

CONJOINED TWINS OR CRANIOFACIAL MALFORMATION? DIPROSOPUS CASE REPORT

Borna Poljak, Kate Navaratnam, Umber Agarwal, Jo McPartland, Caren Landes, Asma Khalil

Introduction

Diprosopus comes from Greek words “di” meaning two and “prosopon” meaning face. This is an extremely rare abnormality reported to occur as 1 in 15 million births (Nair et al). The phenotype of the diprosopus has a wide spectrum from complete craniofacial duplication to partial duplications of the face of varying degrees (i.e. mandible, maxilla, oral cavity, nose). The children with partial diprosopus have invariably better prognosis following surgical correction of the defect and associated congenital abnormalities are less common. The complete diprosopus, however, has very poor prognosis with most infants being stillborn or dying soon after birth. It is also without exception associated with other abnormalities and most commonly reported are cardiac (transposition of the great arteries, ventricular septal defect, dextrocardia, hypoplastic heart) congenital diaphragmatic hernia, malrotation of the bowel, spinal abnormalities, anencephaly, cleft lip, right-left laterality viscera, anomalies in lung lobation, asplenia or polysplenia. There are no reported genetic mutations associated with the condition. There are several theories on etiology and pathophysiology of diprosopus, most of them based on hypothesis of abnormal twinning process and one exploring the involvement of over-expression of Sonic Hedgehog (SHH) gene.

Case report

A 21-year-old primigravida was thought to have a cystic hygroma at her dating scan and underwent CVS locally with normal PCR/microarray. She subsequently had a private gender reveal scan which raised the possibility of conjoined twins with a bilobed skull shape. At 20 weeks and 6 days she was reviewed in the tertiary fetal medicine unit and found to have fetal craniofacial duplication with two complete faces at 60 degrees to each other, double brains with severe ventriculomegaly in four ventricles. There was also a hypoplastic left heart, suspected right congenital diaphragmatic hernia and bilateral duplex kidneys and thoracolumbar scoliosis. The appearance was consistent with diprosopus, a rare anomaly with one head, facial duplication and a single trunk.

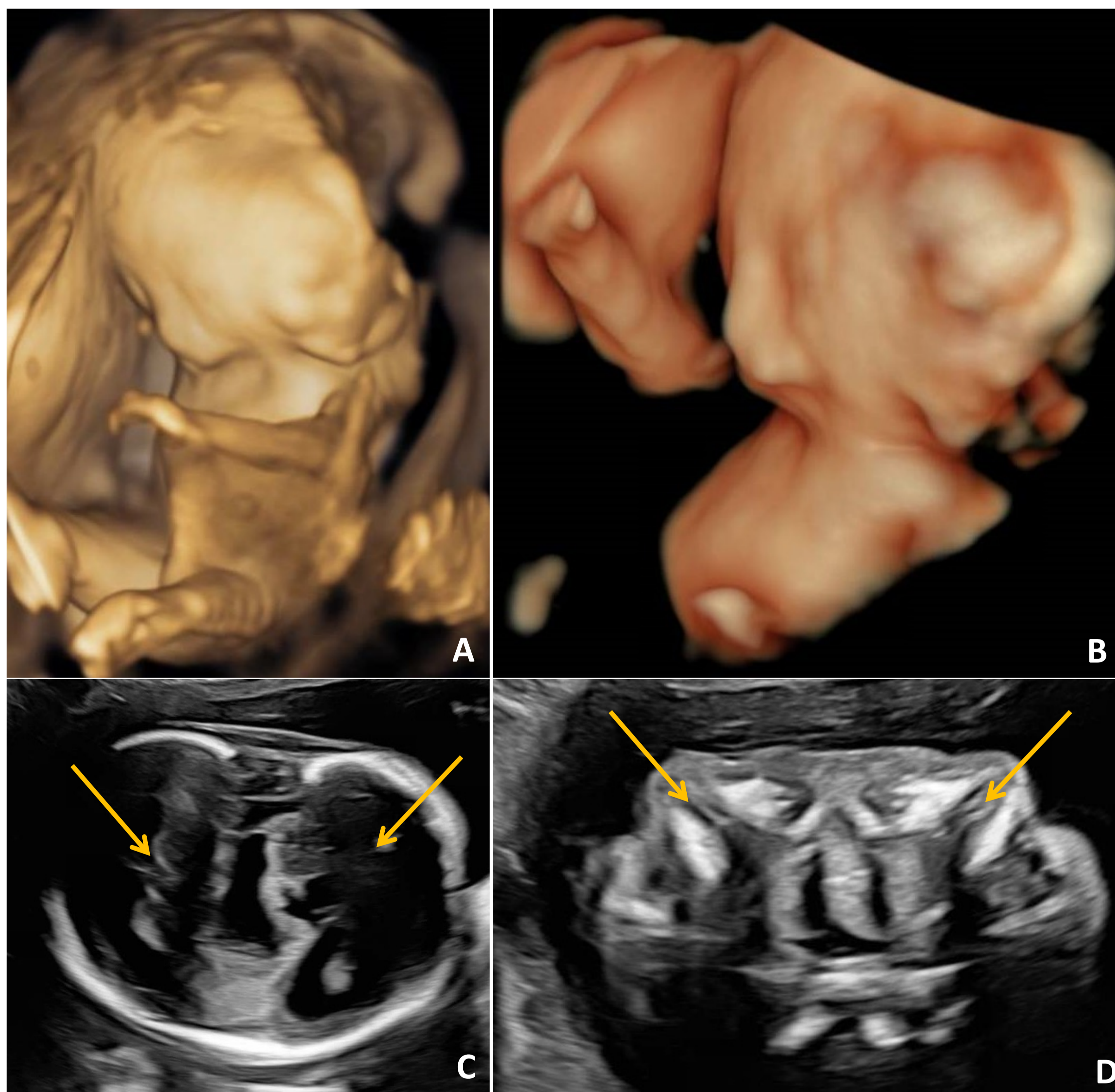


Image 1: A and B – 3D ultrasound images of facial duplication; C – Duplication of brain hemispheres and D – Two noses in transverse view on 2D ultrasound scan

After counselling on ultrasound findings, the patient opted for fetocide due to the severity of multisystem anomalies.

Fetocide was performed at 21 weeks and 4 days and the patient delivered vaginally. The parents agreed to have a postmortem examination.

Postmortem examination confirmed complete diprosopus with four eyes, two noses and two mouths. There was a cleft lip on the right face. There were four frontal and four parietal bones, one occipital bone and two additional bones between each skull. There were three anterior fontanelles. On dissection there was a bifid tongue with a single pharynx and neck structures. The cerebrum was duplicated with four hemispheres but there was a single cerebellum. Fetal heart had absent left atrium and hypoplastic left ventricle with the transposition of the great arteries and ventricular septal defect. Pulmonary venous drainage was not identified. Right diaphragmatic eventration was present close to the midline with part of the stomach, pancreas and spleen in right thoracic cavity. Bilateral duplex ureters were also identified with possible duplex pelvic/ureteral system. The bowel was malrotated.

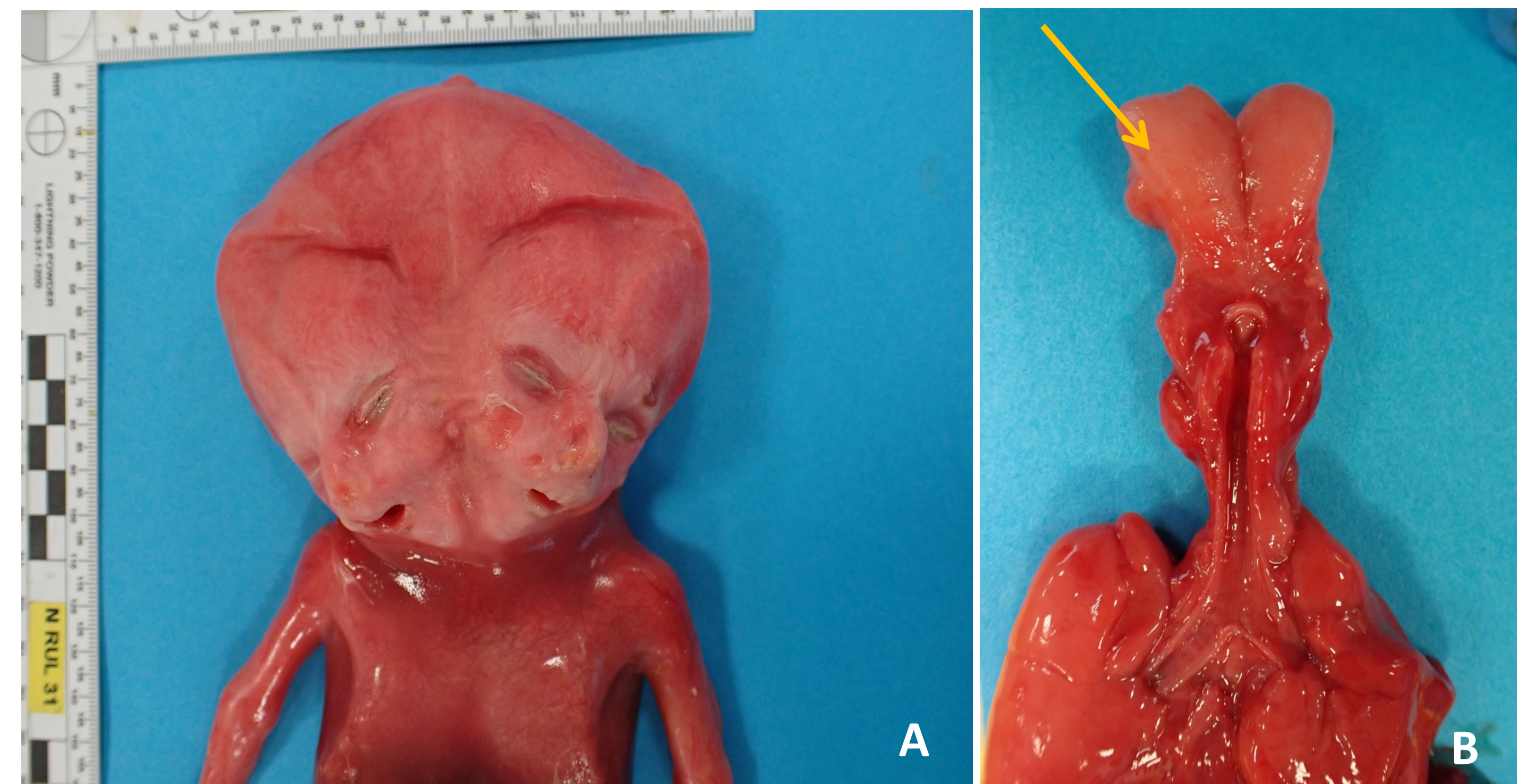


Image 2: A – Postmortem facial duplication image; B – Bifid tongue, single trachea

Supplemental X-ray skeletal survey, CT and MRI scan of the head were also performed. In addition to postmortem findings, postnatal imaging identified absence of the corpus callosum in both brains, two distinct brainstems, including mid brain, pons and medulla, that unite at the level of the foramen magnum to become a single cervical cord. It was difficult to accurately define the pituitary glands but two soft tissue signal areas were described that may represent pituitary glands associated with each brain. There were multiple vertebral segmental anomalies.

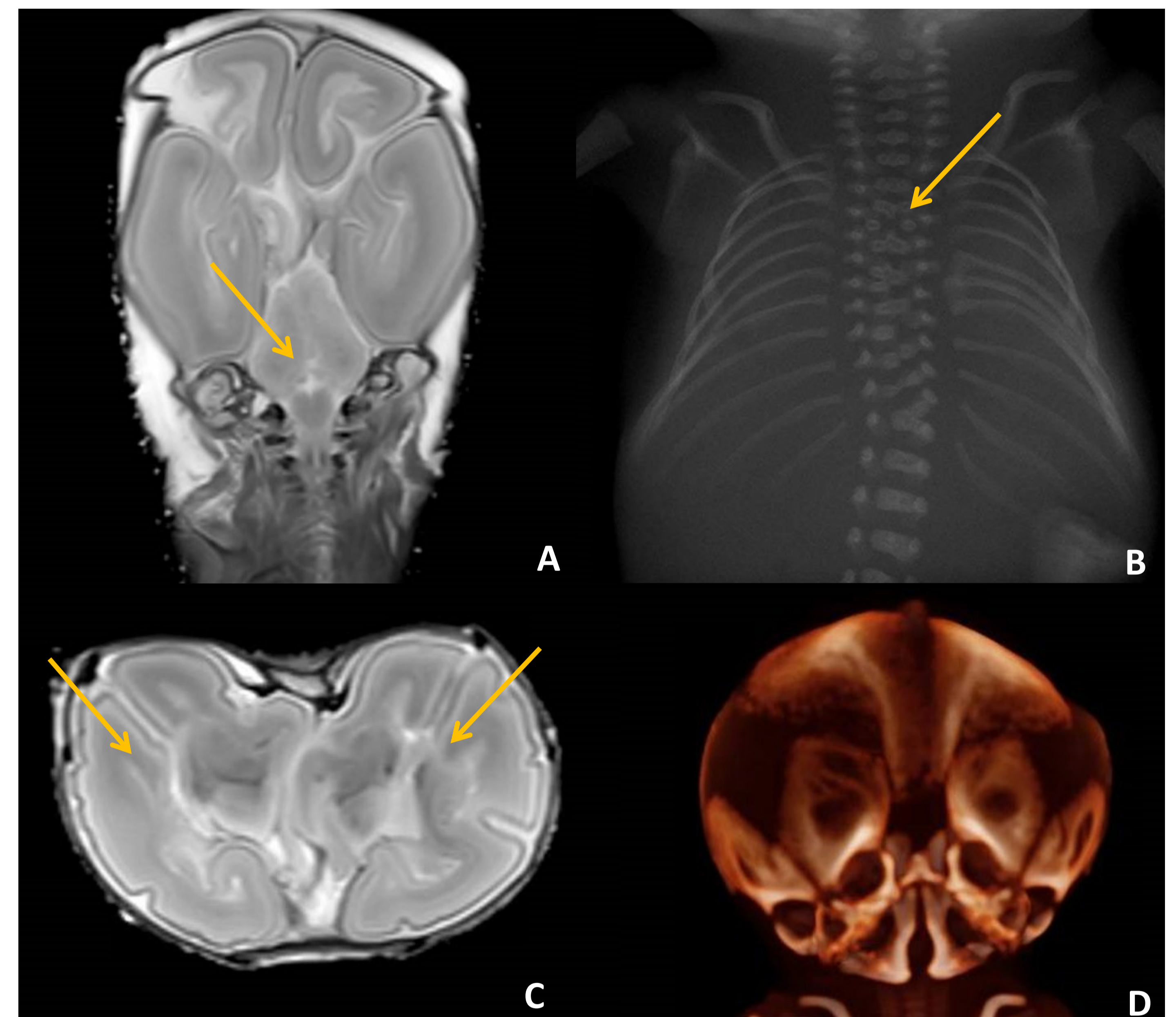


Image 3: A – MRI of single cerebellum; B – X-ray of vertebral segmental anomalies; C – MRI of Duplication of brain hemispheres; D – 3D reconstruction of diprosopus skull

Conclusion

Diprosopus is an extremely rare congenital anomaly and there is limited international experience, except for isolated case reports. Our case presented with typical abnormalities previously described in literature.

In the cases of complete diprosopus patient counselling should involve the options of continuing the pregnancy with the support of palliative care team if available or termination of pregnancy.

They can be reassured that there is no genetic basis for diprosopus and no cases of recurrence have been reported.

In cases of partial diprosopus parents can be counselled about the good prognosis following surgical treatment.

Literature

Bidondo MP, Groisman B, Tardivo A, Tomasoni F, Tejero V, Camacho I, Vilas M, Liasovich R, Barbero P. Diprosopus: Systematic review and report of two cases. *Birth Defects Res A Clin Mol Teratol*. 2016 Dec;106(12):993-1007. doi: 10.1002/bdra.23549. Epub 2016 Oct 5. PMID: 27704687.
Nair NM, Swarr DT, Barnes-Davis ME. Preterm infant with diprosopus and holoprosencephaly. *Clin Case Rep*. 2021 Dec 22;9(12):e05163. doi: 10.1002/ccr3.5163. PMID: 34987809; PMCID: PMC8695654.

Acknowledgements: We would like to thank the patient and her family for their consent to share their story in aim to improve the care of other patients whose pregnancy is affected with this condition.