CASE REPORT

'Mutiny on the crown': two cases of rare cephalic malformations

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SUMMARY

The presentation and management of two bizarre congenital cephalic curiosities at the two extremes of the clinical spectrum are discussed herewith. Case 1: presented to us as a neonate with a scalp swelling mirror-imaging her head and face. The journey from clinics to wards and to the operation theatre and to her home is introspected. Case 2: presented to us as dicephalous dibrachius dipus parapagus conjoined twins. The detailed work-up of individual organ systems, the multidisciplinary approach to management and the final outcome are discussed. This is an unsolved mystery for the anatomists, paediatric surgeons, radiologists and the medical fraternity at large.

BACKGROUND

Congenital cephalic malformations are a common presentation in infants and pose a diagnostic dilemma. They can range from the usual swellings like encephaloceles, nasal gliomas, lipomas, vascular malformations and dermoid to the rare birth defects like aplasia cutis cerebri.¹ We report two cases of bizarre cephalic malformations at the two extremes of clinical spectrum. One turned out to be a benign neoplasm while the other turned out to be a rare congenital birth defect. While one was operable and salvageable, the other was inoperable. However, both the cases represent in true sense medical curiosities and leave behind some food for thought.

CASE PRESENTATION: 1**Case 1: huge congenital benign cystic teratoma**

A 2-day-old baby girl presented to us with an antenatally diagnosed huge cystic lesion in the region of the head. On examination, the general condition of the child was normal. She had an asymptomatic but huge (12 cm × 14 cm) right-sided craniofacial cystic swelling distorting the right half of the face and scalp and disfiguring the right eye. The overlying skin was thin and stretched out with prominent scalp veins. The child received initial supportive management and was started on nasogastric feeds.

INVESTIGATIONS

A bedside ultrasound (USG) was suggestive of a huge cystic scalp swelling; however, its extent and the intracranial extension were confirmed on contrast-enhanced CT scan. CT showed a huge homogenous cystic swelling in the right craniofacial region with thinning and compression of the

underlying crania. The swelling was closely related to the right orbital structures, and intracranial extension was doubtful. No hydrocephalus was seen. The probable diagnosis was a scalp lymphangioma.

DIFFERENTIAL DIAGNOSIS

However, an atypical encephalocele or a scalp teratoma was the differentials. After proper informed consent from the parents, a multidisciplinary team including neonatologist, paediatric surgeon, paediatric neurosurgeon, ophthalmologist, paediatric radiologist and paediatric anaesthetists planned the strategy to debulk this Hercules (figure 1A–C).

TREATMENT

Intraoperative findings revealed a 10×10 cm cystic lesion in the right temporo-parieto-facial region, extending into the neck. Meticulous dissection led to an intact plane between the sac wall and the periosteum, and the tumour was completely removed. However there was no underlying skull bone found. The swelling contained approximately 500 mL of clear serous fluid. Fortunately, there was no intracranial or intraorbital extension. Skin flaps were mobilised to cover the huge defect. Child received postoperative care in the neonatal surgical intensive care unit (ICU). Flap necrosis in the postoperative period was managed conservatively. She was discharged on postoperative day 14 on full oral feeds (figure 2A,B).

OUTCOME AND FOLLOW-UP

What followed was a diagnostic surprise. The histopathologists reported it as a mature cystic teratoma containing elements originating from all three germinal layers (figure 3A,B). The postoperative tumour markers were normal for age. The child was followed up closely until 48 months. There was spontaneous reshaping of the orbital cavity, facial and the skull bones noticed. However, deficiency of the right-sided facial musculature and asymmetry of the face and eyes persisted.

DISCUSSION

Congenital tumours are defined as those presenting within first 60 days of life and account for 0.5%–1.9% of all childhood tumours.² About 50% of the congenital tumours are germ cell tumours (GCT).³ Extragonadal GCTs commonly occur in the sacro-coccygeal region followed by anterior mediastinum, pineal gland, retroperitoneum and, less commonly, the neck, stomach and vagina.⁴ The nasopharynx,

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Figure 1 (A) Neonate with a huge congenital cystic swelling in the right craniofacial region. (B,C) Axial and coronal cuts of the CECT scan showing the extent of swelling and compression of the underlying grey matter.

oral cavity, orbit and cervicothyroidal region are the sites of extragonadal head-neck teratomas.⁵ Congenital cranial teratomas are commonly intracranial in location with some extracranial extension and are usually associated with hydrocephalus.^{6,7} Paediatric scalp teratomas have rarely been reported in literature and are of dismal prognosis.^{4,8,9} They are usually lethal because of their potential for rapid growth, the mass effect, extensive destruction and involvement of the intracranial components, and airway obstruction and respiratory compromise.^{6,10-14} Ours is a rare case of a congenital extracranial craniofacial mature cystic teratoma.

Teratomas appear to arise from diploid cells. There are different theories proposed for the teratoma development. According to one, some of the embryonic germ cells fail to reach the gonadal ridge and become misplaced or migrate aberrantly. These ectopic germ cells are usually embedded in or near midline structures giving rise to teratoma formation later. Another theory is based on the principles of cloning and suggests teratoma formation from a somatic cell. The premise is that because every cell contains the full genetic code, theoretically, any somatic cell, without being a 'germ' cell, could produce any other type of cell. The third theory is that teratomas are a result of incomplete twinning.^{4,7,14,15}

Histology reveals that teratomas contain derivatives of all the three germ layers. Mature teratomas are composed of well-differentiated elements, such as mature cartilage, squamous epithelium, skin appendages, columnar mucosa and neural elements. They are usually benign and are more common in females. Immature teratomas contain primitive elements derived from any or all of the three germinal layers. They are typically malignant and

Figure 2 Intraoperative picture showing complete removal of the swelling.

Figure 3 (A) Gross histology of the specimen. (B) Elements from all three germ layers suggestive of a mature cystic teratoma.

more common in males. Congenital teratomas are mostly benign and show histology akin to that described in our case.^{9,14}

For benign teratomas, a complete excision is the treatment of choice. However, 25% of the neonates die before surgery and hence the dismal prognosis.^{3,8,15} A complete surgical resection is important to prevent recurrences and malignant transformation in a residual tumour. As was noticed in our case, a large cystic teratoma usually has a pseudocapsule that aids in its dissection. Hence, an early decompression is not advisable unless its size hinders proper visualisation. Also, the overlying stretched skin flaps should be preserved to provide cover to the large defect that is left behind. A follow-up with histology, tumour markers and imaging is essential. Spontaneous craniofacial remodelling occurred in most reported cases; a few, however, may later require facial reconstruction from a cosmetic standpoint.¹³

CASE PRESENTATION: 2

Case 2: dicephalous dibrachius dipus parapagus conjoined twins

A 9-hour-old set of female conjoined twins with two heads mirroring each other, a shared trunk, two arms and two legs were referred to our hospital for further management. They were antenatally diagnosed at 8 months of gestation, delivered by caesarean section at term and weighed 3.8 kg at birth. Twins were a product of consanguineous marriage of a 27-year-old multipara. The mother had a 4-year-old son through a previously healthy pregnancy. She had no history of abortion, infertility, medical or drug history, any harmful radiation exposure or significant illness during her present pregnancy. However due to poor socioeconomic background, she was not a booked case and had no previous antenatal USG done. There was no family history of twinning.

On examination, the twins were pink and active, with normal facies and no facial deformities. Their heads were normocephalic with short necks connected to a shared torso. They had two arms, two legs and a normal female-like genitalia with three perineal openings. The HR was 138/min, and breaths sounds were normal; however occasional mild chest retractions were noticed on the left side. The oxygen saturation on the right was 87% and on the left side was 89%. Two spines were palpable, but they seem to fuse at the sacrum. The twins had passed urine and meconium. All extremities moved spontaneously. However, painful stimuli to the right side evoked the right facial cry and vice versa. Also, sometimes the right was active, while the left head was asleep (figure 4A).

Figure 4 (A) Dicephalus dibrachius dipus parapagus conjoined twins. (B) Skygram showing two vertebral columns, single pelvis and no limb defects, enlarged retrosternal cardiac shadow and a liver shadow in both the hypochondria.

INVESTIGATIONS

Supportive treatment was started, and a detailed work-up was planned for further management of the twins. This involved a dedicated multispecialty team consisting of paediatric surgeons, paediatricians, neonatologist, anaesthetists, paediatric radiologist, paediatric cardiologist, paediatric neurologist and surgical neonatal ICU staff. A detailed systemic work-up was planned for further management.

Routine blood investigations were all between normal limits. Plain radiographs showed two skulls, two vertebral columns with no vertebral defects, single pelvis with gas reaching up until the perineum, clear bilateral lung fields, an enlarged retrosternal cardiac shadow and no bony abnormalities of any limb (figure 4B). An abdominal USG revealed two livers probably fused in midline, two normal kidneys and a single urinary bladder. A more detailed systemic evaluation warranted a MRI. Our hospital team geared up to this challenge and arranged for difficult intubation of both these twins in the operation theatre, followed by their transfer to the MRI suite equipped with twin anaesthesia machines. MRI whole body including MR urography, echocardiography, CT angiography, HIDA scan and oral contrast dye study unveiled slowly the mystery of the complex systemic anatomy and physiology of these twins.

Cardiovascular system

A detailed echocardiography of twin A (right) revealed a heart with two ventricles with a large ventricular septal defect (VSD) and a very small septum, atrioventricular canal defect and large primum atrial septal defect (ASD). It also showed aortic regurgitation, right aortic arch and a patent ductus arteriosus (PDA). On CT angiography, the pulmonary artery was seen, and the right aortic origin was seen separate from the left aortic origin. There was a normal relation of great arteries. Right superior vena cava (SVC) was not seen clearly. Twin B (left) had a large ASD and VSD, a hypoplastic left ventricle, transposition of great arteries and a moderate-sized PDA. The pulmonary artery flow was good. Left-sided aorta was not dilated and was normal in relation to the pulmonary artery. There was a single large SVC.

Digestive system

Oral water soluble contrast study (done on separate days) disclosed that twin A had her stomach herniated in the thoracic

Figure 5 Oral contrast dye study. (A) Twin A (right) showing stomach in the right thoracic cavity and mediastinum suggestive of a diaphragmatic defect. (B) Twin B (left) with normally placed stomach.

Learning points

- Congenital extracranial craniofacial teratomas are very rare.
- Our case exemplifies the success of expert thinking, multidisciplinary team effort and a tailored approach to this problem.
- Acceptable functional outcomes in the context of massive congenital extracranial teratomas can be achieved by early radical resection.

cavity through a diaphragmatic defect. Twin B had a normally positioned stomach. Both had separate small intestines and a common rectum and anal opening (figure 5A,B).

Hepatobiliary system

Both twins had a functionally and anatomically normal liver positioned in the right and left hypochondrium, fused in the midline. There were two separate gall bladders and bile ducts; however, the anatomy of the hepatic veins was complex.

Respiratory system

MRI of the thorax showed two separate tracheae and a pair of functionally normal lungs for each twin.

Genitourinary system

The kidney function tests were normal. There were two separate kidneys with respective ureters opening into a single bladder. No signs of obstructive uropathy were present. The external genitalia were female like.

Central nervous system and peripheral nervous system

The MRI brain was normal for each. The spinal cord for the right and the left half was separate.

Limbs and skeletal system

No bony limb abnormalities were present. Both had a separate spinal column with no vertebral abnormality and a shared pelvis (figure 6).

TREATMENT

Hospital course and management

The twins were conservatively managed with oxygen support, intravenous fluids, antibiotics and nursing care. Gavage feeds

Figure 6 MRI brain and spinal cord: two normal brains and spinal cord placed side by side next to each other.

were tolerated well. The children could not be extubated after the MRI was done on day 9 due to the complex cardiac physiology. A Blalock-Taussig shunt for duct dependent cardiac anomaly had been planned for the right twin. However, the size of the right pulmonary artery (2 mm) became the limiting factor for shunt surgery. So the twins progressively deteriorated, developed right upper lobe collapse, had repeated episodes of desaturation and bradycardia and died on day 19 of life.

DISCUSSION

Conjoined twins occur in 1 in 50 000–200 000 live births with female preponderance.^{16–18} About 60% are stillborn or die shortly after birth. Different types of conjoined twins are classified according to their site and extent of fusion (dorsal, ventral or lateral) into omphalopagus who are fixed at the umbilicus (32%), thoracopagus at the chest (40%), cephalopagus at the head, ischiopagus at the hip (6%), craniopagus at the helmet (2%), rachipagus at the spine, pygopagus at the rump (19%) and parapagus side to side (0.5%). Dicephalic parapagus twins have two heads on a single body, one pair of legs and variable degree of duplication of upper body.¹⁹ Our case falls into its most extreme category of dicephalus dibrachus dipus parapagus conjoined twins having one torso, two arms, two legs and two heads and necks situated next to each other.

Two embryonic theories of conjoined twinning are the fission and the fusion theory. The 'fission theory' states that conjoined twins occur when a fertilised ovum begins to split into identical twins but is somehow interrupted during the process and develops into two partially formed individuals who are stuck together.²⁰ The mechanism of the development of dicephalus conjoined twinning is splitting of the embryo only in the cephalic region. In the two-armed or three-armed subtypes, this division is less complete than in the four-armed subtype. It is argued that conjoined twinning cannot possibly result from a 'fission event' and can result from the fusion of mono amniotic twins.²¹ The fusion theory states that two mono-ovular embryonic discs may

lie adjacent to one another at various angles and may become secondarily united dorsally, caudally or laterally and symmetrically or asymmetrically but always homologous.²²

History dates back to AD 375 when Higden's Polychronicon and Capgrave's Chronicle of England both recorded the birth of a two-headed boy in the castle of Emaus. He was divided upwards from the navel and died at the age of 2 years.²³ The famous Scottish brothers were a pair of dicephalic conjoined twins (fused below the waist) born near Glasgow in 1940s who became proficient linguists and could speak in eight different languages. They survived until the age of 28 years.^{24 25} Thereafter until the 19th century, there were many reports of still-born dicephali.^{24 26 27} Dr Manel gave a detailed autopsy account of 8-month 11-day-old Ritta-Christinana, who were born in Paris in 1829. The twins were found to be each others' mirror images with all internal organs transposed. Their hearts were situated next to each other in a shared pericardium with one of the twins having a severely malformed heart.^{16 28} Then history witnessed the most celebrated and the longest lived dicephalic twins—the Tocci brothers. Born in Italy in 1877, they had two heads, two necks, four perfect arms, with bodies fused at the level of the sixth rib and a pair of legs and common male genitalia. Many anatomists, physicians and pathologists including Rudolf Virchow studied the case of the 'two-headed boy' and gave their detailed account. The Tocci family earned a fortune through their exhibition. The Scientific American described them as good looking and intelligent with independent minds and action. They were able to dress and undress themselves and were able to stand but could not walk a single step, instead they crawled about on the floor, using all six extremities.^{29–31} The brothers married two different women and probably had children too.³² The current living example is that of the 27-year-old famous American dicephalous conjoined twin sisters Abigail and Brittany Hensel. Described as 'One body Two souls', the twins have two heads and two spines merging at the coccyx and are joined at the thorax by sections of ribs. They have one broad ribcage with two highly fused sternums and traces of bridging ribs. It contains four lungs, two hearts in a shared circulatory system. They have two arm (rudimentary third arm was amputated) and two legs. Although each twin controls her respective side, they are well coordinated. They can not only walk but run, swim, dance and ride a bicycle too. These twins share the large bowel and probably parts of the small bowel as well, and food eaten by Abigail also nourishes her sister. They have differing characters and personalities. Their parents have never considered the option of surgical separation.^{29 33}

In the era of prenatal diagnosis of conjoined twins, a caesarean delivery is the preferred option, although cases delivered by vaginal route have been reported.^{20 29 34 35} Vaginal delivery is associated with a high potential of birth canal trauma for the mother and may also lead to birth trauma or asphyxia of the newborn.²⁹ The type of twinning is appreciable by the clinical examination and a few basic investigations. Only if surgery is planned, the need for higher imaging study arises. These investigations provide an insight to plan for organ division, use of prosthesis, tissue expanders and grafts preoperatively. A proper consent, multidisciplinary team approach, careful planning, thorough work-up, rehearsal and involvement of the parents in all steps of planning and counselling are of utmost importance.²⁹

World over, many cases of conjoined twins have been successfully separated.^{18 29 34–36} The main predictor of survival is conjunction and abnormality of the hearts. Those with cardiopulmonary malformations that are incompatible with extrauterine life are stillborn. It is desirable to separate less extensively conjoined

twins, because the children's quality of life definitely is much improved.³⁷ However, in cases like ours, where the anomalies of heart command the feasibility of separation, survival is bleak after separation.^{36–39} Some twins are closely conjoined yet may have a better quality of life if left alone rather than being separated. For such cases timely diagnosis, antenatal counselling and appropriate intervention seem to be the answer in today's world.⁴⁰

Learning points

- Management of a case of dicephalus dibrachius dipus parapagus conjoined twins is once in a lifetime opportunity for a clinician.
- A thorough literature search, multidisciplinary team effort, a detailed work-up and a proper consent and parental counselling are essential if separation is planned.
- Cardiovascular anatomy governs the possibility of surgical separation in such cases.

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Contributors Both the cases reported have been managed under the care of MB. MB and KK have assisted during the management of the conjoint twins. PG and AG along with MB have been involved in the management and flow-up of the scalp teratoma case. KK has drafted and prepared the manuscript. MB, PG and AG have substantially contributed to the editing and revision of the report. MB has agreed to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

Competing interests None declared.

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