the entire reading frame of all 8 genes in this locus. This is unlikely to be explained on the basis of deep intronic mutations or regulatory element mutations given the normal results of reverse transcription–PCR. Normal reverse transcription–PCR results also rule out the possibility of gene rearrangement as a potential cause. Therefore, the causative mutation must reside in an as yet unannotated gene or intergenic regulatory element within the minimal linkage interval. Capture of this interval for subsequent next-generation sequencing is an attractive strategy that we are actively pursuing.

Mutations in MFRP were first reported in patients with severe hyperopia. While severe hyperopia is a major feature of PM, the anterior segment involvement in patients with MFRP mutations indicates that the phenotype is best described as nanophthalmos. In addition, another MFRP mutation was described in patients who, in addition to PM, have retinitis pigmentosa, loveoschisis, and optic disc drusen. Therefore, our finding of a novel mutation in 2 patients with the classic nonsyndromic PM phenotype represents the first evidence to date that MFRP is a bona fide nonsyndromic PM gene. While it is premature to draw any meaningful genotype-phenotype correlation for MFRP-related PM, it is noteworthy that our missense mutation is milder than the truncating mutations that characterize the mutational spectrum of nanophthalmos and syndromic PM.

Two other findings are worth highlighting. First, exclusion of linkage to MFRP and chromosome 2q37.1 in family 6 suggests the presence of yet another locus for this genetically heterogeneous condition. Second, the fact that none of the ethnically Saudi families have linkage to MFRP suggests a low frequency for mutations in this gene in this population. Both findings should encourage further research in the genetics of PM in Saudi Arabia, which is likely to identify important effectors of embryonic eye development.

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Subclinical Facioscapulohumeral Muscular Dystrophy Masquerading as Bilateral Coats Disease in a Woman

Coats disease is a nonhereditary retinal vasculopathy that typically occurs unilaterally in young males. We describe a unique case of a woman with bilateral Coats disease–like retinal changes and subclinical facioscapulohumeral disease (FSHD).

Report of a Case. A 39-year-old woman with an unremarkable medical and ocular history had a routine eye examination. She denied any family history of hereditary systemic or ocular disease. Visual acuity was 20/20 OU. The slitlamp examination results and intraocular pressures were normal. Funduscopic examination of the right eye revealed a normal posterior pole with subtle sheathing of retinal vessels without exudation in the far inferotemporal periphery (Figure, A). Funduscopic examination of the left eye also revealed a normal posterior pole with an area of fibrovascular tissue surrounded by lipid exudation in the far temporal periphery (Figure, B). The patient underwent wide-field fluorescein angiography (Optos P200, Optos PLC, Dunferline, Scotland) to obtain superior images from the far periphery (Figure, C and D). The wide-field fluorescein angiogram showed bilateral temporal retinal telangiectasia, aneurysms, and peripheral nonperfusion with leakage and staining of the fibrovascular lesion in the left eye (Figure, C and D). Results of a comprehensive medical workup for infectious and inflammatory causes were negative. Owing to reports of a relationship between bilateral Coats disease–like retinal vascular changes and FSHD, the patient underwent genetic testing for this muscular dystrophy. Genetic testing demonstrated a deletion in chromosome 4q35 and resulted in 27-kilobase (kb) and 24-kb band fragments following diges-
tion with EcoRI and double digestion with EcoRI/BlnI, respectively. These findings provided molecular confirmation of FSHD in our patient. However, neurological examination and electromyography did not show any abnormalities. The patient denied having any hearing deficits. A formal audiology examination was not performed.

Comment. Coats disease is a nonhereditary retinal vasculopathy associated with telangiectasia, aneurysms, capillary nonperfusion, and intraretinal and subretinal exudation. Most cases occur unilaterally in young males. However, there have been reports of Coats disease in adults, in females, and bilaterally. Patients with bilateral Coats disease–like changes may have an underlying systemic or hereditary disease process, most notably FSHD. Facioscapulohumeral disease is an autosomal dominant myopathy (with a high percentage of sporadic cases) characterized by progressive weakness in the facial, shoulder, and upper arm muscles that can be associated with retinal vascular changes in 50% to 75% of patients.

Although bilateral Coats disease associated with severe FSHD has been reported in 3 girls, this has not previously been reported in the literature in an asymptomatic adult patient to our knowledge. Other diagnoses that can manifest similarly were also considered in this patient prior to genetic testing. They include vasoproliferative tumor, familial exudative vitreoretinopathy, dyskeratosis congenita, and Parry-Romberg syndrome. However, there were no feeder vessels (as are sometimes seen in vasoproliferative tumor), no family history of retinal disease (seen with familial exudative vitreoretinopathy), no bone marrow, skin, or nail abnormalities (found in dyskeratosis congenita), and no hemifacial atrophy (as in Parry-Romberg syndrome). Despite the unique manifestation, the clinical picture in conjunction with genetic testing suggested a Coats-like disease in association with FSHD. Genetic testing was critical as the patient did not initially show any symptoms of muscular dystrophy and did not manifest any electromyographic abnormalities.

Of interest is the subclinical nature of FSHD. There is a spectrum of severity in FSHD that appears to be under the influence of several factors. Females are less affected, as are earlier generations of affected family members. The infantile-onset form of FSHD is more severe and is more likely to exhibit features such as sensorineural hearing loss, which is generally absent in patients with the late-onset form of the disorder. The fragment size inversely correlates with the size of the deletion (the smaller the fragment, the larger the deletion). A larger deletion results in an earlier and more severe disease onset. In our patient, the lack of previously affected family members, relatively higher restriction digest fragment lengths (more severely affected patients usually have lengths of 10-18 kb), and female sex may explain the patient’s subclinical manifestation. Also unusual is that in FSHD the myopathy generally manifests initially and the retinal changes, if present, are discovered only later during a screening ophthalmic examination. This is one of only 2 cases to
our knowledge in which retinal findings were detected prior to the diagnosis of FSHD.3

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Three-Dimensional Reconstruction and Analysis of Vitreomacular Traction: Quantification of Cyst Volume and Vitreoretinal Interface Area

Optical coherence tomography (OCT) has made considerable advancements in retinal imaging, especially with the advent of high-resolution, spectral-domain OCT.1 Nonetheless, viewing and analysis of OCT data are limited to 2-dimensional (2D) slice-based scrolling through consecutive scans (Figure 1).2

Video available online at www.archophthalmol.com

In a series of eyes with idiopathic vitreomacular traction, we used a method of rendering 2D raster OCT data into 3-dimensional (3D) volumetric objects. By isolating and quantifying distinct retinal structures within these 3D objects, we sought to determine the following: (1) the correlation between cyst volume and area of vitreoretinal adhesion; and (2) the relationship between individual cysts within the retina.

Methods. In this Weill Cornell Medical College Institutional Review Board–approved study, OCT scans (Heidelberg Spectralis HRA + OCT; Heidelberg Engineering, Inc, Carlsbad, California) of idiopathic vitreomacular trac-