

Friedreich's ataxia

Optic [neuropathy](#) is a near-ubiquitous feature of [Friedreich's ataxia](#) (FRDA).

Friedreich's [ataxia](#) (FA) is the most frequent [hereditary ataxia](#) syndrome, while painful muscle [spasms](#) and [spasticity](#) have been reported in 11-15% of FA patients.

Intrathecal baclofen for Friedreich's ataxia

[Intrathecal baclofen for Friedreich's ataxia](#).

Case series

Thomas-Black et al. investigated 62 genetically confirmed FRDA patients using an integrated approach as part of an [observational cohort](#) study. They included measurement of [frataxin](#) protein levels, clinical evaluation of visual and neurological function, [optical coherence tomography](#) to determine retinal nerve fiber layer thickness and macular layer volume, and volumetric brain MRI.

They demonstrated that frataxin level correlates with peripapillary retinal nerve fiber layer thickness and that retinal sectors differ in their degree of degeneration. They also showed that the retinal nerve fiber layer is thinner in FRDA patients than in controls and that this thinning is influenced by the AAO and GAA1. Furthermore, they show that the ganglion cell and inner plexiform layers are affected by FRDA. The MRI data indicate that there are borderline correlations between retinal layers and areas of the cortex involved in visual processing.

The study demonstrates the uneven distribution of axonopathy in the retinal nerve fiber layer and highlights the relative sparing of the papillomacular bundle and temporal sectors. They show that thinning of the retinal nerve fiber layer is associated with frataxin levels, supporting the use of the two biomarkers in future clinical trials design ¹⁾

¹⁾

Thomas-Black G, Altmann DR, Crook H, Solanky N, Carrasco FP, Battiston M, Grussu F, Yiannakas MC, Kanber B, Jolly JK, Brett J, Downes SM, Moran M, Chan PK, Adewunmi E, Gandini Wheeler-Kingshott CAM, Németh AH, Festenstien R, Bremner F, Giunti P. Multimodal Analysis of the Visual Pathways in Friedreich's Ataxia Reveals Novel Biomarkers. Mov Disord. 2022 Nov 25. doi: 10.1002/mds.29277. Epub ahead of print. PMID: 36433650.

From:

<https://neurosurgerywiki.com/wiki/> - **Neurosurgery Wiki**

Permanent link:

https://neurosurgerywiki.com/wiki/doku.php?id=friedreich_s_ataxia

Last update: **2024/06/07 02:51**

