Unusual association of diseases/symptoms

Cleft palate, arachnoid cyst and posterior urethral valve

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Summary

A 5-month-old infant presented with cleft palate, febrile urinary tract infection and abnormal movements. His urinary stream was poor and investigations revealed posterior urethral valve and grade V vesicoureteric reflux. This baby was investigated for seizure activity and on MRI of the brain was found to have a large arachnoid cyst, splaying the occipital horns and compressing the posterior aspect of third ventricle leading to dilatation of the ventricles.

This seems to be a very unusual coexistence of three abnormalities in one patient for which no plausible explanation can be given. It appears to be a chance finding.

BACKGROUND

An unusual association of rather common abnormalities of cleft palate and posterior urethral valve (PUV) with a relatively uncommon finding of arachnoid cyst prompted us to publish this case report. This case may help progress in the delineation of unknown multiple congenital anomalies entities.

Arachnoid cysts are common in the middle cranial fossa and are usually asymptomatic; this infant had an arachnoid cyst in the posterior fossa and was symptomatic.

In children with PUV, the unilateral vesicoureteric reflux (VUR) is on left side in 92%, while in this infant the reflux was on right side.

CASE PRESENTATION

A 5-month-old baby boy presented to us with an intermittent febrile illness of 5 days. This baby had had a poor urinary stream since birth. The parents gave a history of abnormal movements of 5–10 min duration, involving all four limbs, associated with the rolling up of the eyes and urinary and faecal incontinence. These attacks occurred frequently and used to subside spontaneously.

Born to non-consanguineous parents, the antenatal and birth history were normal. There was no family history of seizure activity. Developmental milestones were normal and the immunisation status was appropriate for the age.

Figure 1 Cleft palate.

Figure 2 Hypertrophied bladder, dilated posterior urethra.

The infant's weight, height and head circumference were 3.88 kg, 56 cm and 37 cm, all of which were below the third percentile. This infant had a cleft palate and low-set ears, but no other dysmorphism (figure 1). Physical examination was unremarkable except for a palpable urinary bladder.

INVESTIGATIONS

Haemoglobin, complete blood count, serum sodium, potassium, urea, creatinine and cerebrospinal fluid analysis were within normal limits. Urine routine examination showed plenty of pus cells and the urine culture grew *Escherichia coli*. The colony count was more than 100 000 colony forming units/ml. Antenatal sonography was not performed.

Ultrasonography revealed bilateral hydronephrosis, hydroureters and thickened bladder wall. Both the kidneys were of normal size and echotexture. The micturating cystourethrogram showed grade V VUR on right side and dilated posterior urethra (figures 2 and 3).

MRI of the brain showed large lobulated cystic lesion in both the occipital lobes, mainly on the left side (the dimensions 6.3×4.8×4.6 cm), crossing the midline, leading to splaying of the occipital horns and compressing the posterior aspect of third ventricle causing moderate ventricular dilatation (figures 4 and 5).

TREATMENT

The infant has been catheterised with an infant feeding tube. After control of urosepsis, endoscopic fulguration of PUV and endoscopic surgery for the arachnoid cyst are being considered.

DISCUSSION

A search of the literature for association of cleft palate and/or PUV with arachnoid cyst did not yield any results.

PUVs are the most common form of urethral obstruction in boys.¹ It is part of the spectrum of obstructive congenital anomalies of the urinary tract. The urethral valves are tissue leaflets fanning distally from the prostatic urethra to the external urinary sphincter. A slit-like opening usually separates the leaflets. Valves are of unclear embryological origin and cause varying degrees of obstruction. Approximately 30% of patients experience end-stage renal disease or chronic renal insufficiency. The prostatic urethra dilates and the bladder muscle undergoes hypertrophy. VUR occurs in 50% of patients.²

There are a wide range of renal changes from mild hydronephrosis to severe renal dysplasia; their severity probably decided by the severity of the obstruction and its time of onset. As in other cases of obstruction or renal dysplasia, there may be oligohydramnios and pulmonary hypoplasia, these two being very important poor prognostic factors.

Affected boys with PUVs are discovered prenatally when maternal ultrasonography reveals bilateral hydronephrosis, a distended bladder and, if the obstruction is severe, oligohydramnios.

Prenatally diagnosed PUVs, particularly when discovered in the second trimester, carry a poorer prognosis than those detected after birth. In the male neonate, PUVs are suspected when there is a palpably distended bladder and the urinary stream is weak. If the obstruction is severe and goes unrecognised during the neonatal period, infants may present later in life with failure to thrive due to uraemia or sepsis caused by infection in the obstructed urinary tract. With lesser degrees of obstruction, children present later in life with difficulty in achieving diurnal urinary continence or with urinary tract infection.

Cystoscopy is considered to be the reference standard for determining the presence of a urethral obstruction. It is

Figure 3 Grade V vesicoureteric reflux (VUR; right side).

supposed to be more reliable than voiding cystourethrography and even more reliable than urodynamic studies.³

The relationship between valves, reflux and dysplasia has been identified. In comparing refluxing and non-refluxing kidneys, it was recognised that infants with unilateral reflux

had markedly different renal function. Reflux was consistently associated with dysplastic and damaged kidneys. It was postulated that the reflux allowed the high bladder pressures to focus on the refluxing kidney, sparing and protecting the non-refluxing kidney. The vesicoureteral reflux

Figure 4 Bilobed arachnoid cyst.

Figure 5 T2-weighted MRI showing arachnoid cyst and dilatation of ventricle.

and dysplasia relationship is found in 13% of patients with valve issues, and interestingly, the refluxing kidney is on the left in 92%.⁴

The proposed mechanism of this protection is that the refluxing collecting system acts as a pressure pop-off valve. It is thought that the protected kidney would ultimately lead to a better long-term prognosis than for boys without unilateral reflux.⁵

Arachnoid cysts are commonly encountered fluid collections in the central nervous system. These are more common in men, as was the case in our patient too. Reporting a series of 20 cases, the authors describe a case series that illustrates the diverse forms of presentation and the treatment modalities commonly used for arachnoid cysts. A total of 70% of the cysts were supratentorial, 5% infratentorial and 25% spinal. Symptoms at presentation included headache (41%), weakness (23%), seizure (14%), hydrocephalus (9%), scoliosis (4%), cognitive decline (4%) and visual loss (4%).⁶

Primary arachnoid cysts are present at birth and are the result of developmental abnormalities in the brain and spinal cord that arise during the early weeks of gestation. Secondary arachnoid cysts are not as common as primary cysts and develop as a result of head injury, meningitis, or tumours, or as a complication of brain surgery. The majority of arachnoid cysts form in the middle crania fossa. Arachnoid cysts involving the spinal cord are more rare. The location and size of the cyst determine the symptoms and when those symptoms begin. Most individuals with arachnoid cysts develop symptoms before the age of 20, and especially during the first year of life, but some people with arachnoid cysts never have symptoms. Males are four times more likely to have arachnoid cysts than females.

Symptomatic arachnoid cyst may be treated by fenestration or cystoperitoneal shunt. Asymptomatic arachnoid cyst may be watched, or drained if expansion or haemorrhage would threaten vital structures.

Learning points

- ▶ posterior urethral valve (PUV) is part of the spectrum of obstructive congenital anomalies of urinary tract with protean presentations.
- ▶ Treatment needs to be tailored to the individual.
- ▶ Arachnoid cyst may be primary or secondary. These are more common in males.
- ▶ Asymptomatic cysts may not need any treatment, whereas symptomatic cysts will need either fenestration or shunting.
- ▶ These two conditions with cleft palate may be associated as a chance finding. This case may help progress in the delineation of unknown multiple congenital anomalies entities.

Competing interests None.

Patient consent Obtained.

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