

Prenatal diagnosis of Cerebriform Sebaceous Naevus with Three-dimensional (3D) ultrasound technology



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Introduction

- Sebaceous naevus (SN) is a rare birthmark found in 0.3% of neonates, characterised as a congenital hamartoma of the pilosebaceous follicular unit^{1,2}.
- SN usually presents on the scalp, trunk and lower limbs.
- Cerebriform Sebaceous Naevus (CSN), a rare variant of SN¹.
- CSN is a challenging diagnosis prenatally due to its flat, smooth and waxy appearance & lack of association with extracutaneous manifestations².
- This case report describes the detection of CSN using three-dimensional (3D) prenatal ultrasound, with CSN manifesting as bilateral infra-auricular masses and tumours in the facial and neck region.

Case Report

- A woman with four previous vaginal deliveries and no previous neonatal auricular or hearing issues, was referred to our Maternal Fetal Medicine (MFM) Service following an abnormal ultrasound at 24 weeks' of gestation.
- Imaging revealed bilateral soft tissue masses around the fetal ears.
- No earlier fetal imaging or aneuploidy screening had been performed.
- The maternal medical history was notable for hypothyroidism (non-compliant with thyroxine). She was a current smoker of tobacco and marijuana.

Fetal imaging at 27.4 weeks of gestation

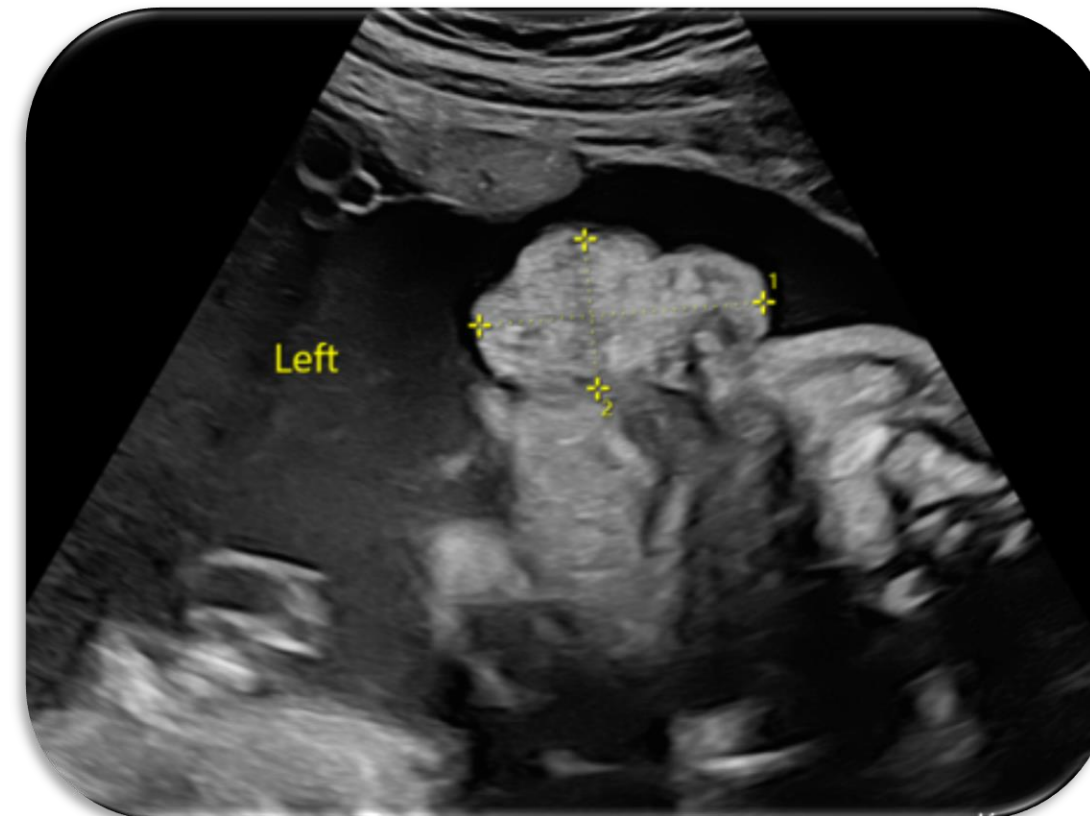


Image 1: Left infra-auricular mass

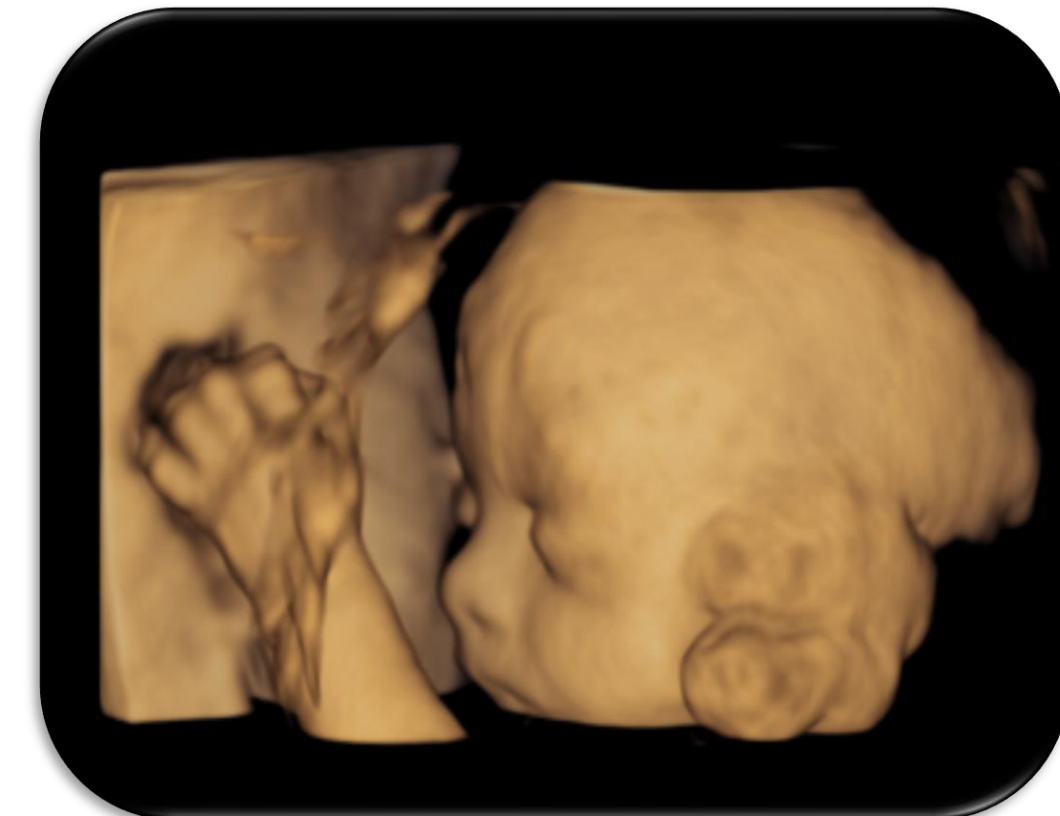


Image 2: 3D of left infra-auricular mass

- 32mm x 15mm x 19mm bilateral soft tissue masses replacing the lower external ears with superior rotation of the pinna and helix.
- No other anatomical abnormalities were evident.

33.1 weeks gestation



Image 5: Fetal face

- Serial ultrasound scans showed a progressive increase in size and vascular supply of the bilateral infra-auricular masses
- The growth of the masses stabilised at 35+1 weeks gestation.
- However, a new vascular right cheek mass developed (Image 6).

35.1 weeks gestation



Image 6: New right cheek lesion

Follow-up

- Multiple specialities, including dermatology, plastic surgery, ear, nose and throat, genetics, cardiology and ophthalmology, assessed the baby and excluded associated syndromes with SN.
- Comprehensive examinations including brain MRI, abdominal and spine ultrasound, newborn hearing check and echocardiogram were normal.
- Biopsy confirmed the lesion as CSN, and chromosomal microarray of the baby's blood was normal.
- Baby was well and discharged on day 6 of age with the CSN lesions remaining unchanged at day 40.
- Ongoing outpatient follow-up with dermatology and plastics were scheduled to consider surgical removal of the CSN.

Fetal imaging at 29.4 weeks of gestation

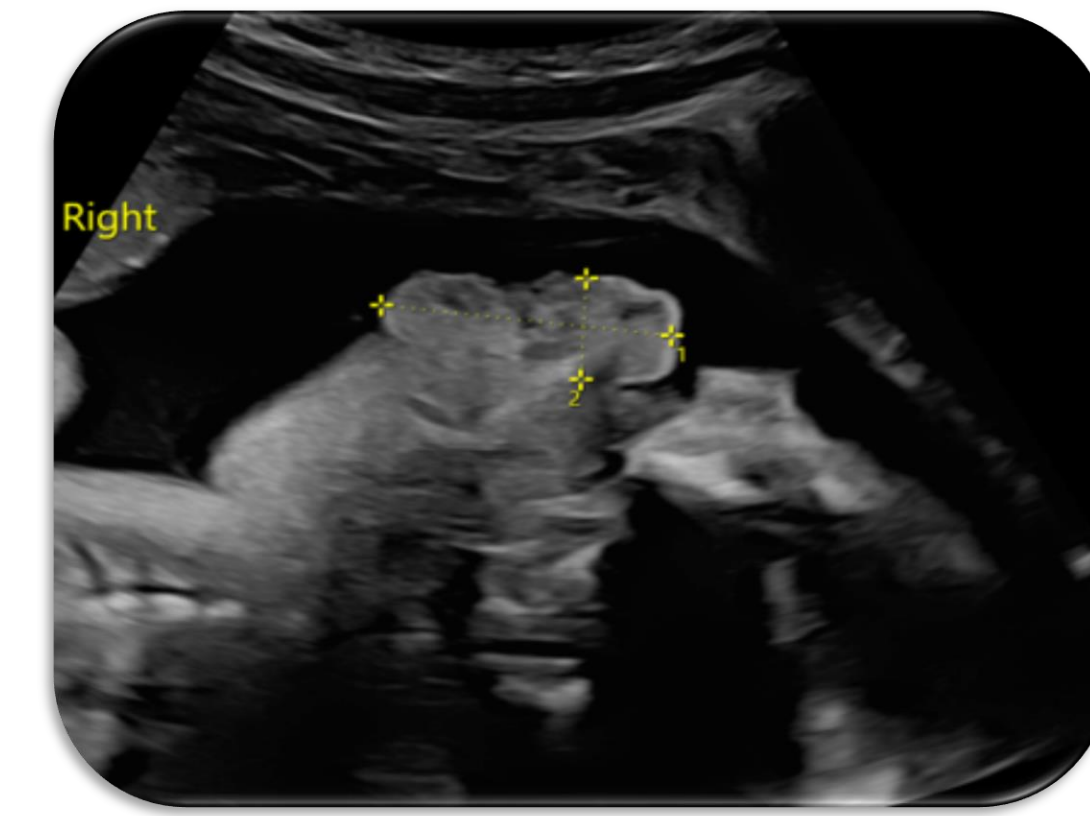


Image 3: Right infra-auricular mass

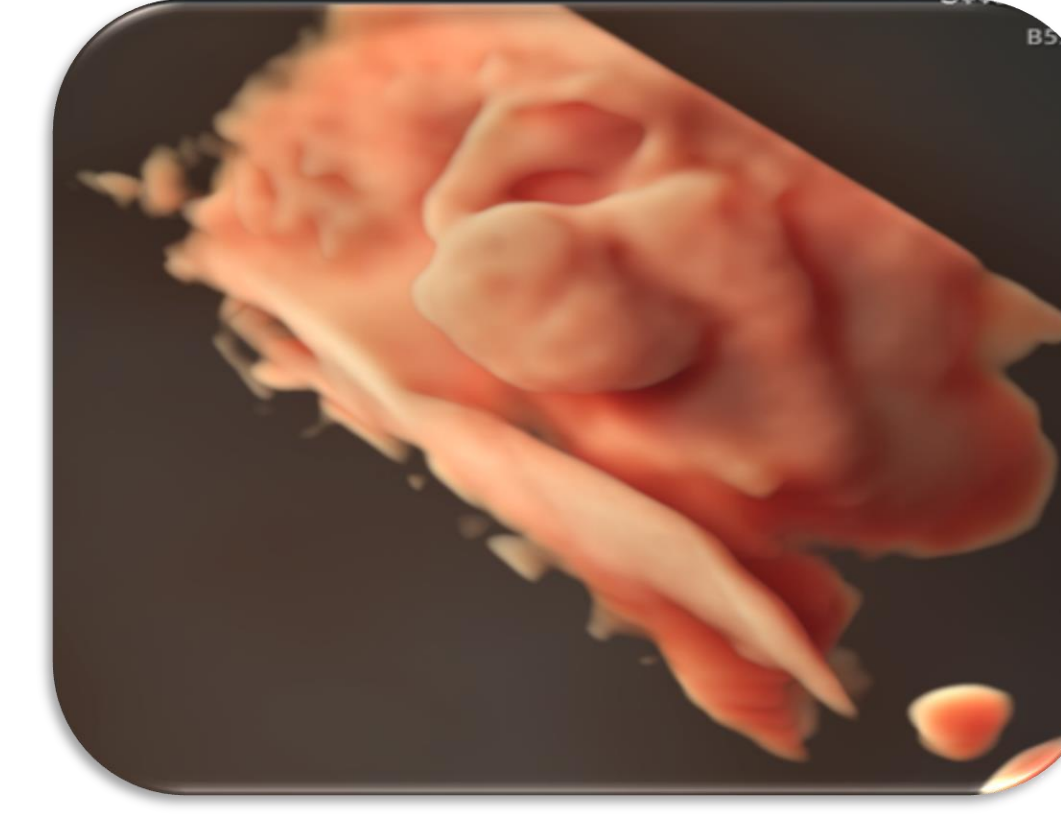


Image 4: Right-infra-auricular mass (3D)

Fetal imaging at 36.4 weeks gestation



Image 7: New lesion medial to right eye

- Imaging at 36.4 weeks gestation revealed a new lesion medial to the right eye (Image 7).

Discussion & Conclusion

- This case highlights the valuable role of prenatal imaging in the detection of rare cutaneous lesions with the aid of 3D ultrasound technology.
- Detection allows for postnatal management planning with multidisciplinary team input.
- Recent advances in ultrasound technology have improved the assessment of normal and abnormal structures, enhancing the detection rates of subtle fetal abnormalities, especially in the evaluation of fetal face and profile.

Outcome

- The baby was delivered vaginally via forceps at 38.1 weeks gestation, with Apgar scores of 9 & 9.

- On examination:
 - Characteristic cerebriform lesions were observed in the bilateral pinna and preauricular regions, right zygomatic arch, temple and right medial canthus extending to the forehead (Images 8, 9 and 10).



Image 8: Right facial profile

- The left pinna was partially visible, and the left exterior auditory canal (EAC) was occluded by polyp-like lesions (Image 9).
- The right superior pinna remained intact, while the cerebriform mass extended inferiorly encasing the lobule, tragus, and tracking preauricular and down along the angle of the mandible.
- The right EAC was patent with no lesions (Image 8, 10).
- The remainder of the clinical examination was unremarkable.



Image 9: Left facial profile



Image 10: Right and frontal facial profile

References

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Disclosure

None