Case Number 10
Dandy-Walker Syndrome

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Reviewed by: Dr. Stephen Attard

Case summary:

Demographic details:
SA, female.
Aged 8.
Resident in Pembroke.

Ms. SA is an 8-year-old known case of Dandy-Walker syndrome managed through supraventricular and infraventricular peritoneal shunts. Having acquired normal developmental milestones till seven years of age, she currently has cognitive impairment, gait abnormality and speech defects following shunt complications.

Presenting complaint:

Cognitive Impairment
Speech Deficit
Locomotor Disturbance

History of presenting complaint:

Ms. SA is a known case of Dandy-Walker syndrome diagnosed at two years of age on the 8th January 2007 following investigation of supposed meningitis. MRI studies revealed the presence Dandy-Walker cyst malformation which was responsible for raised intra-cranial pressure. A supraventriculo-peritoneal shunt was inserted via a right occipital bore-hole.

Ms. SA had reached developmental milestones besides delayed speech development and a clumsy gait. She was described as an intelligent girl and was coping with the educational activities encountered in primary school.

The patient progressed to suffer multiple episodes of shunt failure typically following an upper respiratory tract infection. She frequently presented with fronto-occipital headaches that would wake her up during the night, being exacerbated when lying down and associated with multiple episodes of vomiting and irritability. The first such episode was on 28th February 2008, and a second episode took place on 2nd February 2012. In each case the cause was due to secondary raised intracranial pressure following shunt obstruction and required surgical revision.

On the 6th April 2012 Ms. SA at 7 years of age presented to casualty with a presentation similar to previous episodes of shunt failure but also had an episode of a generalised tonic-clonic seizure. On examination the patient was neurologically intact and had a negative Kernig's sign and no neck stiffness. On fundoscopy bilateral papilloedema was found, being greater on the left side. The shunt was also tense and indicated failure of drainage, dilated ventricular systems were subsequently revealed through CT scanning. A new shunt was inserted to drain the Dandy-Walker cyst through a right parietal borehole which was connected with the supra-ventricular shunt through a Y-connector.

Following the procedure Ms. SA suffered cardiac arrest a total of three times requiring cardiopulmonary resuscitation and developed acute respiratory distress syndrome requiring ITU admission. Following this she went into a coma lasting six weeks, where upon returning to consciousness she was found to have cognitive impairment, loss of speech ability and an impaired gait.

Ms. SA was seen on Wednesday 26th March 2014 at specialist neurosurgical review wherein her gait and speech was assessed. She had a fourth shunt failure in December 2013 with no complications which resulted in revision of the ventriculo-peritoneal shunts. Her speech and gait have greatly improved and she is now capable of conversation but has significant cognitive impairments. She is not able to stand independently but is able to walk with assistance of devices.

Past medical and surgical history:

Past Medical History

- Diagnosed Pulmonary Stenosis at birth with several cyanotic spells requiring admission to SCBU
- Coarctation of the Aorta
- Dandy-Walker Syndrome
- Double kidney on the right diagnosed on 10/01/07
- Missing left little toe and aplasia of her left little finger noted at birth
- 30% bilateral sensorineural hearing loss treated with hearing aids
- Cleft soft palate

Past Surgical History

- Ventriculo-peritoneal shunt first done on the 8/01/07. Revisions of the shunt carried out on 6/14/12 and 28/02/12, 13/12/13
- Adenoidectomy plus grommet insertion February 2008

Drug history:

Patient is currently on no regular therapy and does not suffer from any known drug allergies.

Family history:

No known family history of Dandy-Walker syndrome or any other congenital malformations.

Social history:

Ms. SA is an only child currently residing with her mother and grandfather in Pembroke. She currently attends primary school with the aid of a Learning Support Assistant.

Systemic inquiry:

- General Health: Patient looks well with no recent weight loss.
- Cardiovascular System: No chest pain or palpitations.
- Respiratory System: No cough, wheeze or shortness of breath. The child complains of early morning drowsiness and tiredness through the day with episodes of gasping during sleep indicative of sleep apnoea.
- Gastrointestinal System: No dysphagia, no abdominal pain, slight constipation, no diarrhoea.
- Genitourinary System: No urinary frequency or dysuria.

- Central Nervous System: Suffers from occasional frontal headaches responding to paracetamol.
- Musculoskeletal System: Nil of note.
- Endocrine System: Nil of note.

Current therapy:

The patient currently has multiple ventriculo-peritoneal shunts for the management of Dandy-Walker Syndrome. She also regularly attends physiotherapy and speech therapy.

Discussion of results of general and specific examinations:

The patient has visible facial dysmorphic features with close set eyes, low lying ears and a beaked nose; a missing left little finger was also noted, bilateral hearing aids were also present. On cardiorespiratory examination a loud S2 was noted with no systolic or diastolic murmurs, slight radiofemoral delay was also found.

Ms. SA showed excellent neurological progress from previous examinations with good cognition and improved speech. Her gait is slightly ataxic and she is able to walk with assistance, otherwise being neurologically intact.

Differential diagnosis:

Differentiation from other posterior fossa cystic lesions such as mega cisterna magna and retrocerebellar arachnoid cyst is straightforward through the use of standard neuroimaging. The position of the choroid plexus in the fourth ventricle helps in differential since this is normal with an arachnoid cyst, displaced into the superior cyst wall with a Blake pouch, and absent in Dandy-Walker malformation.

Diagnostic procedures:

Laboratory exams:

Karyotype done on 21/2/2012 showing 46 XX – normal female karyotype

Imaging:

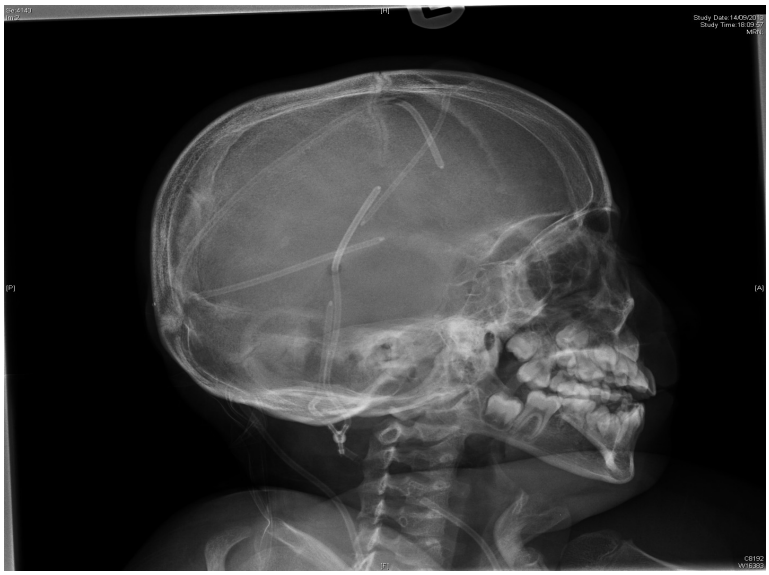

Figure 1: Lateral View Shuntogram

Description of Image

This is a lateral skull X-ray indicating a total of four shunts. Note the Y-connector and valve positioned inferior to skull base and posterior to the vertebral column on the radiograph.

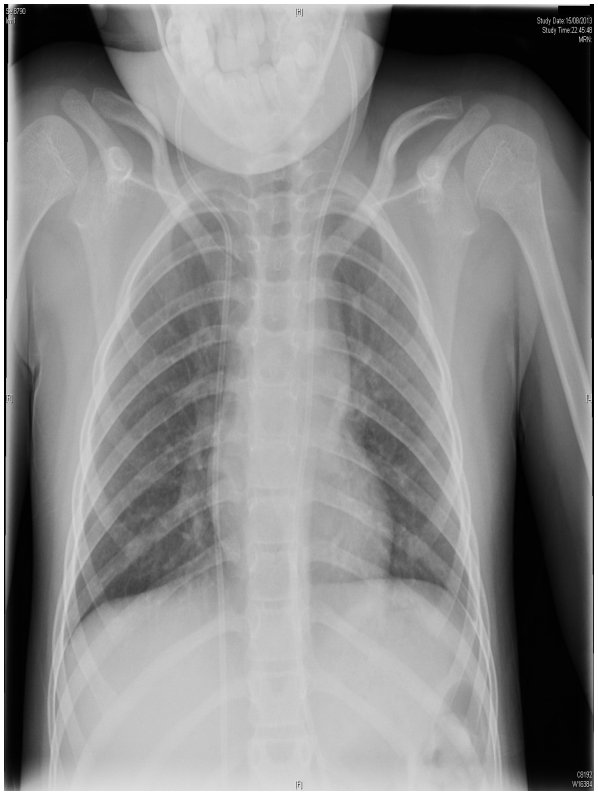

Figure 2: PA Chest X-ray

Description of Image

The above is a posteroanterior chest X-ray of the patient exhibiting the ventriculo-peritoneal shunts.

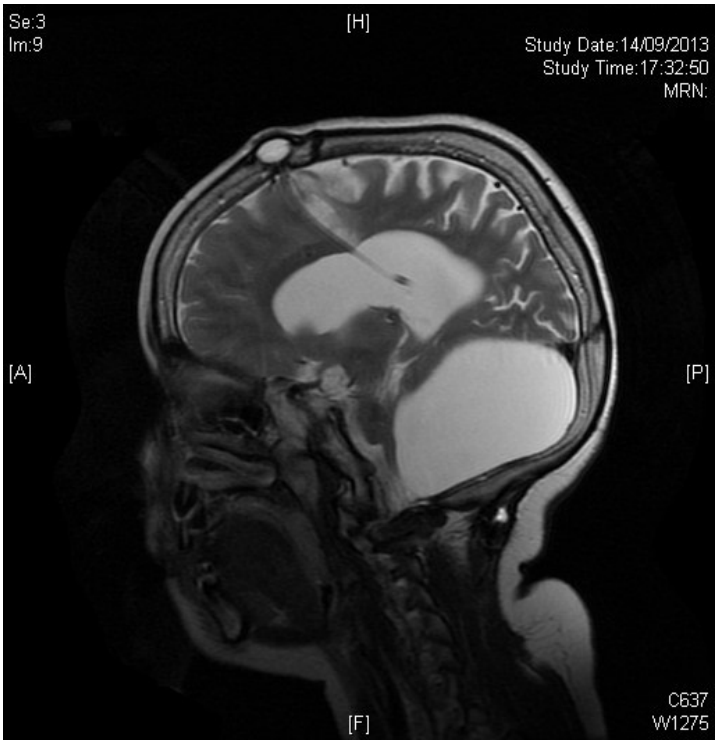

Figure 3: Sagittal T2 weight MRI study

Description of Image

This is a sagittal T2-weighted image. A fluid filled cyst is present in the posterior cranial fossa with superior displacement of the tentorium cerebelli and hypoplasia of the cerebellar vermis consistent with Dandy-Walker syndrome. There is also ventriculomegaly and visible supratentorial shunt.

Diagnosis:

Dandy-Walker Malformation with multiple ventriculoperitoneal shunts.

Final treatment and follow-ups:

To be reviewed at Child Outpatients in six months' time.

Fact Box 10:

Title: Dandy-Walker Syndrome

Brief Overview: The Dandy-Walker syndrome refers to a rare congenital malformation characterised by agenesis or hypoplasia of the cerebellar vermis, cystic dilatation of the fourth ventricle and an enlarged posterior fossa[1]. The newborn infant may be symptomatic or asymptomatic with the majority of cases presenting with a macrocephaly and raised intracranial pressure[2-3]. Around 80% are diagnosed at infancy, whereas others are identified in adulthood[2-5]. The aetiology is not well understood but is probably multifactorial. In most cases the syndrome is isolated but it may also be associated with defined Mendelian disorders, chromosomal abnormalities, and other syndromes such as Ritscher-Schinzel, Hydrolethalus, and Marden-Walker[6-12].

Epidemiology: Osenbach and Menezes, estimated that isolated Dandy-Walker Syndrome has a prevalence of about 1 per 30,000 live births and a study by Hirsch revealed that it accounts for about 2-4% of infantile hydrocephalus[4,13]. A study by Long et al., involving a population based survey of the foetal posterior fossa anomalies found an incidence of about 1 per 11,000[14].

Symptoms and signs:[2-3,15]

- macrocephaly
- symptoms of raised intracranial pressure such as lethargy, vomiting, and irritability
- occipital encephalocoele (unusual presentation)
- apneoa (unusual presentation)
- nystagmus
- motor deficits
- intellectual impairment

Investigations:[12, 16-17]

- Magnetic Resonance Imaging (MRI) is the preferred investigation for detailed imaging.
- Ultrasound is useful in prenatal diagnosis which may reveal an enlarged fourth ventricle at 14-16 weeks gestation as a transient phenomenon and therefore allows diagnosis close to 20 weeks gestation.

Treatment: If progressive hydrocephalus is evident, the current treatment of choice involves various techniques of shunt placement. Some specialists prefer an initial attempt with a standard ventriculo-peritoneal shunt, others prefer a cystoperitoneal derivations, and other recommend combined shunting of the cyst and the lateral ventricles[4,13,18]. Physiotherapy is also an important component of management to ensure optimal psychomotor development.

Prognosis: Survival and outcome are largely dependent on other associated findings, such as cardiac anomalies and other central nervous system malformations. Also, preservation of vermian lobulation and the absence of supratentorial anomalies favour a better intellectual prognosis[19-20]. Shunt dysfunction is a source of complication but sudden unexpected deaths unrelated to shunt problems have also been reported[21].

References:

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