CASE REPORT

Diprosopus tetraophthalmus: CT as a complement to autopsy

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ABSTRACT. Diprosopus is the rarest form of conjoined twinning. This anomaly is characterised by craniofacial duplication to varying degrees and is associated with anomalies of the central nervous, cardiac, respiratory and musculoskeletal systems. We present an infant characterised as diprosopus tetraophthalmus who underwent post-mortem CT, which served as a highly useful complement to autopsy.

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Diprosopus, or craniofacial duplication, is the rarest variant of conjoined twinning which results from incomplete monozygotic, monochorionic and monoamniotic twin division [1, 2]. It is characterised by a wide spectrum of abnormalities that range from simple nasal duplication to two complete faces with a single body. The presence of four eyes is designated tetraophthalmus. The incidence of associated central nervous, cardiac and respiratory system anomalies is high [3]. We report a case of an infant characterised as diprosopus tetraophthalmus for whom CT was used as a complement to autopsy to delineate the associated anomalies.

Case report

A 24-year-old primigravida with no pre-natal care presented to an emergency department in labour at an estimated 32 weeks of gestation. The mother did not smoke or drink alcohol. An ultrasound performed there reportedly showed severe polyhydramnios and hydrocephalus, with additional anomalies suspected. As a result of foetal distress and cephalopelvic disproportion, an emergency Caesarean section was performed and a 3.4 kg female was delivered (Figure 1a). The infant lived for 2 h.

A post-mortem conventional radiograph was obtained to evaluate any skeletal anomalies (Figure 2). There was duplication of the cervical spine and upper thoracic spine to approximately the T3 level, widening of the remainder of the thoracic and lumbar spine, and the sacrum appeared normal. In an effort to better delineate the extensive spinal anomalies, a CT examination of the body (LightSpeed 16, General Electric Medical Systems, Milwaukee, WI) was obtained immediately following the radiographic examination. Two- and three-dimensional reformations were subsequently performed (GE Advantage

Windows workstation 4.4, General Electric Medical Systems, Milwaukee, WI), including bone and surface-rendering algorithms.

The infant's single head with two fused faces and a very wide forehead was evident on the surface-rendered three-dimensional reformations (Figure 1b). The anomalies seen on both the imaging studies and autopsy included four globes (two lateral and two midline) located within three bony orbits, two nasal passages (Figure 3), two cleft lips and palates, a single nasopharyngeal cavity, duplication of the mandible and a single maxilla (Figure 4). There were two single temporal bones, a duplicated clivus, a single central sphenoid body with duplicated posterior clinoids and non-duplicated sphenoid wings laterally. The duplicated cervical spine articulated with the duplicated skull base with two well-defined foramina magna, separated by an interoccipital bone. Within the abdomen, the infant had a midline liver and the gallbladder and spleen were absent. The remaining visceral organs were normal.

Abnormalities, which were identified at autopsy but not on imaging, included a tracheoesophageal fistula, tetralogy of Fallot, extralobar pulmonary sequestration and pulmonary lymphangiectasia. In addition, detailed extent of the brain duplication was not fully appreciated on the CT examination.

The osseous anatomy of the spinal column was clearly defined on the CT examination. Therefore, a detailed dissection of the spinal column was not deemed necessary at autopsy. Complete cervical and thoracic spine duplication to the level of T3 was clearly seen on both the axial and the reconstructed images (Figure 4). In the remainder of the thoracic spine, there was partial anterior vertebral body duplication with separation by a small osseous segment, probably a remnant of fused medial posterior elements. The individual vertebral anatomy was best delineated on the axial images (Figure 5). Fusion of the vertebrae resulted in a widened spinal column, similar to the autopsy photograph (Figure 6). The lumbar spine, bony pelvis and appendicular skeleton were normal.

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Figure 1. Newborn with cranio-facial duplication. (a) Post-mortem frontal photograph. This female infant has a single body with two fused heads. The facial duplication is characterised by a widened forehead, two lateral orbits and one enlarged central orbit, two noses, two cleft lips and a single midline chin associated with a single body. (b) Surface-rendered three-dimensional frontal view. The facial duplication depicted is strikingly similar to Figure 1a. The orbits, cleft lips and single chin are easily delineated.

Discussion

Diprosopus, representing less than 1% of conjoined twinning, spans a wide spectrum of anomalies that range from minimal facial structure duplication to complete

dicephalus [1, 3]. A commonly cited proposed aetiology of the diprosopus phenotype is rostral bifurcation or forking of the notochord, which delineates the embryonic axis and induces neurulation. This incomplete division is thought to occur somewhere between 2 and 3 weeks following fertilisation and results in two adjacent vertebral axes, neural plates and neural crest derivatives [1, 3]. Diprosopus is found primarily in single pregnancies, but also can be seen in dichorionic twin gestations with a normal co-twin [2], then representing a triplet monochorionic/dichorionic pregnancy with an incomplete separation of monozygotic twins. Tetraophthalmus, referring to four eyes, can range from four fully separate globes and bony

Figure 2. Post-mortem frontal conventional radiograph. There is a large cranium. The two lateral bony orbits are seen (arrows) but the single central orbit is not well delineated. Duplication of the cervical and upper thoracic spine with widening of the remainder of the spine to the sacrum is present. The lower thoracic and lumbar spine has two side-by-side osseous components at each level. There are 12 paired ribs and 5 lumbar vertebral segments. The bony pelvis and appendicular skeleton are normal.

Figure 3. Axial CT at the level of the orbits. There are four globes (arrows), two lateral in separate bony orbits and two midline in a third larger fused bony orbit. Note the complete duplication of the nasal passages (asterisks).

Figure 4. Frontal image from the three-dimensional CT reformation in bone window. There is complete duplication of the cervical spine (dotted arrows) and upper thoracic spine. Partial vertebral body duplication extends to the sacrum. The duplicated mandibles (arrows) and three bony orbits are readily seen. There is substantial macrocephaly.

orbits, to two separate globes laterally with partial or full midline orbital fusion [2], as in this patient. Duplication of the eyes is always associated with duplication of the nose, but nasal duplication can be isolated [4].

Chromosomal anomalies have not specifically been identified in diprosopus [2, 3]. As such, there is no reported increased risk of recurrence. However, some authors suggest a multifactorial pathogenesis caused by interaction between environmental and genetic factors [5].

Severe abnormalities associated with diprosopus have been described in the central nervous, cardiovascular, gastrointestinal and respiratory systems, and most neonates are stillborn [4]. The more extensive duplications are associated with more severe central nervous system anomalies. Brain abnormalities include complete or partial duplication of the cerebral hemispheres with varying degrees of fusion of the posterior fossa structures [5, 6].

Figure 5. Axial CT image from the lower thoracic spine. Two separate vertebral bodies and lateral posterior elements are present. There is a small osseous structure (arrow) which probably represents residual fused medial posterior element ossification centres.

Imaging of diprosopus includes two- and three-dimensional sonography performed pre-natally to show the features of facial duplication [2, 5, 7]. Polyhydramnios is a frequent finding [3], presumably as a result of impaired swallowing of the amniotic fluid. MRI and CT have been used to evaluate the cranial abnormalities following birth and in post-mortem particularly if an autopsy is refused [4, 5]. To our knowledge, pre-natal MRI has not yet been described, and description on CT imaging has been limited to the cranium and its contents [3–5, 8, 9].

Like the central nervous system anomalies, malformation of the vertebral column can be varied. Although bone sampling is included in paediatric autopsies, detailed osseous dissection is not routinely performed. Additional prosectioning of bone is usually directed by radiological findings [10], and in our case was replaced by the CT evaluation. The full extent of the anomalies and detailed anatomy of the vertebral column duplication was readily identified with the axial CT images and subsequent reformations.

CT is occasionally used as an adjunct to, or in some cases a substitute for, conventional autopsy [11]. The image acquisition is rapid and restraint and sedation is irrelevant [12]. CT has been cited as an invaluable tool particularly in the area of forensic pathology [11, 12] and specifically in suspected cases of child abuse [11]. In our case of congenital malformation, the bone images were important to allow a more directed autopsy. Also, the surface-rendered three-dimensional reconstructions served as a guide for the radiologist who interpreted the cranial findings but who had not seen the patient at the time of imaging. Findings such as the cleft lips were evident on the surface-rendered views and the orientation of the anatomy was easier to understand.

In this case, there were several non-osseous anomalies that were not identified or were less well defined with

Figure 6. Frontal photograph of the spine obtained at autopsy. Although vertebral anomalies are seen throughout the cervical and thoracic spine, delineation of the abnormalities is limited.

the CT examination. The lack of intravenous and gastrointestinal contrast and extensive lung atelectasis precluded identification of various visceral and cardiac abnormalities. The central nervous system structures, owing to inherent lack of contrast, were less well seen, although severe anomalies were identified. Therefore, CT should be considered a complementary tool that can

potentially limit the extent of, rather than substitute for, autopsy.

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