Prenatal Diagnosis of an Extremely Rare Neurological Disease

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Abstract
We report one clinical case of a 29-year-old, Caucasian woman who is pregnant for the first time. Ultrasound examination showed a singleton fetus with porencephaly. Amniocentesis was done, and the fetal chromosomal analysis revealed a fetus with normal karyotype: 46,XY. After a complex and very difficult genetic counselling the parents opted to terminate the pregnancy. The autopsy confirm the prenatal ultrasound diagnosis.

Keywords: porencephaly, ultrasound examination, amniocentesis, prenatal diagnosis

1. Introduction
Porencephaly is an extremely rare neurological disease characterized by the presence of solitary or multiple degenerative cerebrospinal fluid (CSF) cavities within the brain parenchyma [1]. Porencephaly is prenatally seen as cystic cavities within the brain that usually communicate with the ventricular system, the subarachnoid space or both [2]. Most cases are sporadic [3]. Birth trauma, inflammatory diseases, amniocentesis complications, and twin-to-twin transfusion syndrome are also recognized as possible causes [4]. Scattered familial cases have been reported, and evidence for mutations in genes encoding type IV collagen alpha-1 and alpha-2 (COL4A1 and COL4A2) chains has been demonstrated in patients radiologically classified as either porencephaly or schizencephaly [3].

2. Materials and Methods
A 29-year-old, Caucasian woman, pregnant for the first time, was referred to a private medical center, from Bucharest, Romania, at 16 weeks’ gestation for a routine prenatal ultrasound investigation. The couple was non-consanguineous and clinically healthy [5]. Ultrasound examination revealed a singleton live fetus with porencephaly (Figure 1, 2). Genetic amniocentesis and the fetal chromosome analysis revealed a karyotype of 46,XY (Figure 3). Ultrasound examination at 16 weeks of pregnancy, selective ultrasonography for detection of fetal abnormalities with General Electric Echograph Voluson E8, amniocentesis, fetal chromosomal analysis and genetic counselling was done.
3. Results and Discussion

After the prenatal ultrasound and genetic investigations the following diagnosis was established: Pregnancy 18 weeks in evolution Porencephaly. After a complex genetic counselling the parents opted to terminate the pregnancy. The prenatal diagnosis was confirmed by fetal autopsy. Genetic counseling is appropriate in familial and genetically determined porencephaly. Special attention in genetic counseling must be paid for families carrying COL4A mutations, given the variable phenotypic expression and penetrance [6].

4. Conclusion

Prenatal ultrasound investigation and genetic testing is absolutely necessary for the early prenatal diagnosis of severe malformations.

References


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