

Trisomy 9p

Hydrocephalus caused by excessive **liquor** production due to **choroid plexus hyperplasia** is a rare condition that may necessitate unusual treatment paradigms. It can be seen in **trisomy 9p** where coexisting **congenital heart disease** additionally may complicate the therapeutic approach.

Case report

At 20 months of age, a **Caucasian** girl with **trisomy 9** and a **family history** of an older brother and **twin** sister having the same syndrome displayed signs 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 production** 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 drainage**, and **replacement** was merely temporary alleviating. **Intracranial 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 were 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** ¹⁾.

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Henningsen MB, Gulisano HA, Bjarkam CR. **Congenital hydrocephalus** in a **trisomy 9p** gained **child**: a **case report**. J Med Case Rep. 2022 May 27;16(1):206. doi: 10.1186/s13256-022-03424-5. PMID: 35619116.

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