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Porencephaly a Sign Revealing CMV Infection: Case Report

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Abstract | **Case Report**

This case illustrates a rare prenatal presentation of porencephaly associated with congenital cytomegalovirus (CMV) infection, highlighting the importance of considering viral etiologies in the differential diagnosis of fetal brain malformations. A 28-year-old primigravida was referred at 24 weeks and 3 days of gestation for suspected polymalformative syndrome. Ultrasound revealed multiple anomalies including microcephaly, cerebellar and vermian hypoplasia, Blake's pouch cyst, mild ventriculomegaly, near agenesis of the corpus callosum, agenesis of the septum pellucidum, hyperechoic intestine, and adrenal calcifications. CMV infection was suspected, and confirmed by positive serology and CMV DNA detected in the amniotic fluid; fetal karyotype was normal. A medical termination of pregnancy was performed, and fetopathological examination confirmed porencephaly and signs of CMV infection. This case emphasizes the critical role of detailed prenatal imaging and targeted virological testing in diagnosing congenital infections and guiding perinatal decision-making. Early detection of such severe anomalies allows timely intervention and informed counseling.

Keywords: Porencephaly, Congenital Cytomegalovirus Infection, Prenatal Diagnosis, Polymalformative Syndrome, Fetopathology Case Report.

INTRODUCTION

Porencephaly is a rare neurological disorder characterized by the presence of cysts or cavities within the cerebral hemisphere of the brain [1]. These cavities are filled with cerebrospinal fluid and result from brain tissue damage [2]. It can be genetic or acquired. If acquired, the congenital cytomegalovirus (CMV) infection could be revealed by the existence of porencephaly.

PATIENT AND OBSERVATION

Patient Information

A 28-year-old woman, blood group A positive, gravida 1 para 0 (G1P0A0), with no significant medical or surgical history, and no known consanguinity with her spouse, was referred at 24 weeks and 3 days of gestation for evaluation of a suspected polymalformative fetal syndrome identified during a routine antenatal check-up. There was no relevant family or genetic history, and no prior interventions had been performed before referral.

Clinical Findings

Prenatal morphological ultrasound revealed multiple central nervous system anomalies, including porencephaly (figure 1) microcephaly (figure 2), mild ventriculomegaly (10-12 mm) (figure2), Blake's pouch cyst, vermian and cerebellar hypoplasia (figure 3), near agenesis of the corpus callosum (figure 4), and agenesis of the septum pellucidum. Other associated findings included the "tear drop" sign, hyperechoic intestine, and bilateral adrenal calcifications (figure 5).

Timeline of Current Episode

- 24+3 weeks gestation: Referred for fetal malformation assessment.
- Following referral: Detailed fetal ultrasound performed revealing multiple cerebral and extra-cerebral anomalies.
- Shortly after: CMV serology requested based on the suspicion of congenital infection. Amniocentesis performed; CMV DNA detected in amniotic fluid; fetal karyotype was normal.
- Subsequently: Decision for medical termination of pregnancy taken based on fetal prognosis and maternal request.

- Few days later: Termination performed; stillborn female delivered.

Diagnostic Assessment

The fetal morphological ultrasound was the cornerstone for anomaly detection. CMV infection was suspected and confirmed through maternal serology (suggestive of a primary infection) and molecular analysis of amniotic fluid, which identified the presence of CMV DNA. The normal karyotype excluded chromosomal abnormalities. Diagnostic challenges were minimal due to timely access to virology and genetic testing.

Diagnosis

Presumptive Diagnosis: CMV-related congenital infection with neurological impairment.

Final Diagnosis: Porencephaly and adrenal calcifications due to congenital CMV infection, confirmed by fetopathological examination.

The prognosis was poor due to the extent of brain involvement and associated malformations.

Therapeutic Interventions

Given the severity of fetal anomalies and poor prognosis, and in accordance with the patient's wishes, a medical termination of pregnancy was conducted. The procedure was performed using Misoprostol (Cytotec) following the FIGO protocol. The outcome was the delivery of a stillborn female without externally visible malformations.

Follow-up and Outcome of Interventions

Post-termination, the fetopathological study confirmed the prenatal findings, revealing porencephaly (figure 6), preserved corpus callosum, and adrenal calcifications consistent with CMV infection. The procedure was uneventful, and no complications were reported in the postpartum period. Maternal physical recovery was satisfactory.

Patient Perspective

The patient expressed sadness upon learning the diagnosis but was thankful for the clarity provided by the diagnostic workup. She appreciated being involved in the decision-making process and felt supported throughout. Although emotionally difficult, she felt the medical termination was the most humane decision given the prognosis.

Informed Consent

Written informed consent was obtained from the patient for all diagnostic procedures, the medical termination of pregnancy, and for publication of this anonymized case report.

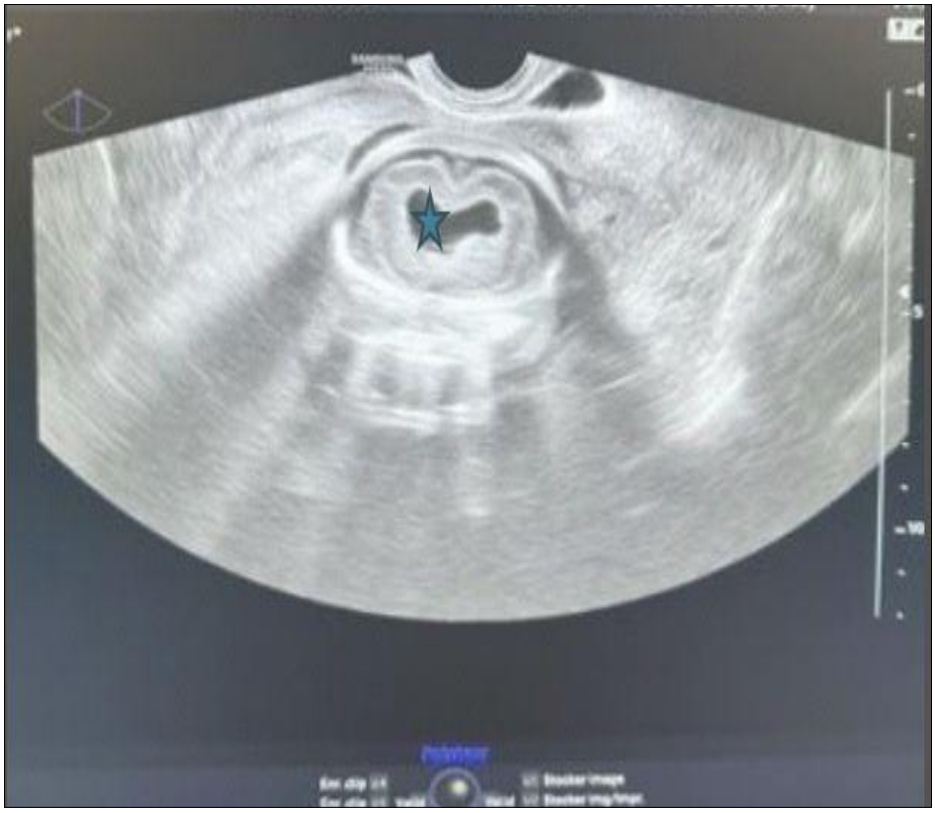

Figure 1: Prenatal ultrasound showing a cystic cavity within the cerebral hemisphere suggestive of porencephaly(star)

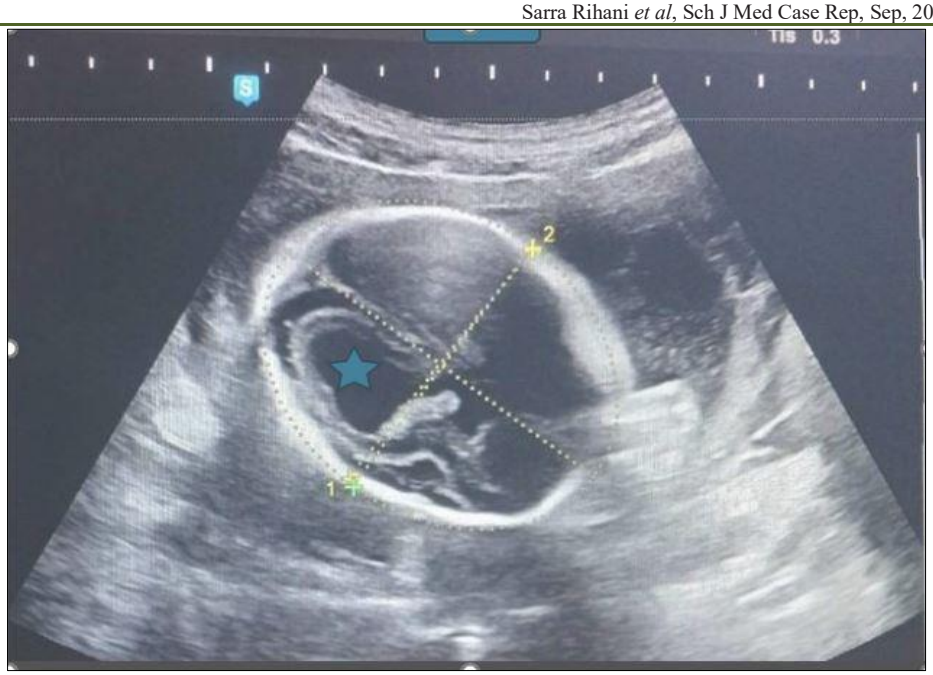

Figure 2: Axial view demonstrating microcephaly and mild ventriculomegaly (10–12 mm) (star)

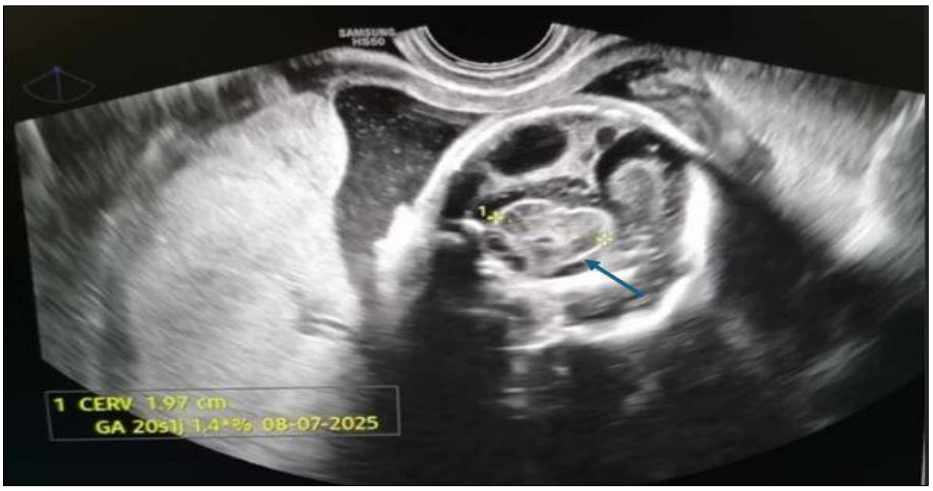

Figure 3: Midsagittal view revealing cerebellar hypoplasia (star)

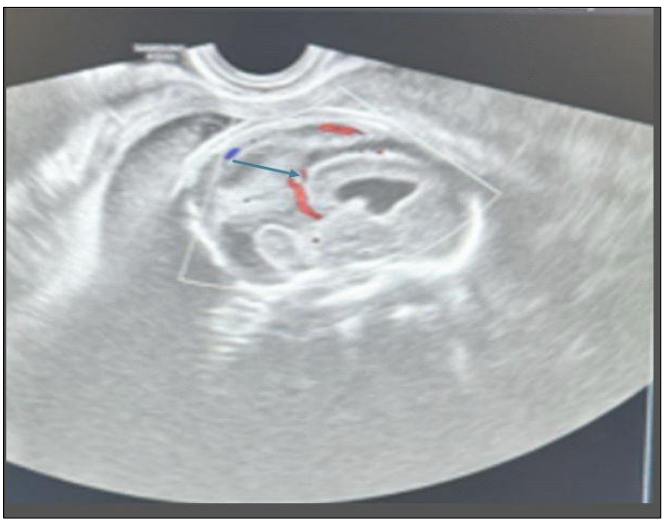

Figure 4: Sagittal image illustrating near agenesis of the corpus callosum (arrow)

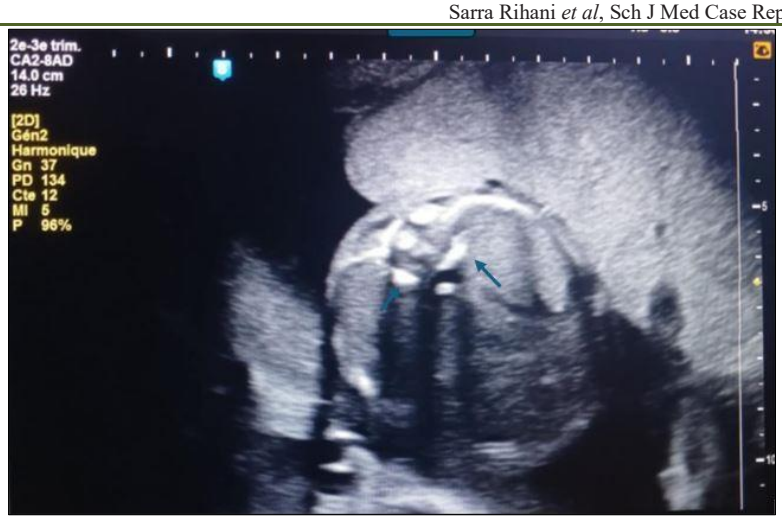

Figure 5: Axial abdominal view showing bilateral adrenal calcifications (arrow)

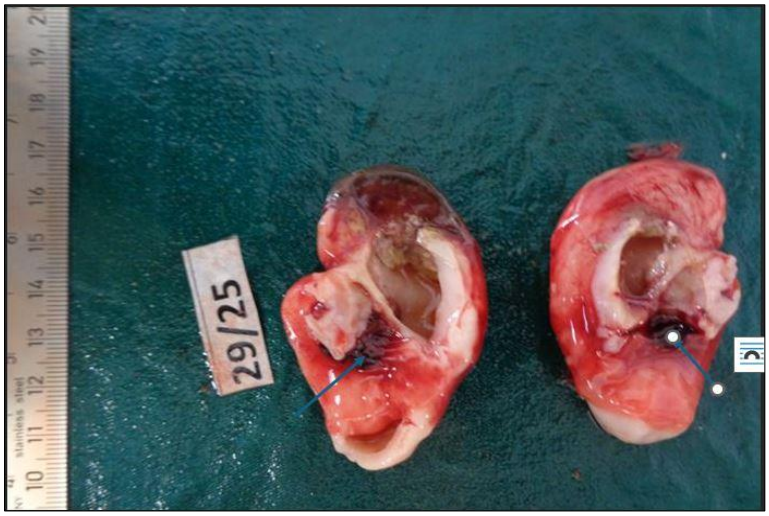

Figure 6: porencephaly confirmed by fetopathological examination (arrow)

DISCUSSION

Porencephaly is a rare neurological disorder characterized by the presence of cerebrospinal fluid-filled cavities or cysts within the cerebral hemispheres, resulting from focal destruction of brain tissue [2]. Its etiology is multifactorial. In utero vascular events—such as periventricular hemorrhages or ischemic strokes—are among the most common causes, particularly in cases diagnosed during infancy [3]. Congenital infections, especially cytomegalovirus (CMV), are also well-documented causes, as they can induce necrosis and cavitary brain lesions [4]. In our case, porencephaly was diagnosed following a detailed prenatal ultrasound that revealed severe cerebral anomalies including microcephaly, cerebellar and vermian hypoplasia, mild ventriculomegaly, near agenesis of the corpus callosum, and the presence of the "tear drop" sign. These findings raised suspicion of an in utero infection, which was confirmed by serological testing and amniocentesis identifying CMV DNA in the amniotic fluid.

Genetic mutations, particularly in the COL4A1 gene, have also been implicated in familial forms of porencephaly, although this was not the case in our patient, who had no relevant personal or familial genetic history, and a normal fetal karyotype was confirmed [5].

The clinical manifestations of porencephaly vary depending on the extent and location of the lesions. Common signs include microcephaly, hypotonia, hemiparesis, epileptic seizures, and global developmental delay [6]. In our case, although the pregnancy was terminated before birth, the presence of porencephaly and extensive CNS malformations strongly indicated a poor neurological prognosis had the pregnancy been continued.

Management of porencephaly is primarily supportive and requires a multidisciplinary approach. Physical, occupational, and speech therapies are fundamental for improving motor and cognitive outcomes. Seizures, when present, are treated with antiepileptic drugs, though response can be variable [7]. In complicated cases involving hydrocephalus, surgical

intervention such as ventriculoperitoneal shunting may be needed [2]. In our patient's situation, the degree of fetal brain destruction associated with CMV infection, combined with other extracerebral anomalies such as adrenal calcifications and hyperechoic intestine, led to the decision for medical termination of pregnancy.

Prognosis in porencephaly largely depends on the severity and extent of cerebral involvement, as well as the presence of associated anomalies or comorbidities [1]. While some patients may achieve acceptable functional outcomes, others suffer severe neurological impairments requiring lifelong support [4]. In our case, the diagnosis of porencephaly in association with multiple brain and visceral malformations underscored the poor prognosis, justifying the decision for termination after multidisciplinary counseling and maternal consent.

CONCLUSION

This case illustrates the rare but severe neurological consequences of congenital CMV infection, notably porencephaly, which is often associated with extensive brain damage and poor prognosis. The constellation of prenatal ultrasound findings—including microcephaly, cerebellar hypoplasia, ventriculomegaly, and adrenal calcifications—should prompt clinicians to investigate infectious causes such as CMV. Early detection through targeted serological testing and amniocentesis plays a crucial role in establishing the diagnosis and guiding parental counseling. In cases with major cerebral involvement, medical termination of pregnancy may be a considered and ethically appropriate option. The main takeaway from this case is the importance of including congenital infections in the differential diagnosis of fetal brain anomalies and acting promptly to optimize outcomes and support parental decision-making.

Competing Interests: The authors declare no competing interest.

Authors' Contributions

Patient Management: Sarra Rihani, Montasar Hafsi, Houssem Ragmoun.

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Manuscript Drafting: Sarra Rihani, Aymen Khalfaoui.

Manuscript Revision: Ikram Ben Abdallah, Mariem Rahmani, Houssem Ragmoun.

All authors have read and agreed to the final version of the manuscript.

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List of Tables and Figures (if any)

List all tables and figures (if any) included in the manuscript, with their detailed legend: Example. Note that Tables should be included at the end of the manuscript, after the reference section. Figures should be uploaded during the submission process.

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