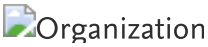

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Intracranial hypertension and papilledema in a large cohort of pediatric Alagille syndrome

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Abstract

Objectives and Study: Ophthalmic abnormalities are amongst the 5 major criteria for diagnosing autosomal dominant Alagille syndrome (AS), of which embryotoxon, pseudo-papilledema (PPO), and pigmentary retinopathy are the most common. Papilledema (PO), with or without intracranial hypertension (ICHT)/pseudotumor cerebri syndrome (PTCS), are rarely described. We here report 9 cases of bilateral PO in an AS cohort, 5 of whom were diagnosed with PTCS that occurred after liver transplantation. Methods: We reviewed ophthalmologic examination data from 85 pediatric patients with clinically (n = 48) and/or genetically (n = 37) proven AS, who were followed in 2 referral hospitals (UCL St Luc Brussels, n = 75; HUG Geneva, n = 10). Patients who received liver transplantation (LT) (n = 40) were compared with pediatric patients (n = 40) with LT for indications other than AS. Results: Sixty-nine patients fulfilled the inclusion criteria; the PO incidence in this cohort was 13% (9/69, 13%). Forty of these patients (40/69, 58%) underwent LT, of whom 2 developed true PO before LT and 6 developed PO after LT (6/40, 15%). One non-LT child (1/28, 3%) had PO, but had a normal neurological examination and rapid resolution. PO resolved spontaneously or remained stable without complications in 4 patients (2 with pre-transplant onset) (4/69, 6%). The remaining 5 patients had a normal neurological examination and cerebral magnetic resonance imaging findings (5/69, 7%). Probable PTCS was diagnosed in 1 patient and definite PCTS was diagnosed in 4 patients. PTCS was treated with steroids alone in 1 patient, and in combination with acetazolamide in 2 patients. In 2 of these 3 patients, ventriculo-peritoneal derivation was ultimately required for severe progressive visual loss. PPO was present in 10 other (10/69, 10%). One patient in the non-AS cohort had PO, yielding an incidence of 2.5% (1/40). Conclusions: True PTCS may be underdiagnosed in AS, but should be considered as a feature of this syndrome. Our findings supports the necessity of close follow-up of

ophthalmologic complications before and/or after LT, because of the risk of severe and irreversible loss of vision.

Drug terms

acetazolamide steroid

Disease terms

Alagille syndrome blindness **brain pseudotumor** complication **papilledema** retinitis pigmentosa

Other terms

Brussels Capital Region child cohort analysis conference abstract diagnosis drug combination drug therapy female follow up human incidence liver transplantation major clinical study male multicenter study neurologic examination nuclear magnetic resonance imaging ophthalmology patient referral pediatric patient peritoneum remission surgery

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