A case of Waardenburg syndrome

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Objective

To present a prenatally diagnosed Waardenburg syndrome type 4 (WS4) newborn with a novel mutation in EDN3 gene through the amniotic fluid genetic examination.

Methods

The antenatal record of a case of prenatally diagnosed Waardenburg syndrome type 4 reviewed retrospectively.

Results

A 27-year-old pregnant woman was referred to a specialized perinatologist for second-trimester detailed ultrasonographic examination and genetic counseling because of intestinal enlargements and an obstetric history of hearing loss and constipation in siblings. Parents were healthy and non-consanguineous. On USG evaluation, there was prenasal skin thickening and large intestinal loops. Amniocentesis revealed a novel frameshift homozygous mutation of the EDN3 gene p.D104Gfs*63 (c.307_308) ins A gene causing early stop codon. At term, a 2820 g, 50 cm male boy was delivered by cesarean section. In physical examination, he had a white forelock hair on the frontal side and hypopigmented skin patch on the chest wall. Meconium passage was delayed, and progressive abdominal distension was observed. Abdominal radiography with water soluble contrast enema showed marked colonic dilatation with a narrow transition zone and deletion of haustra. Retained barium contrast in the bowel was seen on delayed film which was obtained 48 h after the first colon graph. Biopsies were taken during laparotomy, and they indicated total colonic aganglionosis. The aganglionic segment was removed surgically, and ileostomy was created. He also had sensorineural hearing loss. With this genetic and clinical features, the diagnosis of WS4 was confirmed. The patient had enteral feeding intolerance therefore was on total parenteral nutrition. On the 56th day postpartum, he died due to sepsis.

Conclusion

Up to now, approximately 80 cases of Shah-Waardenburg syndrome have been reported in the literature. The case is presented because of it's rare occurrence and prenatally detected novel mutation on the genetic basis.