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ABSTRACT

Purpose

To evaluate the clinical and genetic spectrum of inherited retinal diseases (IRDs) in a Kuwaiti tribe.

Methods

Forty-four patients with IRDs from 28 nuclear families from the tribe, were evaluated for presenting symptoms, visual acuity, fundus examination, OCT, microperimetry, full-field (ff), and multifocal electroretinography (mERG) and genotyping.

The ffERG and mERG were performed with the **Metrovision** MonoPackOne model system using ERG-jet contact lens electrodes following the standards, guidelines, and extended protocols of the International Society for Clinical Electrophysiology of Vision

Results

Seventeen patients were diagnosed with autosomal recessive retinitis pigmentosa (arRP) associated with *RP1* c.606C>A with onset of nyctalopia in the third decade, myopia, and macular atrophy by the age of 50; eleven with autosomal recessive cone/rod dystrophy or macular dystrophy associated with *RP1* c.606C>A (p.Asp202Glu) mutation with color and central vision deterioration in teenage, myopia, paracentral ring scotoma and macular atrophy; eleven were with arRP associated with *PDE6B* c.992 + 1 G > A mutation with onset around 5 years, myopia, cataract, retained central fixation, and ellipsoid zone and late perimacular atrophy; five—with Leber congenital amaurosis associated with homozygous *RPGRIP1* for c.1107delA mutation with extinguished

ffERG and electrophysiological phenotype of rod and cone; and one patient—with autosomal recessive rod-cone dystrophy associated with homozygous *PDE6B* c.992 + 1 G > A, who was homozygous *ABCA4* c.5882 G > A and heterozygous *EYS*; c.2137 + 1 G > A.

Conclusions

This study represents a typical tribe from the Middle East with high rate of consanguinity for many generations that harbors multiple mutated genes associated with IRD. It demonstrates the predominant phenotype and its variability in retinal disorders caused by identical mutations and illustrates the nuances in the clinical presentation and disease progression of patients with pathogenic mutations in more than one gene.

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