Lens dislocation and increased axial length associated with Marfan Syndrome

Svein Ove Semb

EARLY diagnosis and targeted treatment can help to improve the long-term prognosis of patients suffering from ocular complications associated with Marfan Syndrome, according to Svein Ove Semb MD PhD.

"Marfan Syndrome is currently diagnosed using criteria based on an evaluation of family history, molecular data and six organ systems. In 1995, a group of leading clinicians and investigators in Marfan Syndrome proposed revised diagnostic criteria, known as the Ghent criteria," said Dr Semb, University Eye Department, Ullevål Hospital, University of Oslo, Norway.

He noted that the Ghent criteria identify major and minor diagnostic findings, based largely on clinical observation of various organ systems. In terms of ocular complications, more than half of all people with Marfan Syndrome experience dislocation of one or both lenses of the eye. The lens may be slightly dislocated posteriorly (subluxation) or superotemporally (luxation). The dislocation may be minimal or it may be pronounced and obvious.

The Ghent criteria also identified three minor ocular criteria associated with Marfan Syndrome, namely a flat cornea, increased axial length and hypoplastic iris. At least two of these minor criteria must be present for a positive diagnosis of Marfan Syndrome, said Dr Semb. Other possible ocular complications of the disorder include a higher risk of retinal detachment, early glaucoma or cataract.

Dr Semb conducted a prospective population-based study of 105 persons who had been diagnosed as suffering from Marfan Syndrome, who were then investigated for all features of the Ghent criteria.

"We wanted to investigate the prevalence of lens dislocation, flattened cornea, increased axial length and hypoplastic iris in adults with verified Marfan Syndrome based on the Ghent criteria. We estimate that about 200-300 people in Norway have Marfan Syndrome," said Dr Semb.

The control group was made up of 17 out of the 105 patients who had an absolutely negative Marfan Syndrome diagnosis, but who were referred to the clinic for confirmation or rejection of the diagnosis.

Measurements taken by the research team included keratometry (Humphrey), corneal topography (O揉scan II), wavefront aberrometry (Zywave II), axial length and pachymetry (A-Scan, Tomey), and standard slit-lamp examination.

Some 88 of 105 patients fulfilled the Ghent criteria in the study, said Dr Semb. He found an above average axial length, with a mean value of 25mm. Some 66% of the patients had an axial length greater than the 23.5mm specified in the Ghent criteria and 40% were greater than 25mm.

The mean corneal power, or mean K, was 41.7 D. In the Marfan Syndrome group, 86 (50%) of 171 eyes showed a mean K lower than 41.5 D versus only three (8%) of 34 eyes in the control group. Twenty-six eyes (15%) of the group had mean corneal power below 40.0 D, compared to zero in the control group, which corresponds to a considerably flattened cornea.

Mean spherical equivalent refraction was -3.84 (±0.87 D) in right eyes and -3.22 D (±0.90 D) in left eyes. There was a highly significant correlation between right and left eye spherical equivalent refraction, said Dr Semb, although no significant difference was found in mean spherical equivalent refraction between men and women or between the Marfan Syndrome group and the control group.

Dr Semb also noted that there was no evidence of megalocornea found in this study. "In the literature there are some reports claiming that patients with Marfan Syndrome have megalocornea. We therefore measured the corneal horizontal diameter, white-to-white, and found normal values. The central corneal pachymetry was also within the normal range," he said.

The central corneal pachymetry measured with O揉scan was 510 ± 4 (n=165) in the Marfan group and 484 ± 17 (n=32) in the control group, and measured by A-Scan 544 ± 3 (n=173) and 537 ± 13 (n=32) in the Marfan group and control group respectively. Only three patients showed signs of hypoplastic iris. Dislocated lenses were found in 61% (54) of the 88 patients diagnosed with Marfan Syndrome. This also included the patients who had had the lens removed, said Dr Semb. Over one-quarter (26%, n=23) of the patients presented subluxated lenses. "The luxation was bilateral and symmetrical in nearly all patients," said Dr Semb.

He also noted that such subluxation is sometimes undetected by the ophthalmologist because of insufficient dilatation of the pupil.

Dr Semb advised that patients with Marfan Syndrome should have a thorough series of eye examinations to test for dislocation of the lens as well as myopia and other potential complications such as glaucoma and retinal detachment.

Dr Semb presented his findings at the ESCRS Winter Refractive meeting. Dr Semb's co-author of the study was Svend Rand-Hendriksen MD, Sunnaas Rehabilitation Hospital, TRS, a national resource centre for rare disorders in Norway.

s.o.semb@medisin.uio.no