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# Consanguineous marriage resulting in homozygous occurrence of X-linked retinoschisis in girls

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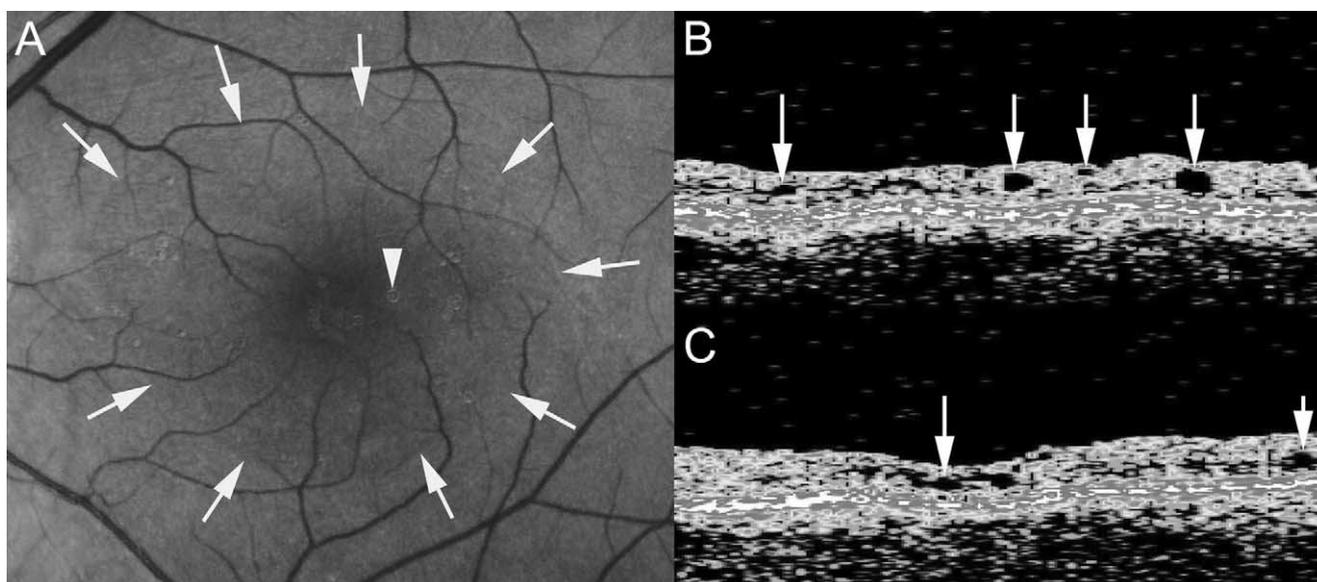
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**FIGURE 1.** (A) A red-free fundus photograph reveals several vacuoles within the area of internal limiting membrane peeling (demarcated by white arrows). The arrowhead in the middle shows a vacuole. Horizontal scans using optical coherence tomography 8 months after surgery slightly superior to the fovea (B) and through the fovea (C) show intraretinal vacuoles, largely confined to the middle layers of the retina.

silicone oil with eventual deposition of silicone oil within the retina, observable only within the region of ILM peeling. It is possible that the intentional iatrogenic defect created by ILM removal augmented the ability of the emulsified oil to penetrate into the retinal tissue. The presence of the silicone droplets could be detected by ophthalmoscopy and optical coherence tomography.

The anatomic success of macular hole surgery with ILM peeling using intraocular gas as a tamponade is greater than 90%.<sup>4</sup> The success rate for limited head positioning is also very high. Although silicone oil tamponade has been used in patients with macular hole surgery to avoid the need for strict positioning, the number of associated possible complications, along with the need for additional surgery to remove the oil, raises questions as to the true need for and utility of silicone oil tamponade in macular hole surgery other than for patients who will require air travel in the immediate postoperative period.

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## Consanguineous Marriage Resulting in Homozygous Occurrence of X-linked Retinoschisis in Girls

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**PURPOSE:** To investigate a family afflicted with X-linked retinoschisis and to determine the pedigree leading to such an occurrence.

**DESIGN:** Observational case series.

**METHODS:** Visual acuities were measured at The Aga Khan University Hospital Ophthalmology clinic. Slit-lamp examination, direct and indirect ophthalmoscopies were performed on each of the seven members of this family. The pedigree was established by appropriate questioning about the rest of their family.

**RESULTS:** The pedigree revealed the homozygosity of the four daughters for the XLRs mutations borne out of the

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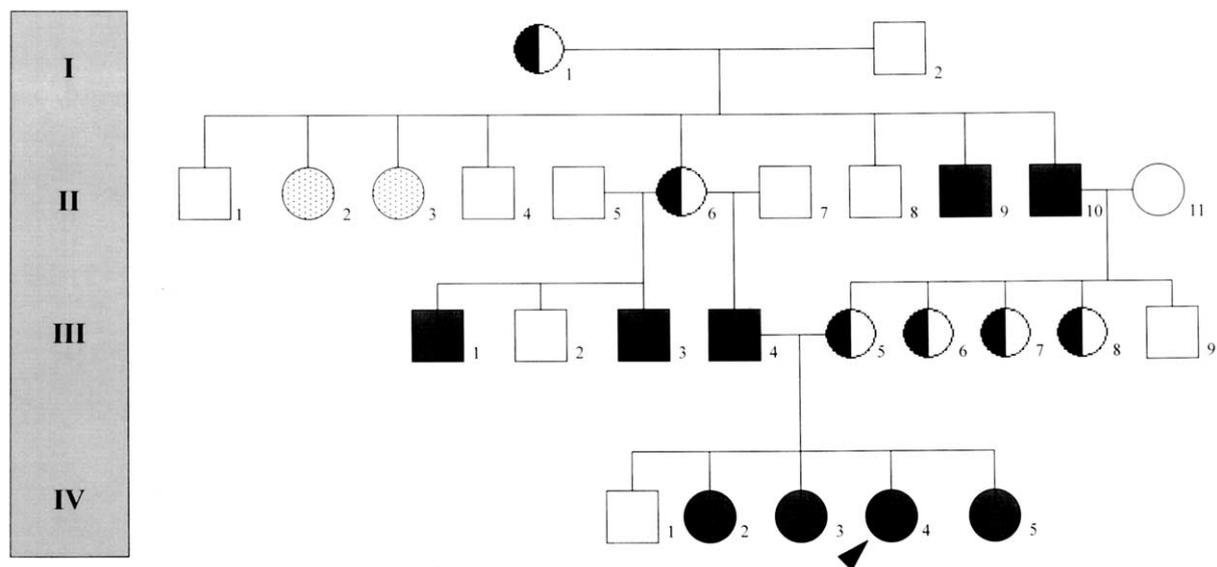


FIGURE 1. Family tree of a family afflicted with X-linked retinoschisis. Open circle = normal woman; black circle = affected woman; open square = normal man; black square = affected man; white/black circle = carrier woman; speckled circle = 50% chance of being a carrier or being normal.

consanguineous marriage between an affected father and a carrier mother.

**CONCLUSIONS:** This is the first report showing that consanguineous marriages lead to homozygous pairing of XLR5 genes in female children, as concluded after a MEDLINE search. (*Am J Ophthalmol* 2003;136:767-769. © 2003 by Elsevier Inc. All rights reserved.)

**A** GIRL WAS CLINICALLY DIAGNOSED WITH X-LINKED juvenile retinoschisis (XLR5). Owing to the unusual severity of this disease in a female, the whole family was checked for the manifestations of this X-linked recessive disorder. Their family tree for four generations is shown in Figure 1 and reveals the homozygosity of the four daughters for XLR5 mutations.

A girl (IV, 4 in Figure 1) presented to us with bilateral nystagmus and light perception in the right eye only. In the right eye, there was total rigid retinal detachment. The left eye had gross searching horizontal nystagmus with leucoria. There was no fundus view on the left side, and the B-scan showed total retinal detachment in the left eye.

The father (III, 4 in Figure 1) was diagnosed with XLR5 as a child. His best-corrected acuities were 6/18 in the right eye and counting fingers at 2 m in the left eye. He had a left divergent squint and relative afferent pupillary defect in the amblyopic left eye. There was extensive macular scarring and signs of peripheral retinoschisis in both eyes. His wife (III, 5 in Figure 1) and a 13-year-old son (IV, 1 in Figure 1) were clinically normal with unaided 6/6 vision in both eyes, clear media, and no retinal abnormalities.

Their 10-year-old daughter (IV, 2 in Figure 1) had best-corrected visual acuities of 6/60 in the right eye and 3/60

in the left eye. There were punctate lens opacities in the right eye, but the left lens was clear. There was obvious macular schisis of the typical bicycle wheel configuration, with long-standing inferior retinoschisis extending to the inferior arcades in both eyes.

Their 5-year-old daughter (IV, 3 in Figure 1) had left esotropia. Her best-corrected visual acuities were 6/36 in the right eye and 6/60 in the left eye. There was a zonular cataract in the left eye, and she had bilateral inferior scars with pigmentary changes accompanying peripheral and macular retinoschisis. Her optic disks were healthy, and there was no evidence of retinal detachment.

The youngest daughter (IV, 5 in Figure 1) was 1 year old. She had left esotropia first noticed when she was 3 months old. Being too young for an accurate assessment of vision, she seemed to have better vision in her right eye compared with the left eye. She had bilateral retinal detachment and retinoschisis with macular involvement. There were secondary changes in the areas of detachment with markedly atrophic retinae.

X-linked juvenile retinoschisis is an incurable hereditary vitreoretinal degeneration and the most common cause of juvenile macular degeneration in males.<sup>1</sup> The mode of transmission of XJR is X-linked recessive, primarily affecting males. Female patients with XJR from consanguineous marriages have been reported for the first time in this case series. A father and his daughter from a consanguineous marriage have been reported by Yamaguchi and Hara<sup>2</sup> to have autosomal recessive juvenile retinoschisis. After a MEDLINE search of the literature, we conclude that this is the first report of homozygous XLR5 occurrence in females from a consanguineous marriage.

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## Nonmechanical Posterior Lamellar Keratoplasty Using the Femtosecond Laser (femto-PLAK) for Corneal Endothelial Decompensation

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**PURPOSE:** To assess the potential of a short pulsed laser to cut a posterior graft and bed for posterior lamellar keratoplasty (PLAK).

**DESIGN:** Experimental study.

**METHODS:** Using the laser FEMTEC (20/10 Perfect Vision, Heidelberg, Germany), posterior lamellar dissections (wave length approximately 1  $\mu\text{m}$ , pulse energy < 10  $\mu\text{J}$ , spot size < 10  $\mu\text{m}$ , repetition rate 12.5 kHz, 6-mm–7 mm diameter, 31 s and 90 s) were performed in 18 freshly enucleated porcine eyes and 10 human donor corneas starting from the anterior chamber and ending with the lamellar bed.

**RESULTS:** Before removal, 50- $\mu\text{m}$  to 500- $\mu\text{m}$ -thick flaps were delineated by partly confluent gas bubbles (maximum 2-mm long) with minute tissue bridges (typically 5- to 10- $\mu\text{m}$ ) in between. Scanning electron microscopy displayed smooth cut surfaces and rectangular corners with minor remaining tissue bridges (approximately 5  $\mu\text{m}$ ). By transmission electron microscopy, the cut edges were lined by a delicate, electron-dense layer (5 nm–10 nm in width) and essentially normal adjacent collagen fibers.

**CONCLUSIONS:** Femtosecond laser technology seems to offer a promising approach to minimally invasive posterior lamellar keratoplasty (femto-PLAK) through small tunnel incisions in corneal endothelial diseases. (*Am J Ophthalmol* 2003;136:769–772. © 2003 by Elsevier Inc. All rights reserved.)

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**A**FTER UNCOMPLICATED NORMAL-RISK PENETRATING keratoplasty, high or irregular corneal astigmatism and ametropia are among the major complications impairing the optical quality of the graft. These problems may be solved in part by nonmechanical corneal trephination in donor and recipient using the 193-nm excimer laser along metal masks with eight “orientation teeth.”<sup>1</sup>

Among the most frequent indications (30%–40%) for penetrating keratoplasty in developed countries are pseudophakic or aphakic bullous keratopathy and Fuchs dystrophy. For these corneal endothelial diseases, Melles and associates<sup>2</sup> in Europe and Terry and Ousley<sup>3</sup> in the United States introduced the technique of posterior lamellar keratoplasty, or deep lamellar endothelial keratoplasty, to selectively exchange endothelium, Descemet membrane, and as little adjacent stroma as possible. In contrast to the “open sky” approach with full-thickness keratoplasty techniques, the recipient eye is opened with a tunnel type of incision, thus reducing the amount of tissue transplanted, reducing the risk of infection, expulsive hemorrhage, and suture-related complications (graft rejection due to suture loosening, vascularization, suture infection) thus requiring fewer frequent postoperative follow-up visits. However, there are several challenges that remain in this procedure. The manual dissection of a deep lamellar plane through a 9.0-mm scleral limbal incision remains difficult, with a significant risk of inadvertent perforation. The greatest technical challenge in this procedure is the intralamellar trephination and scissors excision of the recipient posterior disk supported by only an air bubble in the anterior chamber.<sup>3</sup> In an attempt toward standardization of these difficult steps, the purpose of this study was to assess the potential of a short pulsed laser to cut a posterior graft and bed in a controlled, automated manner.

In this laboratory study 18 freshly enucleated porcine eyes and 10 human donor corneas not suitable for corneal transplantation were treated. Using the laser FEMTEC (20/10 Perfect Vision, Heidelberg, Germany) we performed posterior lamellar dissections (wave length approximately 1  $\mu\text{m}$ , pulse energy < 10  $\mu\text{J}$ , spot size < 10  $\mu\text{m}$ , repetition rate 12.5 kHz) starting from the anterior chamber and ending with the lamellar bed. Pulse energy and spacing of the spots were varied. After the laser action, complete flaps (6 mm–7 mm diameter) were either removed manually (nine pig eyes, five human corneas) or left in situ and were fixed in 10% buffered formaldehyde solution and processed for periodic acid–Schiff and hematoxylin and eosin stain light microscopy, transmission electron microscopy (in two pig eyes and one human cornea after manual flap removal), and scanning electron microscopy (in two pig eyes and one human cornea with the flap left in situ). Main outcome measures included feasibility of the flap creation, configuration of the flap/bed, regularity of the cut surfaces, and thermal damage adjacent to the cut edge.