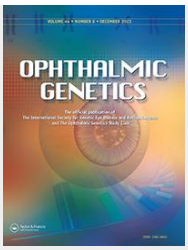

Ophthalmic Genetics

PHARC syndrome which an ultra-rare syndrome with retinitis pigmentosa and cataracts: case report and review of the literature

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Background

PHARC syndrome (MIM:612674) is a rare neurodegenerative disorder characterized by demyelinating polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataracts (PHARC). The syndrome is caused by mutations in the *ABHD12* gene, which encodes αβ-hydrolase domain-containing protein 12 related to endocannabinoid metabolism. PHARC syndrome is one of the rare diseases; so far, only 51 patients have been reported in the literature.

Methods

We evaluated the 25-year-old male patient referred to us due to vision loss, cataracts, and hearing loss. Ophthalmological examinations and genetic analyses were performed using targeted next-generation sequencing.

Results

In the genetic analysis, the patient was diagnosed with PHARC syndrome by detecting homozygous (NM_001042472.3): c.871del (p.Tyr291IlefsTer28) novel pathogenic variation in the *ABHD12* gene. Following the molecular diagnosis, he was referred to the neurology department for reverse phenotyping and sensorimotor demyelinating polyneuropathy was detected in the neurological evaluation.

Conclusions

In this study, we report a novel variation in ABHD12 gene in the first Turkish-origin PHARC patient. We present this study to contribute genotype-phenotype correlation of PHARC syndrome and emphasize the importance of molecular genetic diagnosis in order to determine the appropriate clinical approach. This report is essential for expanding the phenotypic spectrum in different populations and understanding the

genotype-phenotype correlation of PHARC syndrome via novel pathogenic variation in the ABHD12 gene.

KEYWORDS:

- Retinitis pigmentosa
- cataracts
- PHARC syndrome

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Disclosure statement

The authors report no conflicts of interest. The authors alone are responsible for the content and writing of this article.

Ethical approval

This study was performed in accordance with the Declaration of Helsinki Principles. Written informed consent was obtained from the patient for the publication of this report and accompanying images. This study was approved by the medical ethics committee of Marmara University School of Medicine (No: 09.2022.624).

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