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Familial exudative vitreoretinopathy in a 4 generations family of South-East Asian Descendent with *FZD4* mutation (c.1501_1502del)

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Abstract

Background: Familial Exudative Vitreoretinopathy (FEVR) is a hereditary disorder characterized by peripheral avascular retina with neovascularization. Although FEVR has been thoroughly described in multiple literature publications from different countries, there are currently limited articles describing the phenotypes of FEVR among South-East Asian Descendent. This paper describes the clinical phenotype of the *FZD4* gene with c.1501_1502 deletion in a 4-generation case series of a South East Asian family.

Methods: We reviewed a 4-generation case series of a South-East Asian descendent family consisting of 27 family members with 10 members diagnosed with FEVR. We observed the clinical phenotype of these series of patients, including some of the family members who underwent whole-exome sequencing, PCR amplification and DNA sequencing techniques to identify the mutated gene.

Results: Frameshift mutation (c.1501_1502del) were found in *FZD4* gene in this series of patients with the age ranging from 1 month old to 69 years old. There was a 100% (4/4) of our paediatric patients being diagnosed within 21 days of life. It was also found that 75% of patients (6/8) less than 40 years old exhibited disease asymmetry of 2 stages or more and 80% (8/10) had a history of vitreoretinal surgery or diode laser photocoagulation, with a further 50% of the adult patients identified as legally blind; the mean age of blindness was 18-years-old.

Conclusions: Phenotypic manifestation of *FZD4* gene with c.1501_1502del mutation can be identified within the neonatal period. They have relatively greater clinical asymmetry of 2 stages or more compared to other mutations. Without treatment, most of them will have bilateral severe visual impairment around the adolescent age group.

Keywords: Familial exudative vitreoretinopathy, FEVR, FZD4 genes

Background

Familial exudative vitreoretinopathy (FEVR) was first described by Criswick and Schepens in 1969 as congenital, bilateral vitreoretinopathy with no history of premature birth and oxygen therapy [1]. FEVR is characterized by the peripheral avascular retina and subsequently lead to complication due to retina ischemia [2]. These includes peripheral neovascularization, vitreous haemorrhage, retinal traction with temporal dragging, macular dragging, falciform retinal fold and retinal detachment [2, 3].

FEVR can be inherited in different modes which include autosomal dominant, autosomal recessive and

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