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Editorial note

Heterozygote PACS1 Mutation in Schuurs-Hoeijmakers Syndrome

Dilber B*

Karadeniz Technical University, Department of Pediatric Neurology, Turkey

EDITORIAL NOTE

PACS1 gene is found on the long arm of the 11th chromosome (11q13.1-13.2). Mutations showing autosomal dominant inheritance in this gene cause Schuurs-Hoeijmakers syndrome. For the first time, Schuurs-Hoeijmakers et al. diagnosed PACS1 with de novo mutation because of similar findings in two boys who had no cognation. These boys had findings such as similar typical facial appearance, intellectual and motor retardation, and cryptorchidism. De novo c.607C>T mutation was detected in both boys, and clinical pictures were overlapped. Later, three further patients with similar findings were described by Gadzicki et al. in 2014, PACS1 associated symptoms were introduced with their findings. Reported de novo missense PACS1 (c. 608G>A,p.Arg203Gln) mutation. In our case, heterozygous missense mutation c.607C>T was detected in PACS1 gene and the patient was diagnosed with whole exome sequencing (WES). We underlined that this chromosomal abnormality presents with typical facial appearance, retardation in speech skill, and motor skills. Furthermore, we stated that this condition may be found in a wide spectrum and accompanied by several anomalies. However, we establish the diagnosed with WES, which we have sent considering typical facial appearance and mild motor retardation that were prominent in our patient. We thought that our contribution to these patients is raising awareness for the diagnosis, and provide facility for the problems that can be experienced in their families and for future follow-up. We present the first description of PACS1 mutation in Turkey.

Cranial nerve migration anomalies resulted from replacement of arginine by tryptophane in furin binding region are thought to play a role in the etiopathogenesis of PACS1, and similar facial findings are explained by zebrafish method. In our case, although there were difficulties in the differential diagnosis, because of the characteristic facial findings that were compatible with Baraitser Winter syndrome, Cornelia de Lange syndrome, Mowat-Wilson syndrome, and Kabuki syndrome; we found that differences and similarities were slightly different and confirmed the diagnosis with WES.

This PACS1 mutation is characterized by mental retardation, prominent craniofacial characteristics hypertelorism, mild ptosis, a large mouth, depressed and wide nasal bridge, long philtrum, flat eyebrows, downward localized eyelids, prominent ears, and thin upper lip), psychomotor retardation, mild-to-moderate intellectual retardation, inability to speak, hypotonia, seizures, structural malformations (heart, brain, eyes, kidneys, and orthopedics etc), and additional congenital anomalies. In WES analysis performed in our patient, we detected missense mutation c.607C>T (p.R203W) was detected in PACS1 gene (heterozygous). This alteration was also showed as heterozygous in DNA sequencing analysis with Sanger method. The parents were also screened and found to be normal.

***Correspondence to: Dilber B**, Karadeniz Technical University, Department of Pediatric Neurology,Turkey, E-mail: beriltem@gmail.com

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