1 de Dezembro **08h30 | 10h00 – Sala 1**

Retina Médica | Medical Retina

Moderadores | Chairs: Maria Luz Cachulo (CHUC), Diogo Cabral (HGO), Luis Mendonça (HB)

CO 12

OPTICAL COHERENCE TOMOGRAPHY (OCT) AND OPTICAL COHERENCE TOMOGRAPHY ANGIOGRAPHY (OCT-A) FEATURES IN PATIENTS WITH FACIOSCAPULOHUMERAL MUSCULAR DYSTROP

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Introduction and Objectives: Facioscapulohumeral muscular dystrophy (FSHD) is one of the most common muscular dystrophies, with an incidence of 1/ 20,000. It is an autosomal dominant disease characterized by its muscle involvement pattern, which is typically asymmetric, involving facial muscles, shoulder girdles, and upper arms. Disease severity is highly variable and progression to other muscles is slow, but the overall lifespan is not shortened. Patients may also present with extramuscular manifestations, such as vision and hearing loss, arrhythmias, intellectual disability and epilepsy.
About half of patients have been reported to have asymptomatic peripheral retinal vascular abnormalities, such as telangiectasias, microaneurysms, and capillary nonperfusion. Vessel irregularities and tortuosity are also common but reports concerning macular edema as a complication of these vascular changes are rare. Nonetheless, severe cases of intraretinal and subretinal exudative retinopathy, a condition resembling Coats disease, may occur.
Non-invasive imaging techniques such as OCT and OCTA provide a detailed imaging of retinal structure and may provide complementary information on retinal pathology in FSHD.
The objective of this study is to describe retinal findings using OCT and OCTA in a cohort of FSHD patients, in order to increase the knowledge on the pathophysiology of the ophthalmological abnormalities of the disease.

Materials and Methods: Patients with FSHD were recruited from the neuromuscular disease consultation at the Neurology Department of Hospital de São João from January to March 2022.
All patients underwent a thorough evaluation, including a clinical assessment with refraction, BCVA and slit lamp exam and OCT and OCT-A were performed. Central macular and subfoveal choroidal thickness in EDI images were measured manually using the virtual ruler tool of the Heidelberg Eye Explorer Software. Automated RNFL thickness was registered. For OCT-A images, a posterior quantitative analysis was made using the Fiji software Image J, including vessel density measurements in superficial capillary plexus (SCP), intermediate capillary plexus (ICP), deep capillary plexus (DCP), choriocapilar and choroid and an analysis of the foveal avascular zone (FAZ) area, perimeter and circularity index.
A group of 10 healthy controls with matching age and gender was recruited.

Results and Discussion: A total of 22 eyes of 11 FSHD patients were examined. 7 patients were female (64%), and the mean age was 41 years (±20; 8–77).
Mean central macular thickness was lower in the FSHD group, with a mean value of 260 µm versus 278 µm in the control group ($p<0,05$). Subfoveal choroidal thickness and RNFL thickness showed no differences between the two groups, as well as vessel density for any of the analyzed layers. FAZ area in the DCP, FAZ perimeter in the ICP and FAZ circularity index in the DCP were significantly higher in the FSHD group ($p<0,05$). FAZ circularity in the ICP is reduced in the FSHD group ($p<0,05$).

Conclusion: To conclude, retinal abnormalities are frequent but almost always subclinical in patients with FSHD and, in addition to the microvascular changes described in the literature, include arterial tortuosity and foveal abnormalities. More studies need to be performed, but the importance of new non-invasive tests in the early detection, evaluation and follow-up of all patients with FSHD, even asymptomatic, is unquestionable.