Pattern dystrophy in a cohort of patients with McArdle disease

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Introduction

- A pattern dystrophy has been reported in some patients with McArdle disease (type V glycogen storage disease), mostly as single case reports.[1-7]
- We recently reported four cases.[3]
- We have since examined a cohort of patients referred from a specialist neuromuscular clinic to explore prevalence of the pattern dystrophy.

Methods

- Patients seen in a specialist neuromuscular disease clinic with a molecularly confirmed diagnosis of McArdle disease (owing to bi-allelic variants in PYGM) were referred for retinal examination.
- Retinal imaging included spectral domain optical coherence tomography (OCT), infrared reflectance and short wavelength autofluorescence (AF).

Results

- Twenty-five patients underwent retinal imaging in our clinic.
- Eleven were female (mean (SD) age, 47 (14) years; median 52 years; range 24-65 years) and 14 were male (mean (SD) age, 55 (15) years; median 59.5 years; range 32-72 years).
- A clear pattern dystrophy appearance was evident in 3 patients (all male, ages 64, 67 and 69 years).
- The abnormalities were more evident on infrared reflectance and short wavelength autofluorescence than on clinical examination. OCT confirmed changes at the level of the outer retina and retinal pigment epithelium (RPE). Figure 1 shows an example.
- Figure 2 displays ages of male and female patients grouped by presence or absence of pattern dystrophy.
- Males with pattern dystrophy in our cohort were older on average than those without (p=0.01).

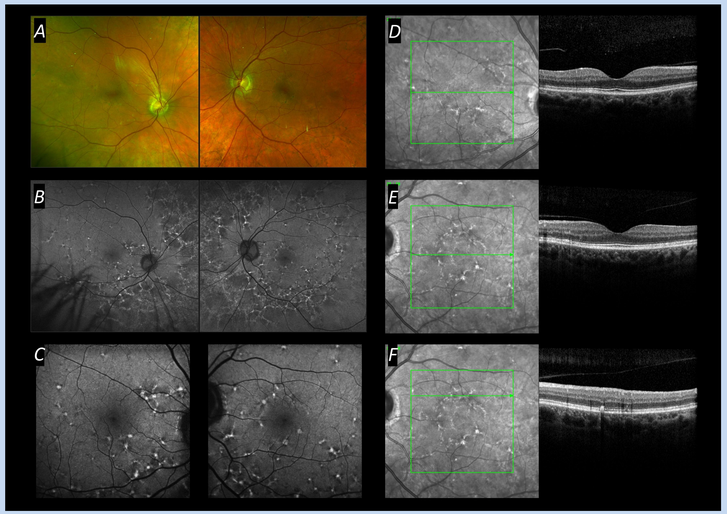


Figure 1 (left). Multimodal retinal imaging findings in a 64 year old male with pattern dystrophy and McArdle disease. *A*, pseudocolour fundus images. *B*, 532 nm AF images. C, 488 nm AF images. *D-F*, Spectral domain OCT images with infrared reflectance en face image. *D*, Horizontal scan through right eye foveal centre. *E*, Horizontal scan through left eye foveal centre. *F*, Horizontal scan superior to foveal centre, showing areas of outer retinal disruption.

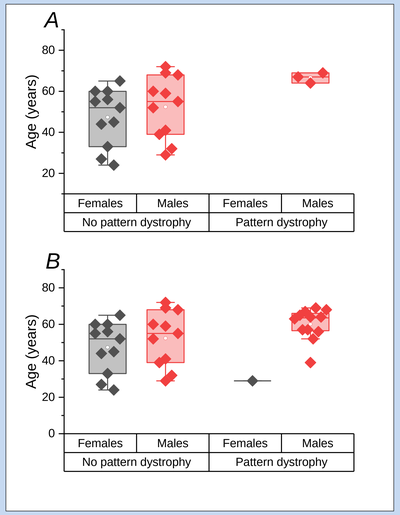


Figure 2 (right). Ages of male and female patients grouped by presence or absence of pattern dystrophy. *A*, Patients in the present study. *B*, Patients in this study as well as previous cases reported in the literature of patients (10 cases) with pattern dystrophy and McArdle disease, including 4 previously reported cases from our group.[3]

Conclusions

- Our study identified three further patients with a pattern dystrophy phenotype (out of 25 examined) showing that this is not a rare finding in McArdle disease.
- Interestingly, a sex imbalance was observed in this cohort, with no females affected by the pattern dystrophy. Of the ten cases currently in the literature,[1-7] only one is female.[7]
- The muscle form of glycogen phosphorylase is expressed in RPE, and these findings might provide insights into aspects of glycogen metabolism in the retina.

References

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