

## References

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J Singh Gandhi

Worcester Royal Ophthalmic Unit, Worcester, UK  
E-mail: doctorjsjg@gmail.com

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Sir,

### Alternative diagnoses with ectopia lentis

We read with great interest the case presented by Moore *et al.*<sup>1</sup> suggesting a correlation between Sturge–Weber syndrome and ectopia lentis. As the authors discuss, the ophthalmic features of Sturge–Weber syndrome do not classically include ectopia lentis, with only two previous reports of such an association.<sup>2,3</sup> The first of these reports is over 35-years old,<sup>2</sup> whereas the second may have been attributable to trauma.<sup>3</sup> Moore *et al* therefore seem to be presenting the first case of ectopia lentis associated with Sturge–Weber syndrome in the era of modern genetic analysis.

The most common cause of spontaneous ectopia lentis is Marfan syndrome, which the authors excluded on clinical grounds in this case, although no family history is discussed. Although normal mentation is mentioned, other syndromes associated with ectopia lentis have not been explicitly discussed by the authors. Nevertheless, if all associated syndromes are excluded, the condition of isolated ectopia lentis (IEL) must be considered. IEL has been established for over 30 years, with both dominant and recessive inheritance reported.<sup>4,5</sup> This can occur with marked asymmetry (Chandra A. ARVO Abstract (2766)(2011)) and indeed be unilateral.<sup>6</sup> Although estimates of IEL prevalence and incidence are yet to be established, up to 31% of congenital ectopia lentis in a national study were not associated with a nosological classification.<sup>7</sup> These may thus be termed IEL, suggesting that the condition of IEL is far more common than an association with Sturge–Weber syndrome, which Moore *et al* are reporting. We have previously shown that mutations in *FBN1* and *ADAMTSL4* have been associated with IEL.<sup>8</sup> Other genetic mutations have also been associated with ectopia lentis.<sup>9</sup>

Although we do not deny that the association suggested in this case by Moore *et al* may be true, it is important to exclude more common causes of ectopia lentis beyond Marfan syndrome, including IEL. Interrogation for known genetic mutations would be

an important and crucial step before such an association can be suggested.

### Conflict of interest

The authors declare no conflict of interest.

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A Chandra<sup>1,2</sup>, JA Aragon Martin<sup>2</sup>, AH Child<sup>2</sup>, G Arno<sup>2</sup> and DG Charteris<sup>1</sup>

<sup>1</sup>Vitreoretinal Department, Moorfields Eye Hospital, London, UK

<sup>2</sup>Sonalee Laboratory, Cardiovascular Sciences, St Georges University of London, London, UK  
E-mail: aman.chandra@moorfields.nhs.uk

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Sir,

### Response to ‘Alternative diagnoses with ectopia lentis’

Chandra and colleagues<sup>1</sup> rightly wonder about other possible causes for ectopia lentis seen in our patient with Sturge–Weber syndrome (SWS).<sup>2</sup> Because of space constraints, we did not report on the full differential diagnosis of ectopia lentis.<sup>3</sup> As we noted, our patient denied even a remote history of trauma; her normal mentation and body habitus, and her ocular examination and the unilaterality of her condition made several possible diagnoses unlikely, including ectopia lentis and