

Case of peters-plus
syndrome. the
parents were not



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Case Report A 23-years-old woman gravida 1 partus 0, referred to our clinic at the 21st week of her pregnancy due to suspicion of fetal hydrocephalus. The fetal biometry scan showed appropriate biparietal diameter, head circumference, and abdominal circumference measurements, while long bone measurements, including femur, fibula, radius, and ulna were one week shorter than the expected gestational age. The detailed scan showed a female fetus with moderate ventriculomegaly, absence of cavum septum pellucidum, dilated third ventricle, bilateral echogenic lenses, retrognathia (inferior mandibular angle $<50^\circ$), hypotelorism (binocular distance at 5th percentile and interocular distance at 50th percentile) and microphthalmia (ocular diameter <5 th percentile) (Figure 1). Karyotyping and fetal magnetic resonance imaging (MRI) scheduled; considering these findings may be related to a chromosomal anomaly or a syndrome.

Fetal MRI showed agenesis of the corpus callosum, ventriculomegaly, hypotelorism and bilateral congenital cataracts. Amniocentesis showed 46, XX chromosomes. An intrauterine fetal death occurred at the 23rd gestational week. A 500 gram, female fetus delivered vaginally after cervical preparation and proper induction. Pathologic autopsy showed narrow palpebral fissures, a long philtrum, cupid's bow upper lips with a thin vermilion border and facial hirsutism and low set ears (Figure 2), bilateral absence of corneal endothelium and descemet membrane, bilateral optic nerve degeneration (Figure 3) bilateral cataracts, agenesis of the corpus callosum and hydrocephalus. The autopsy council, including ophthalmologists, confirms the diagnosis of Peters-Plus syndrome. The

parents were not consanguineous, and their relatives did not indicate a history of such anomalies.

Further array analyses on chromosome 13q12. 3 did not reveal any mutations in the beta-1, 3-galactosyltransferase-like gene (B3GALTL).

Discussion Peters' anomaly is a rare congenital ocular anomaly caused by defective dysgenesis and cleavage of the anterior chamber of the eye causing central corneal opacity (leukoma), the absence of the posterior corneal stroma and Descemet membrane and a variable degree of iris and lenticular attachments to the central aspect of the posterior cornea (1). If typical ocular anomalies of Peters' anomaly accompanies with multiple malformations, this situation referred to as Peters-plus syndrome (2). Peters-plus syndrome previously known as Krause-kivlin syndrome or Peters' anomaly with short-limb dwarfism (OMIM ? 261540) is an autosomal-recessive inherited congenital disorder caused by a mutation in the B3GALTL gene on chromosome 13q12.

Peters-plus syndrome is a rare anomaly, with no known incidence, with equal sex ratio and a high incidence of consanguinity and with reported a bit more 70 cases in the postnatal period and eight cases in the prenatal period (3-7).

The classic triad of Peters-plus syndrome includes anterior segment defects (100%), short stature (100%) and brachydactyly (95%) (8). The clinical features of Peters-Plus syndrome includes a prominent forehead, narrow palpebral fissures, a long philtrum, cupid's bow upper lips, cleft lip and palate, preauricular ribs, micrognathia, a broad neck, cataract and glaucoma, short limbs, brachydactyly, clinodactyly, microcephaly, brain atrophy,

agenesis of the corpus callosum and variable developmental delay and
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intellectual disability. Some of these features may be present prenatally or at birth, and some of them may occur at later ages (3). In prenatally detected cases of the Peters-Plus syndrome, including our case no B3GALTL gene mutation reported until now.

(6). There can be some explanations for this situation; these cases may be a variant or a phenotypic overlap of the Peters-plus syndrome (9) or these cases may carry a distinct mutation, which needs further investigations. Prenatal detection of Peters-plus syndrome requires a handful of dedicated clinicians, including obstetricians, ophthalmologists, genetic specialists, and pathologists. Particularly, obstetricians and ultrasonographers should take attention to the eye, and if ocular anomalies suspected, craniofacial and skeletal system and fetal growth should also be controlled, in the prenatal detection of the Peters-plus syndrome. In the differential diagnosis of Peters-plus syndrome similar syndromes like SHORT, Abbruzo-Erickson, GMS, Weill-Marchesani, Michels, Rieger, Walker-Warburg, Cornelia de Lange, Robinow and Fetal alcohol syndrome can be detected according to Orphanet data ([http: 77www. orpha.](http://www.orphana.net)

net). Herein we report the first case of prenatal detection of the Peters-plus syndrome from Turkey in a patient without any family history. The diagnosis made solely by prenatal ultrasound despite normal fetal chromosomes and no mutation in the B3GALTL gene in this sporadic case. As a result, in case of fetal anterior segment defects, obstetricians can remember Peter's anomaly, Peters-plus syndrome and Peters-plus syndrome like-phenotypes, and they can also scan other accompanying features of these diseases, and they can perform prenatal invasive tests, including specific gene mutations.
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