Gastrointestinal tract in association with heterozygous CFTR gene mutation G 1716A

Mladenović - Segedi Lj, Segedi D, Bogavac M, Bjelica A
University of Novi Sad, Medical faculty, Clinical centre Vojvodina, Department of obstetrics and gynecology Novi Sad, Serbia, Novi Sad, Serbia

Objective

The aim of our study was to present a case of progressive dilatation of fetal gastrointestinal tract (GIT) in the second trimester of pregnancy, which is associated with heterozygous CFTR gene mutation G 1716A.

Methods

Retrospective descriptive analysis of the case records from the Clinical Center, Clinic of Gynecology and Obstetrics, Novi Sad.

Results

Nulliparous 29 year old women with a low risk for aneuploidy in first trimester screening. The ultrasound examination a week 24 of pregnancy showed initial segmental dilatation of the distal region of GIT and of anal region of GIT, respectively. In the next two weeks, additional segmental dilatation of the distal GIT had progressed (anze width up to 10 mm and lengths up to 40 mm), and had developed multiple graded dilatation of the jejo-ileum. Extraintestinal anomalies were not present. There was a suspicion of anal atresia and cystic fibrosis amniocentesis was performed with genetic analysis of both parents. Since the fetal bowel dilatation progressively increased and occupied the new segments of the small intestine, there was the threat of meconial ileus. It was therefore decided to terminate the pregnancy in week 27. An autopsy of the fetus did not prove the existence of anal atresia or atresia of any other part of GIT. Genetic analysis confirmed that both mother and fetus have a heterozygous mutation in the CFTR gene G 1716 A. This mutation in the CFTR gene is itself associated with cystic fibrosis, so there is the possibility of the presence of other autosomal recessive mutations.

Conclusion

Whole genome sequencing in families with autosomal recessive pattern of inheritance should be done in order to identify new mutations with a role in development of GIT atresia, which would have significant prenatal diagnostic value.