WAGNER SYNDROME: A REVIEW OF A RARE VITREORETINOPATHY

Authors: Amanda Geara[1], Carolina Rebello Hilgert[2], Carlos Augusto Moreira-Neto[3], Carlos Augusto Moreira Junior[3]

(1) Retina and Vitreous Fellow at Instituto Professor Moreira, Hospital de Olhos do Paraná - Curitiba, Paraná
(2) Resident at Instituto Professor Moreira, Hospital de Olhos do Paraná - Curitiba, Paraná
(3) Ophthalmologist, Retina and Vitreous specialist at Hospital de Olhos do Paraná– Curitiba – PR, Brazil.

PURPOSE

To report a case of Wagner Syndrome (WS), a rare vitreoretinopathy.

METHODS

Case report through analysis of medical records and multimodal exams.

RESULTS

A 29 year-old male patient presented a 2-day history of abrupt-onset low visual acuity (VA) and inferonasal hemifield loss in the left eye (OS). He denied previous pathological history and reported blindness in a brother and a cousin due to retinal detachment (RD).

The best corrected VA in the right eye (OD) was 20/20 and counting fingers in OS. Biomicroscopy revealed cataract in OS. Tonometry was normal in both eyes (OU). Fundoscopy showed peripheral retina degeneration and multiple retinal breaks OU, in addition to an inferior macula-off RD in OS. Wide-angle retinography was requested (fig 1-3).

A prophylactic 360º-laser was made in OD. In addition to phacoemulsification, intraocular lens implant, and triamcinolone intravitreous injection, pars plana vitrectomy PPV with silicone oil and scleral buckle were performed in OS. Based on clinical features a presumed diagnosis of WS was made. Although instructed about family screening and genetic counseling, the patient declined the request of genetic testing and is being clinically followed.

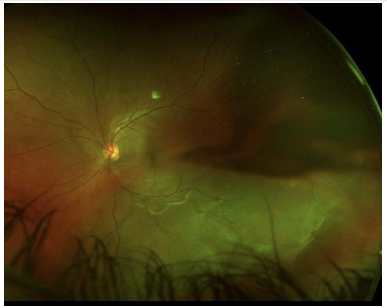

Fig.1 wide-angle retinography: pre-op OS: inferior macula-off retinal detachment, peripheral retina degeneration and multiple retinal breaks.

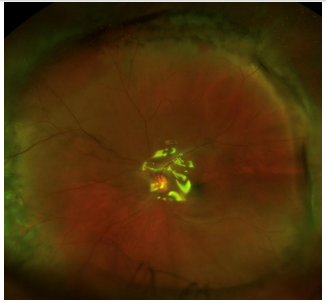

Fig.2 wide-angle retinography: post-op OS: posterior pole silicon oil reflex, scleral buckle introflection reflex, 360º laser, peripheral retina degeneration and multiple retinal breaks

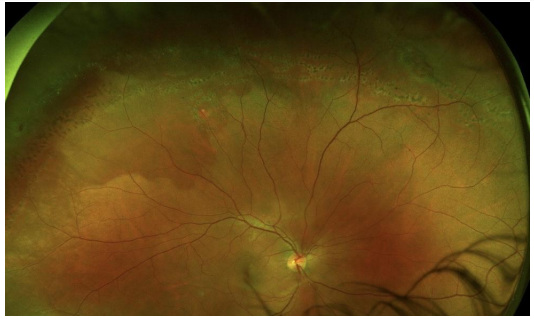

Fig.3 wide-angle retinography: post-laser OD laser 360º, retina degeneration and multiple retinal breaks

DISCUSSION

WS is a rare (estimated prevalence is less than 1:1,000.000) autosomal dominant vitreoretinopathy caused by a mutation in VCAN gene, resulting in aberrant forms of versican. This mutation leads to great reduction of the amount of glycosaminoglycans in proteins, and therefore the premature liquefaction of vitreous and an optically empty vitreous. Other ocular findings are vitreous membranes, avascular vitreous veins, presenile cataract, myopia, retinal breaks and RD. Systemic features are not found in WS, different of Stickler Syndrome, another disease in the hall of vitreoretinopathys. Full-field ERG is attenuated and visual field testing can show constricted patterns. OCT may demonstrate outer retinal disruption, thinning of all retinal layers and high reflectivity membranes at the vitreoretinal surface. The diagnosis of WS is established on a patient with a positive family history (FH) and the corresponding clinical findings. In case of no FH, a genetic testing for mutation of the VCAN is suggested. A vitreoretinal specialist should follow the patient annually and a referral to genetic assessment is recommended. The immediate family of any patient diagnosed with WS should be thoroughly evaluated for subclinical abnormalities.

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