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Prenatal diagnosis of rare Aicardi and Anderman syndromes and management tactics

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Objective

Prenatal assessment analysis diagnosis, frequency, structure of concomitant perinatal outcomes in agenesis of the corpus callosum.

Methods

"A comprehensive clinical-instrumental, ultrasound, laboratory study was carried out on the basis of the Republican Center ""Screening and the Child"" of the Republic of Uzbekistan from 2021 to 2023. In order to study the early diagnosis of Aicardi's syndrome and Anderman's syndrome a retrospective analysis of the results of a comprehensive dynamic examination of 21 pregnant women complicated by agenesis of the corpus callosum in the fetus was carried out.

Results

According to the results of the work done of the 21 cases, 11 were terminated in connection with the identified anomalies. 10 pregnancies ended in childbirth - 9 term births and 1 premature. More than half of the examined pregnant women (66.7%) were at a young age - up to 25 years, dominated by residents of the region - 60%. Primiparous 40%, multiparous - 60%. Of the complications of pregnancy, there were acute respiratory viral diseases in % of cases, the threat of interruption in 58%, infections transmitted sexually - 58%. Prenatally, during the ultrasound examination of the fetuses, the diagnosis of agenesis of the corpus callosum was made in all cases, the average time of diagnosis – 24±4 weeks. Agenesis of the corpus callosum was detected at 24–28 weeks. Among the 21 included cases, 8 had arachnoid cysts associated with curvature of the interhemispheric fissure associated with complete or partial agenesis of the corpus callosum. The triad in the presence of other CNS malformations such as polymicrogyria, heterotopias, ventriculomegaly in each case, cerebral asymmetry, and abnormalities outside the CNS represent the characteristic presentation of Aicardi's syndrome in the patient. Agenesis of the corpus callosum was combined with other congenital malformations of the central nervous system in 11 cases: in 9 with internal hydrocephalus, in 1 with encephalomeningocele, in 1 case with porencephalic cysts. Among the concomitant pathology of other organs and systems 2 cases of congenital heart defects were identified, 2 cases of kidney anomaly, 1 case of diaphragmatic hernia, 1 case of limb defects. Only in two cases, agenesis of the corpus callosum was an isolated malformation of the central nervous system, not accompanied by concomitant pathology of other organs and systems. Anderman's syndrome was confirmed in two cases.

Conclusion

Aicardi syndrome proposed diagnostic triad: corpus callosum agenesis, inter-hemispheric cyst and distortion of the inter-hemispheric fissure. Anderman's syndrome congenital heart defects were identified, kidney anomaly, diaphragmatic hernia. To confirm the prenatal diagnosis of agenesis of the corpus callosum, differential diagnostics allows the combination of the results of the CNS and extra-CNS in the fetus to outline the characteristic pattern of visualization of agenesis of the corpus callosum, and choose the correct tactics for managing pregnancy.