Conflict of interest
The authors declare no conflict of interest.

References
1 Wu W, Dawson DG, Sugar A, Elner SG, Meyer KA, McKey JB *et al*. Cataract surgery in patients with nanophthalmos: results and complications. *J Cataract Refract Surg* 2004; **30**(3): 584–590.
2 Yuzbasioglu E, Artunay O, Agachan A, Bilen H. Phacoemulsification in patients with nanophthalmos. *Can J Ophthalmol* 2009; **44**(5): 534–539.
3 Yalvac IS, Satana B, Ozkan G, Eksioglu U, Duman S. Management of glaucoma in patients with nanophthalmos. *Eye* 2008; **22**(6): 838–843.
4 Faulborn J, Kolli H. Sclerotomy in uveal effusion syndrome. *Retina* 1999; **19**(6): 504–507.
5 Rufer F, Varde MA, Roider J. Intravitreal triamcinolone and bevacizumab injections for alternative treatment of nanophthalmic uveal effusion syndrome. *Klin Monatsbl Augenheilkd* 2008; **225**(6): 594–596.

E Gosse, A Gittos and J Lochhead

Department of Ophthalmology, St Mary's Hospital, Isle of Wight, UK
E-mail: emilygosse@doctors.org.uk

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Sir,
A possible genetic answer to a recently reported novel phenotype

We read with interest the recent article of Shen *et al*,[1] who report a unique Chinese pedigree with the features of ectopia lentis and varicose great saphenous vein. Marfan's syndrome (MFS), clinically diagnosed by characteristic multiple-system abnormalities, lies at one end of a phenotypic spectrum. At the other end of that spectrum are members of the general population who have one feature common to those with MFS.[2] Of those patients who fulfil the modified Ghent criteria for full MFS, up to 97% are found to have FBN1 mutations.[3] However, the patients presented by Shen *et al* are atypical. An alternative approach would be to try to postulate an all-encompassing molecular diagnosis that best fits the clinical signs. Venous varicosity is usually secondary to valvular incompetence, a condition that has been strongly associated with heterozygous mutations in the FOXC2 gene on chromosome 16.[4] Similarly, a range of anterior segment phenotypes have been described, with mutations in FOXC2 inherited in a dominant manner.[5] When aiming for a genetic diagnosis in this family, we would therefore advocate including FOXC2 in the screening set of genes. If a FOXC2 mutation were found in this family, this would represent an interesting extension to the associated phenotype.

Conflict of interest
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References
1 Shen W, Fu Q, Sui R, Wu J, Liu L. Unique phenotype in a Chinese family pedigree: ectopia lentis with varicose great saphenous vein. *Eye* 2010; **24**(10): 1614–1617.
2 Pyeritz RE. The Marfan syndrome. *Annu Rev Med* 2000; **51**: 481–510.
3 Loeys BL, Dietz HC, Braverman AC, Callewaert BL, De Backer J, Devereux RB *et al*. The revised Ghent nosology for the Marfan syndrome. *J Med Genet* 2010; **47**(7): 476–485.
4 Mellor RH, Brice G, Stanton AW, French J, Smith A, Jeffery S *et al*. Mutations in FOXC2 are strongly associated with primary valve failure in veins of the lower limb. *Circulation* 2007; **115**(14): 1912–1920.
5 Smith RS, Zabaleta A, Kume T, Savinova OV, Kidson SH, Martin JE *et al*. Haploinsufficiency of the transcription factors FOXC1 and FOXC2 results in aberrant ocular development. *Hum Mol Genet* 2000; **9**(7): 1021–1032.

K Khan, M Ali and C Inglehearn

Leeds Institute of Molecular Medicine, St James' University Hospital, Leeds, Yorkshire, UK
E-mail: medknk@leeds.ac.uk

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Sir,
Response to Khan *et al*

We thank Khan *et al*[1] for their insightful comments on our recent paper.[2] Their suggestion of including *FOXC2* in the screening set of genes is very interesting. It opens a possibly new aspect of an interesting extension to the associated phenotype in the reported Chinese family. We have already collected 18 genomic DNA samples from three generations of this family. Linkage to *FBN1* locus cannot be ruled out by microsatellite markers. A novel missense mutation was identified in *FBN1* gene, which co-segregated with the ocular phenotype (data not published). Association of single-nucleotide polymorphisms in *TGFβR2* gene was not confirmed. Obviously, this is different from the features associated to disposition to aortic dilatation and dissection of a UK family reported by Law *et al*.[3] *FOXC2*[4,5] or other genes may be the possible genetic factors as Khan *et al* pointed out. Whole-genome scanning using single-nucleotide polymorphism chips is our future strategy, which we hope can answer the question shortly.

Conflict of interest
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References
1 Khan K, Ali M, Inglehearn C. A possible genetic answer to a recently reported novel phenotype. *Eye* 2011; **25**: 529.
2 Shen W, Fu Q, Sui R, Wu J, Liu L. Unique phenotype in a Chinese family pedigree: ectopia lentis with varicose great saphenous vein. *Eye* 2010; **24**(10): 1614–1617.
3 Law C, Bunyan D, Castle B, Day L, Simpson I, Westwood G *et al*. Clinical features in a family with an R460H mutation in transforming growth factor b receptor2 gene. *J Med Gent* 2006; **43**: 908–916.