Abstract 47

EARLY-ONSET MYOPIA AND RETINAL DETACHMENT WITHOUT TYPICAL MICROCORIA OR SEVERE PROTEINURIA IN PIERSON SYNDROME DUE TO A NOVEL LAMB2 VARIANT

Poster

Magliyah M., Alshamrani A.*, Alsulaiman S.

King Khaled Eye Specialist Hospital ~ Riyadh ~ Saudi Arabia

Purpose:

Background: Pierson syndrome is a genetic disease first described as microcoria and congenital nephrotic syndrome that progresses to fatal renal failure. It results from mutations in the laminin B2 gene (LAMB2), one of the glycoproteins that are expressed in the basement membrane of glomeruli, ocular structures, and neuromuscular structures.

Methods:

Purpose:To describe the ocular and renal features as well as outcomes of retinal detachment repair in patients with a novel homozygous LAMB2 mutation.
Methods:The eyes, urine, and serum DNA were evaluated in the affected patients.
Results:Eleven patients (22 eyes) from 4 families with a mean age at presentation of 6.0 years (range 1 to 26 years) were included. All were confirmed to have a homozygous variant c.619T>C p.(Ser207Pro) in the LAMB2 gene. None of the study eyes had microcoria, and none of the patients had nephrotic range proteinuria. Seven patients were tested for proteinuria, and all demonstrated mild proteinuria <100 mg/dL

Results:

All patients had moderate to high axial myopia, with a mean refraction of -5.6 diopters(range -4.0 to -9.0 diopters). Among 15 eyes with clear view to the fundus, tessellated myopic fundus, avascular peripheral retina evident clinically or on fluorescein angiography, and rudimentary fovea were universal (100% of the eyes). Optic disc pallor was observed in 10 eyes(66.7%). Peripapillary atrophy was present in only 1 eye (6.7%). Straightened retinal vessels, abnormal vascular emanation from the optic disc, supernumerary vascular branching at the optic disc and vascular tortuosity were observed in 10 (66.7%), 2 (13.4%), 2 (13.4%), and 2 (13.4%) eyes respectively.

Conclusions:

Conclusions: This study describes a distinct phenotype of Pierson syndrome with a novel homozygous LAMB2 mutation and further expands the spectrum of ophthalmic and renal features, and molecular genetic basis of LAMB2-related disease. The retinal features may be the only clue to suspect the diagnosis.