

Malformations of cortical development (MCD): a case series and literature review

Enoch Q. S. Barreto^{1, 2}, Javier Miguelez², Thiago S. Oliveira², Larissa R. Esporcatte², Bryda B. Cunha³, Mariana A. Carvalho^{1, 4}, Mario H. Carvalho^{1, 4}

1. Fleury Medicina e Saude, Brazil. 2. Hospital e Maternidade Escola de Vila Nova Cachoeirinha, Sao Paulo, Brazil. 3. Medical School - Araquara University, Araraquara, Brazil. 4. Department of Obstetrics and Gynecology, University of São Paulo, Brazil.



This is a retrospective observational study of 5 cases of malformation of cortical development diagnosed during pregnancy between 2021 and 2026 in two centers (HMME Vila Nova Cachoeirinha and Fleury Medicine and Health) and literature review.

In 2 cases the findings were observed during second-trimester scan; 2 cases were referred to fetal neurosonography because unilateral ventriculomegaly; and 1 case because polycystic kidneys.

In 2 cases exome sequencing (WES) was performed with the this results: (a)case 2: a pathogenic deletion in long arm: chr21: 14982835-28340026 (13.4Mb); (b)case 3: a variant of undetermined significance (VUS) c.9620G>A: p.(Arg3207His), in heterozygosity (55%), in exon 60 of the RELN gene.

In all 5 cases, the presence of midline distortion, atypical anterior complex, cavum septi pellucidum distortion, ventriculomegaly, ventricular asymmetry with frontal horn dysmorphism, and dysgyria was observed. In 4 cases (80%), frontal horn dilatation, persistence or cavitation of the ganglionic eminence, asymmetric Sylvian fissure, and delayed gyration were observed. The other signs were less frequent. These signs are consistent with the literature.

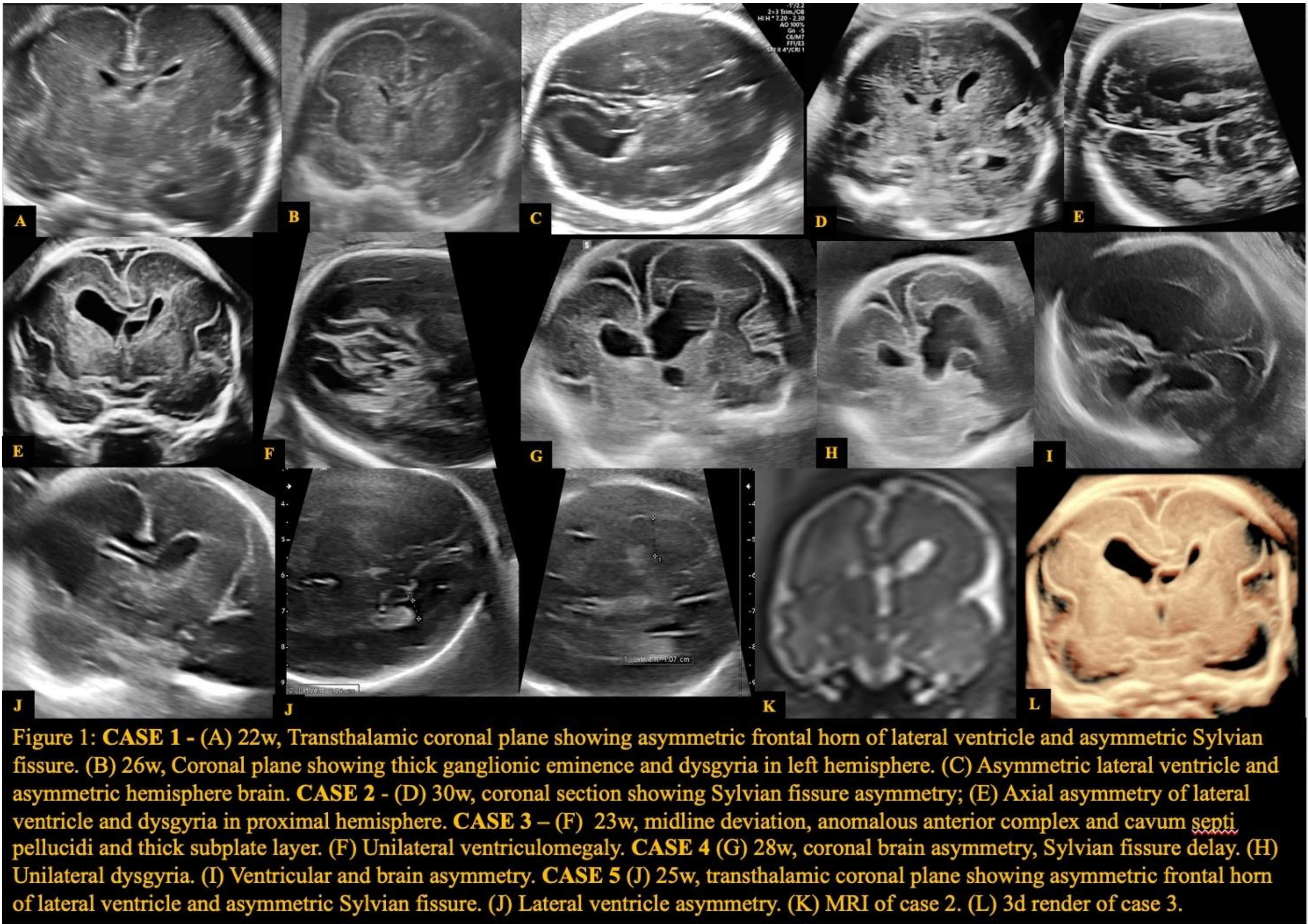


Figure 1: **CASE 1** - (A) 22w, Transthalamic coronal plane showing asymmetric frontal horn of lateral ventricle and asymmetric Sylvian fissure. (B) 26w, Coronal plane showing thick ganglionic eminence and dysgyria in left hemisphere. (C) Asymmetric lateral ventricle and asymmetric hemisphere brain. **CASE 2** - (D) 30w, coronal section showing Sylvian fissure asymmetry; (E) Axial asymmetry of lateral ventricle and dysgyria in proximal hemisphere. **CASE 3** - (F) 23w, midline deviation, anomalous anterior complex and cavum septi pellucidi and thick subplate layer. (F) Unilateral ventriculomegaly. **CASE 4** (G) 28w, coronal brain asymmetry, Sylvian fissure delay. (H) Unilateral dysgyria. (I) Ventricular and brain asymmetry. **CASE 5** (J) 25w, transthalamic coronal plane showing asymmetric frontal horn of lateral ventricle and asymmetric Sylvian fissure. (J) Lateral ventricle asymmetry. (K) MRI of case 2. (L) 3d render of case 3.

The described ultrasound signs associated with cortical developmental malformations are important for screening and diagnosis. Among the series of cases presented, the most frequently found signs were: midline distortion, atypical anterior complex, distortion of the cavum septi pellucidi, ventriculomegaly, ventricular asymmetry with frontal horn dysmorphism, dysgyria, frontal horn dilatation, persistence or cavitation of the ganglionic eminence, asymmetric Sylvian fissure, and delayed gyration.