Robinow syndrome

Robinow syndrome is a rare entity with a characteristic appearance, such as hypertelorism, short stature, mesomelic shortening of the limbs, hypoplastic genitalia, and rib as well as vertebral anomalies. We had treated a patient with Robinow syndrome who developed hydrocephalus and craniosynostosis which is not usually associated.

Case presentation: The ventricle enlargement was detected during pregnancy in a female infant. She did not develop hydrocephalus just after birth. Her facial appearance was fetus-like, so the pediatricians had suspected Robinow syndrome. During follow-up examinations, a rapidly enlarging head circumference was detected when she was 3 months old. Her conscious level was not disturbed, but she had a tight fontanel and sunset phenomenon was recognized. Hydrocephalus was diagnosed by radiographic imaging so that she underwent ventriculo-peritoneal shunting (VPS). Her irregular head enlargement seized. Six months after surgery, her parents noticed the brachycephalic shape of her head. A computed tomography (CT) and magnetic resonance (MR) scan were conducted and showed that her bilateral coronal, bilateral lambdoid, and the sagittal suture were fused in addition with a tonsillar herniation. Since the sutures were not remaining, we diagnosed that this was a primary pan synostosis rather than secondary craniosynostosis due to VPS. Posterior cranial vault distraction with foramen magnum decompression (FMD) was conducted. The distractor was extended by 1 mm per day up to 30 mm. After a consolidation period of 2 months, the distractors were removed. Through this intervention, a 15.4% increase (+196cc) of the intracranial space with an improvement of the chronic tonsillar herniation was achieved.

Conclusion: To confirm the diagnosis of Robinow syndrome, a genetic test was conducted. The analysis showed ROR2 Exon3 (c233 c>t p. Thr 78 Met), which is found in the recessive type of Robinow syndrome. We report this patient as, to our best knowledge, the first case documented case of Robinow disease presenting with hydrocephalus and craniosynostosis. Posterior cranial vault distraction with FMD is a useful way to treat this condition [1)].

A 16-year-old female with osteosclerotic Robinow syndrome complicated by slit ventricle syndrome presented with refractory intracranial hypertension following external ventricular drain placement. Symptoms included severe headaches and altered mental status. Given the severe intracranial volume restriction secondary to massive calvarial thickening (2.5 cm), the patient was taken to the operating room for urgent surgical decompression. After frontal and parietal craniectomy, burr and osteotome contouring were used to remove two-thirds of the endocranial calvarial bone flap thickness resulting in a 9% cranial vault expansion while preserving an overall normal head size. There were no immediate postoperative complications. At over 3 years postoperatively, the patient had reduced headaches, maintained adequate shunt function, and has not required further vault reconstruction [2)].

A 13-year-old girl with Robinow syndrome presented with multiple transient ischemic attacks consisting of speech arrest and generalized weakness. Evaluation revealed probable bilateral moyamoya syndrome. Initially, she was treated conservatively due to the rarity of her symptoms and relatively unremarkable angiographic findings. However, she ultimately had progression of her disease both clinically and radiographically, leading to surgical management. She underwent staged bilateral pial synangiosis without complication and has done well subsequently.

Conclusion: The potential difficulty of detecting symptoms related to cerebral ischemia in the Robinow's population - where cognitive impairment may obscure initial neurologic symptoms - can result in delayed diagnosis and treatment. Given the excellent outcomes of moyamoya patients when treated prior to the development of fixed neurologic deficits, case reports such as this and identifying syndromic associations serve to highlight conditions that may result in improved patient outcomes through earlier diagnosis and treatment. The clinical and radiographic features of moyamoya syndrome associated with Robinow syndrome seem comparable to those of primary moyamoya disease. The presence of moyamoya syndrome should be considered in the evaluation of patients with Robinow syndrome who present with transient ischemic attack-like symptoms [3].

1)
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