What is your diagnosis?

A 28-year-old woman (gravida 5, para 2, live 2, abortion 1) was referred from elsewhere with multiple foetal anomalies, detected on morphology scan at 21 weeks. This was a natural conception. There was no family history of twins or congenital anomalies. There was no history of intake of teratogenic drugs. She had a dating scan at 9 weeks which was told to be normal.

A follow-up scan at our centre, revealed a foetus with two fused faces-oriented opposite to each other, a single body, and a set of upper and lower limbs. Two-dimensional (2D) grey scale ultrasonography (USG) showed four lateral ventricles, tetraophthalmos (two sets of eyes with lateral ones fused together), two separate noses and mouths, both showing cleft lips (Figure 1).

There was a single heart with transposition of the great arteries (TGA) and ventricular septal defect (VSD), a single spine, thoracic and abdominal cavity, a solitary stomach, and urinary bladder. There were no other foetal anomalies.

Diagnosis was confirmed using three-dimensional (3D) USG where both the faces could be seen, fused together, on the same plane (Figure 2).

The parents were informed about the ultrasound findings and decided to terminate the pregnancy. Termination of pregnancy was done using vaginal misoprostol (PGE1) tablets and resulted in expulsion of the malformed fetus vaginally. There were no problems encountered during the expulsion of fetal head. Autopsy findings confirmed the scan findings (Figure 3a, b). Informed written consent was obtained from the parents for presenting this case.

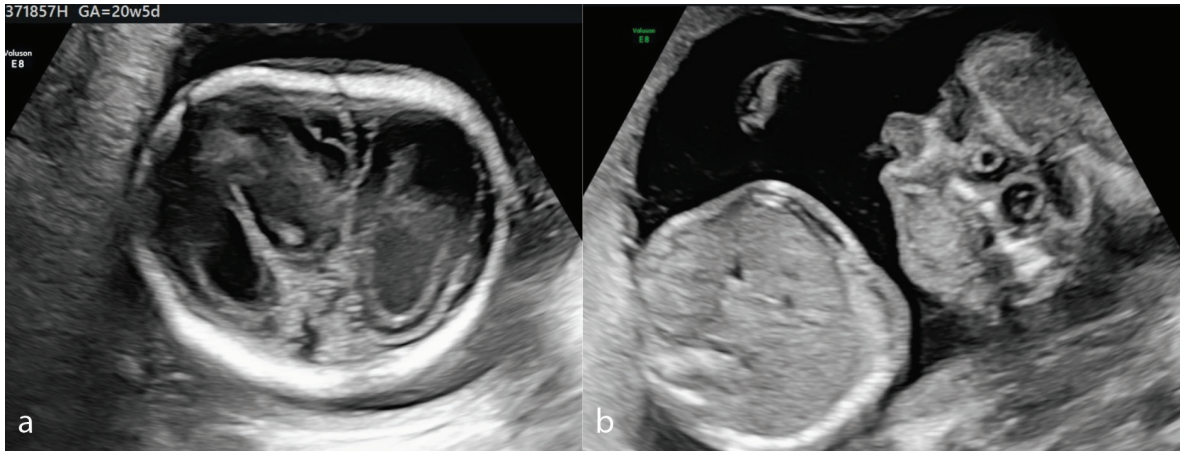

Figure 1. 2D USG shows distorted fetal head with multiple lateral ventricles (a). Tetraophthalmos with fused lateral eyes is seen in the center. There was a single thoracic and abdominal cavity (b)

2D USG: Two-dimensional ultrasonography

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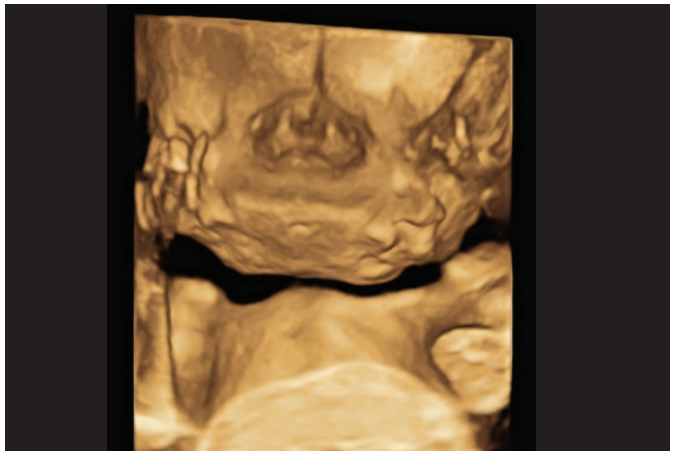

Figure 2. 3D USG shows two faces fused together laterally and oriented in opposite directions

3D USG: Three-dimensional ultrasonography

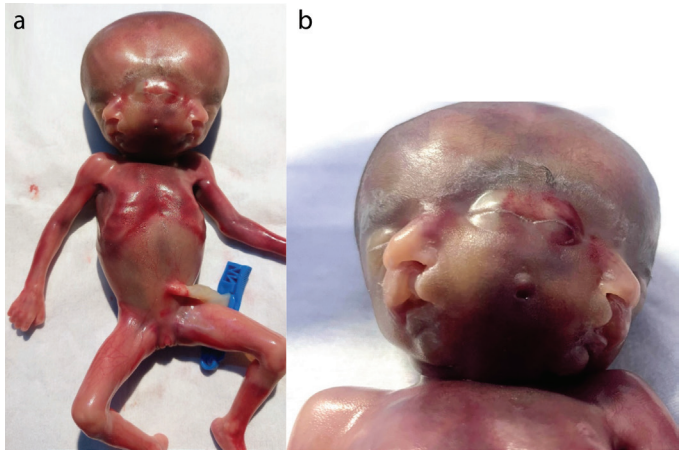

Figure 3. Post-abortal images revealed craniofacial duplication with a single body and a set of upper and lower extremities. No other organs were shared (a). Note the cleft lip on both the faces (b)

Answer

Parapagus diprosopus is a rare type of conjoined twinning which involves craniofacial duplication on a single body. Based on the site of attachment, conjoined twins are classified into: thoracopagus (42%); parapagus dicephalus (12%); cephalopagus (6%) and omphalopagus (6%). Diprosopus means duplication of the face; the trunk and limbs are normal (1).

This is an extremely rare condition, with an incidence of 1in 180,000 to 15,000,000 births (2). To date, only 40 cases have been described in the medical literature (3). Gorlin et al.'s (4) classification of the craniofacial duplication is the most widely accepted. The various types described are: 1) single mouth with duplication of maxillary arch; 2) supernumerary mouths laterally placed with rudimentary segments; 3) single mouth with duplication of mandibular segments; and 4) true facial duplication diprosopus (4).

Complete facial duplication, as in our case, is seen in only 50% of cases. The prevalence is higher in females (3). The aetiology is thought to be a disruption in the Sonic Hedgehog pathway, which controls the craniofacial tissue differentiation in the embryo (5).

The majority of cases (96%) are associated with cranial abnormalities, such as anencephaly, encephalocele, and craniorachischisis, with the most common being anencephaly (6). Other associated anomalies include: cardiac (86%); cleft lip/palate (63%); and congenital diaphragmatic hernia (42%) (3).

In the presented case there was evidence of cleft lip in both the faces (Figure 3a, b) and TGA with VSD. 2D USG raised the suspicion of foetal anomaly but 3D USG confirmed the diagnosis, since both the fused faces could be clearly seen on one plane (Figure 2). Conjoined twins are at risk of other structural anomalies, their presence impacts the prognosis adversely (3,7). In the presented case, the foetus had VSD with TGA.

Prognosis is typically poor with most of these babies dying in the first few months of life due to cardiorespiratory arrest (3). The remainder are either stillborn or abortuses. Hence early prenatal diagnosis is of paramount importance, as it gives enough time for parents to consider termination of pregnancy.

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