Sorsby’s Fundus Dystrophy: Novel mutations, novel phenotypic characteristics and treatment outcomes

Supplementary Material

1. Supplementary Figure 1
2. Supplementary Figure 2
Supplementary Figure 1: Subfoveal Choroidal Thickness of Patients with Sorsby’s Fundus Dystrophy

Subfoveal choroidal thickness was measured using enhanced depth imaging optical coherence tomography. Patients with Sorsby’s fundus dystrophy (SFD) without visible fundus changes as well as those with drusen-like deposits revealed choroidal thicknesses measures within normal limits. Presence of choroidal neovascularization (CNV) had no impact on choroidal thickness. Patients with chorioretinal atrophy showed a marked thinning of subfoveal choroidal thickness. Continuous line marks the linear regression line of controls and the dotted line shows the 95% confidence interval.
Supplementary Figure 2: Late-Phase Indocyanine Green Angiography in Sorsby’s Fundus Dystrophy.

In subjects not carrying pathogenic TIMP3 mutations (controls) late-phase indocyanine green (ICG) angiography revealed a largely uniform fluorescence throughout the posterior pole 30-40 minute after dye injection (A-D). Asymptomatic
carriers of *TIMP3* mutations without visible fundus changes on other imaging modalities (E, F) as well as one subject with unilateral choroidal neovascularization and a contralateral normal appearing fundus (G) presented a distinct central hypofluorescence with multilobulated borders. Patients with drusen-like deposits showed a more pronounced hypofluorescence at the entire posterior pole (H-L). In two patients with later disease stage, there was a large area of reduced fluorescence with a more peripheral multilobulated border towards a peripheral zone of preserved fluorescence (M-N).