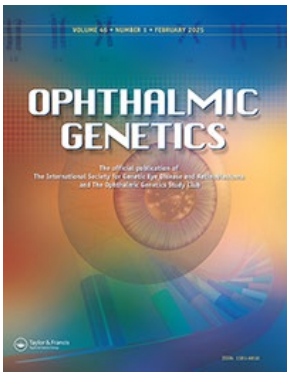

Case report

Intellectual disability and retinitis pigmentosa due to a homozygous null *SCAPER* variant: a clinical and genetic insight with review of the literature

Zehra Manav Yiğit, Osman Semih Dikbaş, Erol Erkan, Gözde Şahin Vural, Gökay Bozkurt

Received 26 Dec 2024, Accepted 20 Mar 2025, Published online: 30 Mar 2025

- https://doi.org/10.1080/13816810.2025.2485222

ABSTRACT

Introduction

Variations in the SCAPER gene are associated with Intellectual Developmental Disorder and Retinitis Pigmentosa (IDDRP), characterized by visual and neurological symptoms. Despite limited data, SCAPER plays a critical role in cell cycle regulation and ciliary function, which may explain its diverse phenotypic effects. This study aims to report a homozygous NM_020843.4: c.2605 A>T; p.(Lys869*) nonsense variant in SCAPER gene, expanding the phenotypic spectrum of IDDRP and contributing to its clinical and genetic understanding.

Methods

Genetic testing and multidisciplinary evaluations were performed on an 11-year-old girl with intellectual disability, retinitis pigmentosa, and dysmorphic features. Clinical exome sequencing identified a homozygous null SCAPER variant, confirmed by Sanger sequencing.

Results

Clinical findings revealed bilateral epiretinal membranes, thinning of the ellipsoid zone, and hyperfluorescent rings in fundus autofluorescence imaging. Neurological evaluation showed intellectual disability, ADHD, and corpus callosum abnormalities. Skeletal anomalies, including short stature and genu valgum, were also noted. The variant was classified as likely pathogenic based on ACMG guidelines.

Discussion

This report describes the first case of a homozygous c.2605 A>T variant in SCAPER, highlighting its role in ciliary and cell cycle dynamics. These findings contribute to a better understanding of SCAPER-related phenotypes and emphasize the importance of genetic testing in similar cases.

KEYWORDS:

- ***SCAPER***
- **retinitis pigmentosa**
- **rare variant**
- **nonsense**
- **intellectual disability**