New insights into ocular findings in Marfan syndrome

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Key points:

- Ectopia lentis may develop or progress in adulthood.
- Patients with Marfan syndrome reported increased symptoms of photophobia and glare.
- Understanding pupil size and changes in pupillary function associated with Marfan syndrome is important if surgery is needed.

On September 5, 2022, Gunhild Falleth Sandvik defended her thesis "An ophthalmological study of adults with Marfan syndrome: Ten-year of follow-up and an evaluation of photophobia, glare and pupillary response" at the Faculty of Medicine at University of Oslo (UiO). The PhD project was conducted at the Department of Ophthalmology at Oslo University Hospital (OUH) and the Institute of Clinical Medicine at UiO. The main supervisor was Olav Kristianslund MD PhD, Department of Ophthalmology, OUH, Institute of Clinical Medicine, Faculty of Medicine, UiO, with co-supervisors Professor Liv Drolsum, Department of Ophthalmology OUH, Institute of Clinical Medicine, Faculty of Medicine at UiO and Svend Rand-Hendriksen MD PhD, Sunnaas Rehabilitation Hospital.

Patients with Marfan syndrome often have ocular involvement. The most prevalent ocular manifestation is ectopia lentis — a dislocation of the biological lens— which has been assumed to be present from birth or early childhood in these patients. Other findings commonly seen include myopia, increased axial length, flatter corneal curvature, and hypoplastic iris. These features have been investigated in several studies, but a long-term follow-up has been missing. The diagnosis of Marfan syndrome is based on a list of criteria, and ectopia lentis is one of the cardinal features. If ectopia lentis develops in adulthood in persons previously not fulfilling the criteria, this could possibly lead to a definitive diagnosis. The visual prognosis for patients with Marfan syndrome is often good. However, it is known that Marfan syndrome is associated with a higher prevalence of retinal detachment and cataract. In addition, it has been speculated that patients with Marfan syndrome have increased symptoms of photophobia; however, no findings have previously been published.

This PhD study is the ophthalmological part of the Norwegian Marfan study, where all relevant organs of 44 patients with verified Marfan syndrome were examined at baseline in 2003-2004 and follow-up in 2014-2015. Further, a matched control group for the follow-up examination was included. The thesis aimed to assess changes in ocular features during a ten-year period and to compare photophobia, glare, and pupillary response between patients with Marfan syndrome and a matched control group.

Results from the study revealed that ectopia lentis may develop or progress in adulthood. Other ocular manifestations were stable over the ten-year period; however, an increased risk of cataract and retinal detachment was seen. Independent of these findings, the visual prognosis was good. Evaluation of photophobia and glare showed that patients with Marfan syndrome reported increased symptoms, and measurements of ocular straylight revealed a higher value compared to the matched control group, even after adjustments for cataracts and several other features prevalent in Marfan syndrome. When measuring the pupillary response, we found a smaller pupil size, slower contraction velocity, and longer re-dilation time. The small pupil size was associated with increased straylight in the eye; however, it could not fully explain the difference in straylight between the two groups.

Overall, these results indicate that lacking fulfillment of the diagnostic criteria for Marfan syndrome may change during adulthood, e.g., if ectopia lentis develops. Further, the findings in this PhD project may help when informing newly diagnosed Marfan syndrome patients about what symptoms and changes they may experience in the years to come.

Remaining questions:

- What is the reason for the increased photophobia and glare in patients with Marfan syndrome?
- Could findings of ocular manifestations at an early age predict other disease characteristics in patients with Marfan syndrome?

References

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