Although historically conceptualized as driven by sporadic genetic events, a growing body of literature supports the possibility of an inherited predisposition.

Systematic screening for Familial colloid cysts may facilitate early recognition and treatment of indolent cysts, thereby preventing the rapid deterioration that can occur with an unrecognized third ventricular tumor. Furthermore, identifying a transmission pattern may yield more insight into the molecular and genetic underpinnings of colloid cysts [1)].

Case reports

At 20 months of age, a Caucasian girl with trisomy 9 and family history of an older brother and twin sister having the same syndrome displayed sign of congenital hydrocephalus due to increasing head circumference. Magnetic resonance imaging revealed enlarged lateral ventricles and a prominent choroid plexus, and the girl was treated with a ventriculoperitoneal shunt, which 2 days later had to be replaced with a ventriculoatrial shunt as cerebrospinal fluid formation greatly exceeded the ability of the patient's abdominal absorptive capability. At 16 years of age, the patient was diagnosed with cardiomyopathy and diminished ejection fraction. Some months later, she was admitted to the neurosurgical ward showing signs of shunt dysfunction due to a colloid cyst in the third ventricle. Cystic drainage through endoscopic puncture only helped temporarily. Revision of the shunt system showed occlusion of the ventricular drain, and replacement was merely temporary alleviating. Intracerebral pressure was significantly increased at around 30 mmHg, prompting externalization of the drain, and measurements revealed high cerebrospinal fluid production of 60-100 ml liquor per hour. Thus, endoscopic choroid plexus coagulation was performed bilaterally leading to an immediate decrease of daily cerebrospinal fluid formation to 20-30 ml liquor per hour, and these values where stabilized by pharmaceutical treatment with acetazolamide 100 mg/kg/day and furosemide 1 mg/kg/day. Subsequently a ventriculoperitoneal shunt was placed. Follow-up after 1 and 2 months displayed no signs of hydrocephalus or ascites.

High cerebrospinal fluid volume load and coexisting heart disease in children with trisomy 9p may call for endoscopic choroid plexus coagulation and pharmacological therapy to diminish the daily cerebrospinal fluid production to volumes that allow proper ventriculoperitoneal shunting [2)].

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