

Pericallosal lipoma with callosal dysgenesis and coexisting monosomy X mosaicism

Albert Guarque Rus¹, Gemma Soro², Glòria Gual¹, Lourdes Martín¹, Linda Puerto¹, Montserrat Inglès¹, Marta Jané¹,
Mónica López¹, Mónica Ballesteros¹

¹ Hospital Universitari de Tarragona Joan XXIII, Tarragona, Spain. ² Obstetrics and Gynecology, Institut Català de la Salut, Barcelona, Spain

1) INTRODUCTION

Pericallosal lipoma is a rare congenital lesion adjacent to the corpus callosum. It is frequently associated with callosal abnormalities and may be detected during prenatal neurosonography. When structural CNS findings coexist with chromosomal abnormalities, counselling becomes substantially more complex.

2) CASE PRESENTATION

A singleton pregnancy was referred at 28+3 weeks after second-trimester ultrasound showed borderline posterior ventricular horns (9.5 and 9.4 mm) without additional abnormalities. Detailed neurosonography demonstrated corpus callosum dysgenesis and an echogenic pericallosal lesion suggestive of lipoma. Fetal MRI confirmed the findings.

3) IMAGING FINDINGS

Prenatal imaging suggested a partial callosal anomaly associated with a pericallosal lipoma. Pathological examination later confirmed partial agenesis of the corpus callosum, with absence of the posterior body and splenium, together with a pericallosal lipoma.

4) GENETICS / OUTCOME

Karyotyping revealed mosaicism: 46,X,der(X)(?)[10]/45,X[10]. Array-CGH identified terminal deletions involving Xp22.33p11.21 and Xq21.2q28 with preservation of a pericentromeric region, consistent with a ring X chromosome. This pattern reflects mosaicism combining monosomy X and a predominant ring X cell line. Prenatal exome sequencing was non-conclusive. Following parental decision, pregnancy termination was performed.

5) CONCLUSION

Callosal dysgenesis is rarely reported in Turner syndrome and in mosaic cases involving ring X chromosomes. Although causality cannot be established, the coexistence of structural brain findings and chromosomal mosaicism changed the overall prognosis and influenced parental decision-making.

KEY MESSAGE

A pericallosal lipoma should prompt detailed assessment of the corpus callosum and careful genetic evaluation when associated anomalies are present. The combination of neurosonography, MRI and cytogenetics was essential for diagnosis and counselling in this case.

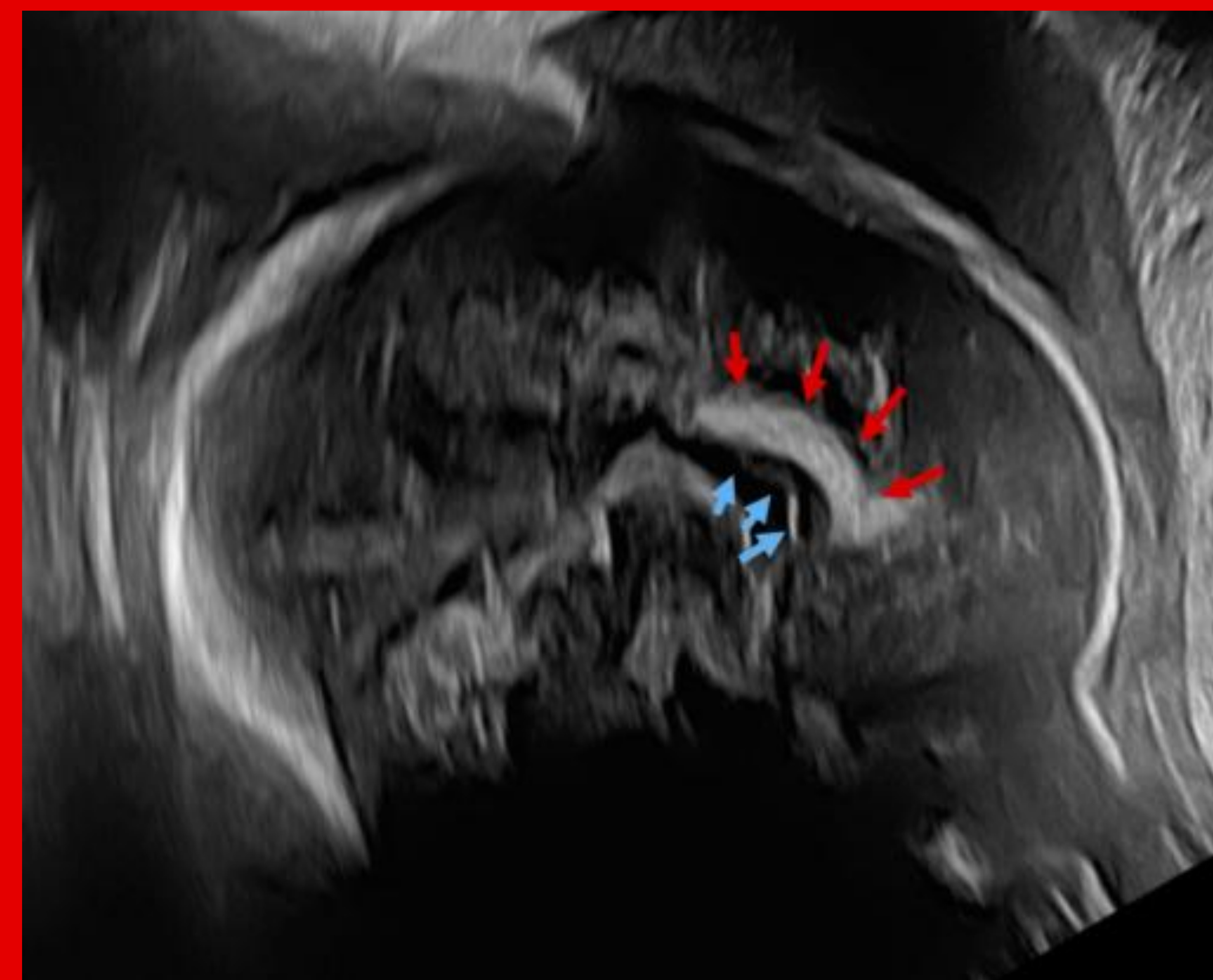


Figure 1. Mid-sagittal neurosonographic view of the fetal brain at 28+3 weeks. The echogenic curvilinear pericallosal lesion is identified along the superior aspect of the corpus callosum, consistent with a pericallosal lipoma (red arrows). Associated callosal dysgenesis is also demonstrated, with abnormal morphology and incomplete development of the posterior portion of the corpus callosum (blue arrows). The coexistence of these findings prompted further fetal neuroimaging and genetic assessment.