Case Report

Jalili Syndrome

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A thirty years old Pakistani male was referred from University of Lahore, Pakistan for clinical evaluation and interpretation of his ERG. Clinical examination revealed Cone – Rod dystrophy. Systemic examination showed discolored and abnormally shaped teeth. The patient was otherwise mentally and physically normal. His ERG showed Scotopic (rod responses) within upper normal limits in right eye while markedly reduced in the left eye. Scotopic rod and cone combined responses (indicating generalized retinal functions) were also decreased. Photopic (cone responses) were reduced in both eyes, with more marked fall in the left eye as compared to the right eye.
Key Words: Rod-cone dystrophy, amelogenesis imperfect, Electro-retinogram, pigmentary retinopathy

Jalili Syndrome is a rare cone-rod dystrophy which is associated with amelogenesis imperfecta. Few cases are reported in literature so far. The very first report of Jalili syndrome was published in 1988 by an Iraqi ophthalmologist named Ismail K. Jalili. Jalili and Smith reported 29 individuals from a single highly inbred Arab family from the Gaza strip. Later, other families with this syndrome were reported mainly from middle east. Only one case from North America and Indian sub-continent are reported. We report a case of Jalili syndrome from northern areas of Pakistan. Environmental factors and genetic association of this syndrome is also mentioned with reference to the hypotheses already presented in different papers globally.

CASE REPORT

We present a case of thirty years old Pakistani male, referred from University of Lahore, Pakistan for clinical evaluation and interpretation of his ERG. For reporting of this case, informed consent was taken, according to the principles of the Declaration of Helsinki. The patient did not give consent to publish his face photographs but allowed the publication of fundus pictures and Electro retinogram.

History revealed that the patient complained of decreased visual acuity in both eyes since childhood. He was prescribed glasses but the compliance was poor. Day and night vision were equally affected. There was no irritation and redness in both eyes. Systemic history revealed discolored and abnormally shaped teeth. There was no history of any medicine intake specially Tetracycline group of drugs. Family consanguinity was positive but there was no history of such disease in any of the relatives. He had two brothers and one sister and all were normal with no ocular and systemic disease.

On examination, he was an average built male who was well oriented in time and space. He was Orthotropic. Auto-refraction showed mild myopia. His best corrected visual acuity was finger counting in each eye. Anterior segment showed no abnormality. On dilated fundus examination, there were tilted oval discs in both eyes. There was arteriolar attenuation and bone spicule pigmentation scattered all over the retina. Pigmentation was also visible in the macular area. Large choroidal vessels were visible.

General physical examination showed normal vital signs. Teeth were yellow in color and distorted in shape. Clinical features led to the diagnosis of Jalili syndrome.

His ERG showed Scotopic (rod responses) within upper normal limits in right eye while markedly reduced in the left eye. Scotopic rod and cone combined responses (indicating generalized retinal functions) were also decreased. Photopic (cone

responses) were reduced in both eyes, with more marked fall in the left eye as compared to the right eye.

Fig. 1: Cone rod dystrophy in a patient with Jalili Syndrome. Macular excavation, arteriolar attenuation and pigmentary retinopathy.

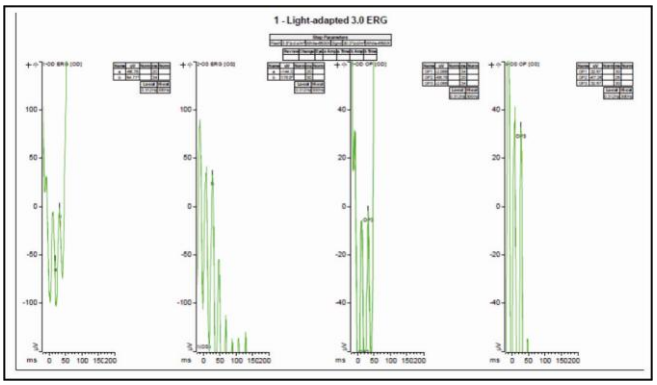

Fig. 2a: Light adapted ERG.

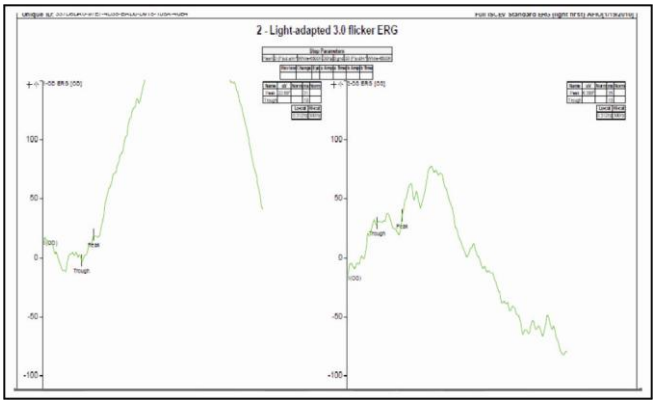

Fig. 2b: Light adapted flicker ERG.

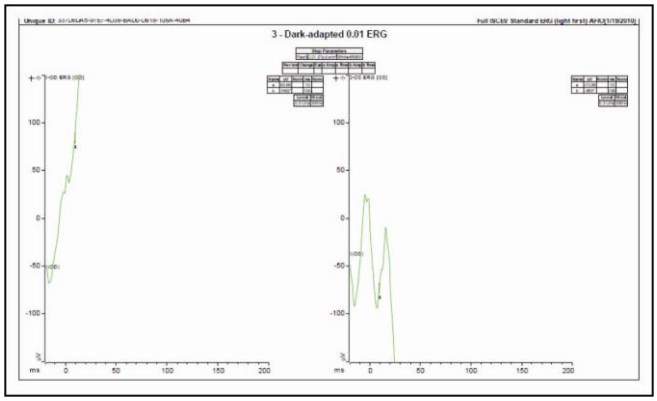

Fig. 2c: Dark adapted ERG.

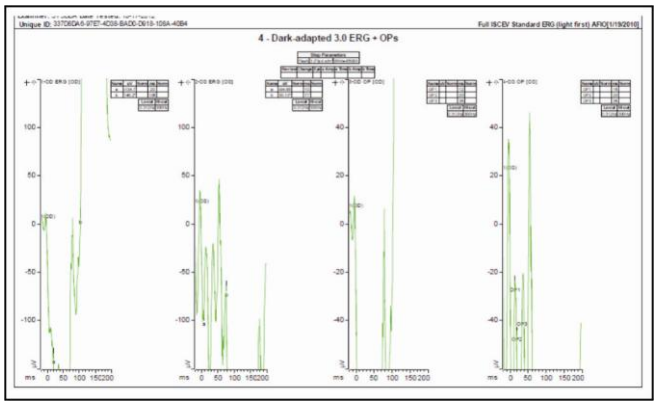

Fig. 2d: Dark adapted ERG + Ops.

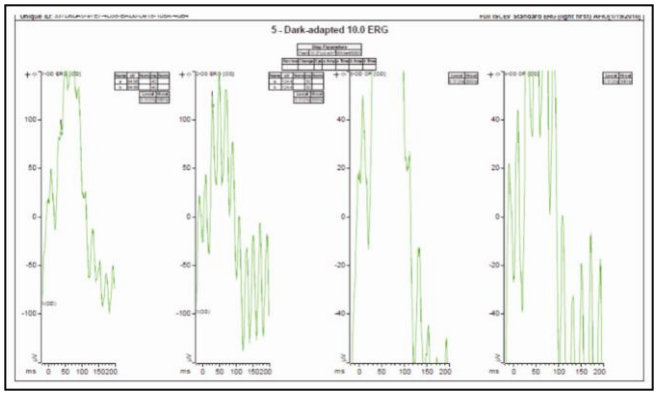

Fig. 2e: Dark adapted 10 ERG

DISCUSSION

Cone – rod dystrophies are part of a genetically diverse group of progressive photoreceptor disorders, which are categorized on the basis of the photoreceptor cells primarily involved in the disease process. Three main groups are identified; cone-rod, rod-cone, and mixed receptors dystrophies. Jalili

syndrome is a cone-rod dystrophy which is associated with amelogenesis imperfecta.

Amelogenesis was defined by Crawford *et al.* as "a group of conditions of genetic origin that affect the structure and clinical appearance of enamel of all or almost all of the dentition, and that may be associated with morphological or biochemical changes in other parts of the body".[1]

In cone-rod dystrophy, there is initial involvement of cone dysfunction; loss of central vision, color vision and photophobia.[2]

Jalili syndrome was first reported in 1988 by an Iraqi ophthalmologist named Ismail K. Jalili. Jalili and Smith reported 29 individuals from a single highly inbred Arab family from the Gaza strip. All patients had photophobia, loss of color vision but normal night vision. Teeth of all the individuals were discolored and malformed from the very beginning.[3] Three phenotypic variations were identified in Gaza Strip. Type A had early onset macular lesion leading to macular excavation and coloboma in early age. Type B had more peripheral involvement and had resemblance to Retinitis Pigmentosa but without night blindness. Third type C was similar to type A, but it appeared in late age. Our patient had early onset of retinal signs and had excavated type of maculopathy similar to type A.

Initially, Jalili syndrome was only found in the Gaza Strip and not in the West Bank[4]. Different environmental and genetic factors are described in literature which can lead to Jalili syndrome. One such environmental factor is family consanguinity in people of Gaza strip. Our patient also had a strong history of intra family marriages.

Another important factor regarding the Jalili syndrome was hypothesized to be high fluoride levels in ground water of Gaza strip. This resulted in dental fluorosis[5]. Literature shows that high fluoride levels in water are toxic and the toxicity is dose dependent.[6] It was also hypothesized that a disrupted magnesium transport was involved in the development of the dental abnormalities observed in Jalili syndrome[7]. Unfortunately, we were not able to get the Fluoride and Magnesium levels in ground water of that area (our patient's residence).

Genetic factors are well recognized for this disease. Nine mutations are described in literature; three mis-sense changes, three termination mutations, two large deletions, and a single base insertion.[8]

Recently, more cases were reported from other parts of the world. In 2013 first family of Jalili syndrome was identified in North America.[9] There was only one case of Jalili syndrome reported from Sub continent[10]. He had situs inversus totalis, keratoconus and ectopia lentis. He belonged to an area with high fluoride levels in the ground water and a positive history of consanguineous marriage among his family members. There were no such ocular abnormalities in our patient.

Another case of a 9-year-old child with neurofibromatosis type 1 (NF1) and Jalili syndrome was reported in the literature[11]. Similarly, different phenotypes were also seen in the same family in a study.[12]

The short comings in our case report were lack of genetic study and chemical analysis of ground water.

CONCLUSION

Our literature search found only one case reported from the Indo-Pak sub-continent. Family consanguinity and environmental factors favor the prevalence of Jalili Syndrome in our part of the world but few reports might be because of under diagnosis of the disease.

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