Asociación de Síndrome de Axenfeld-Rieger y Síndrome de Peter’s en un mismo paciente

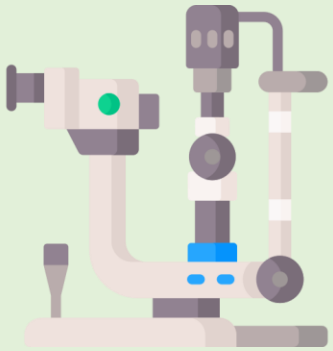
Marta Caminal, Alicia Serra, Nuria Planas

CASO CLÍNICO

Recién nacida **3 días de vida**
IC por **opacidad corneal AO**

- Rasgos craneofaciales dismórficos (macrocefalia, ventriculomegalia)
- Foramen oval permeable
- Ectasia renal
- Extremidades cortas

CASO CLÍNICO



OD

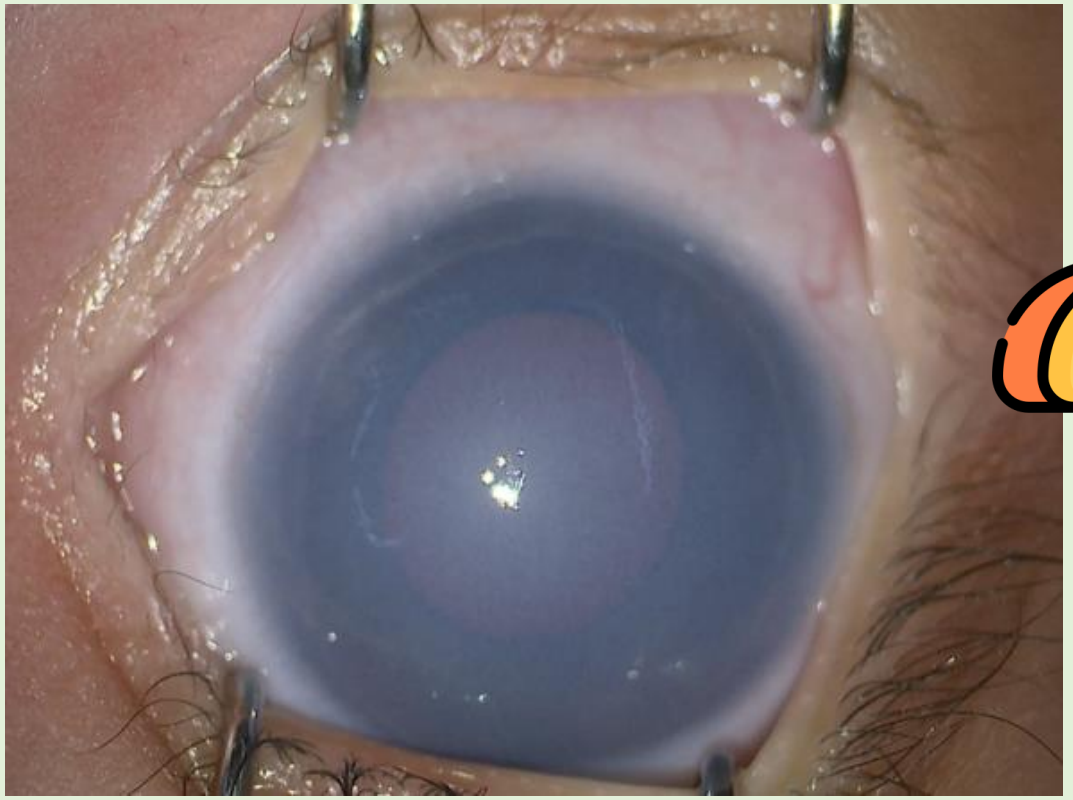
- Opacidad difusa corneal
- Megalocórnea
- PIO 34

OI

- Opacidad corneal densa vascularizada
- Diámetro corneal normal
- PIO normal

OD Dorzolamida + timolol cada 12h + Acetazolamida oral

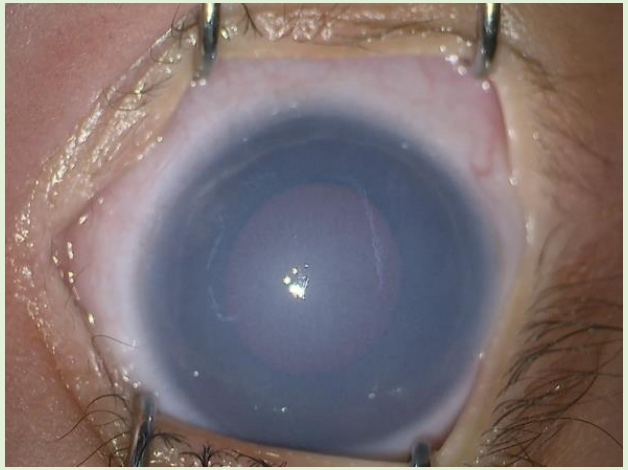
EXPLORACIÓN QUIRÚRGICA



Sd.Axenfeld-Rieger

Sd. Peter’s

EVOLUCIÓN



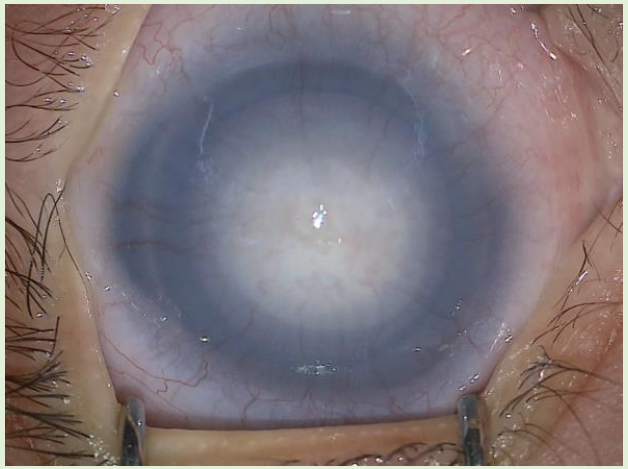
PIO 34

Dorzolamida + timolol cada 12h +

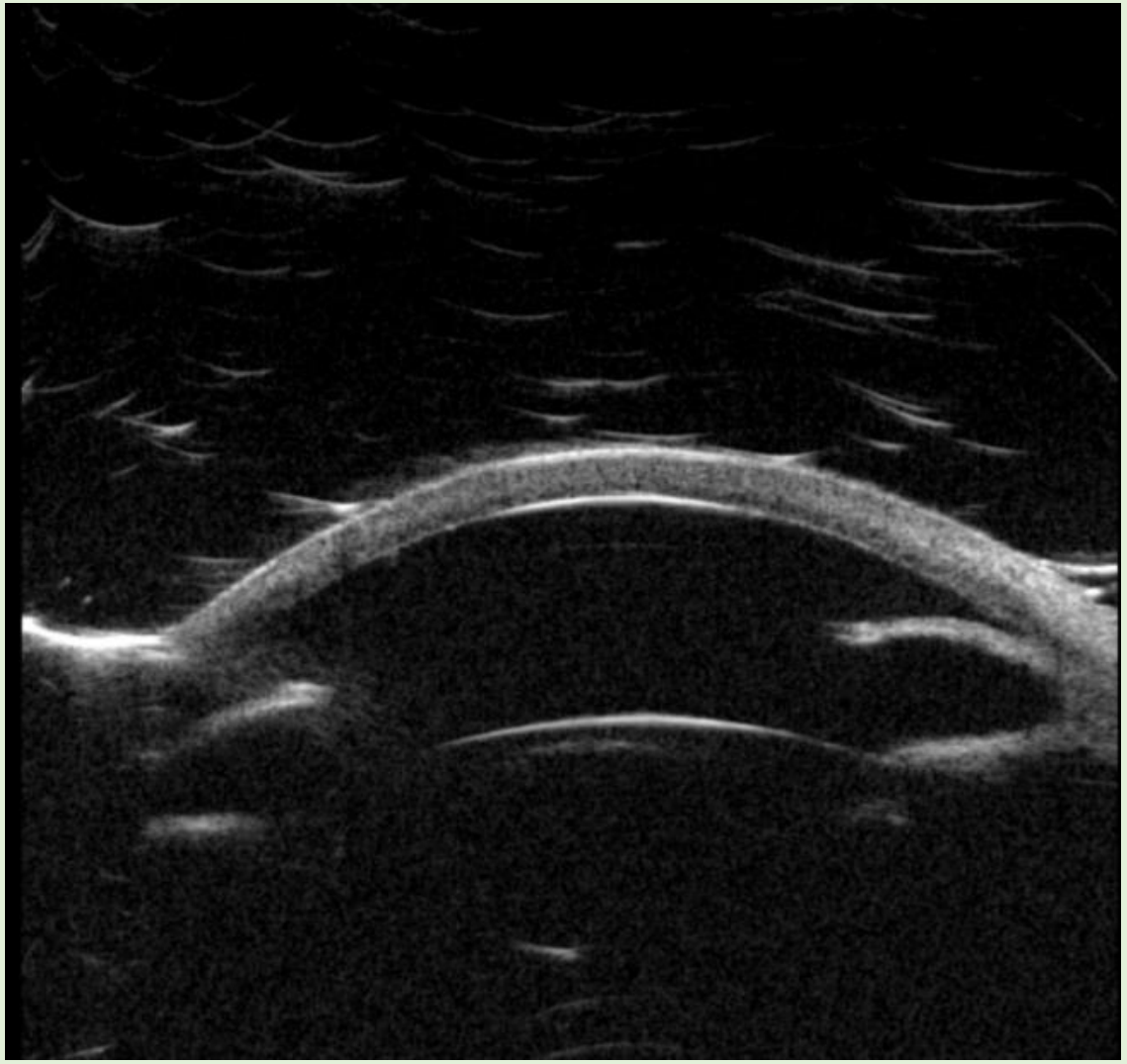
Acetazolamida oral

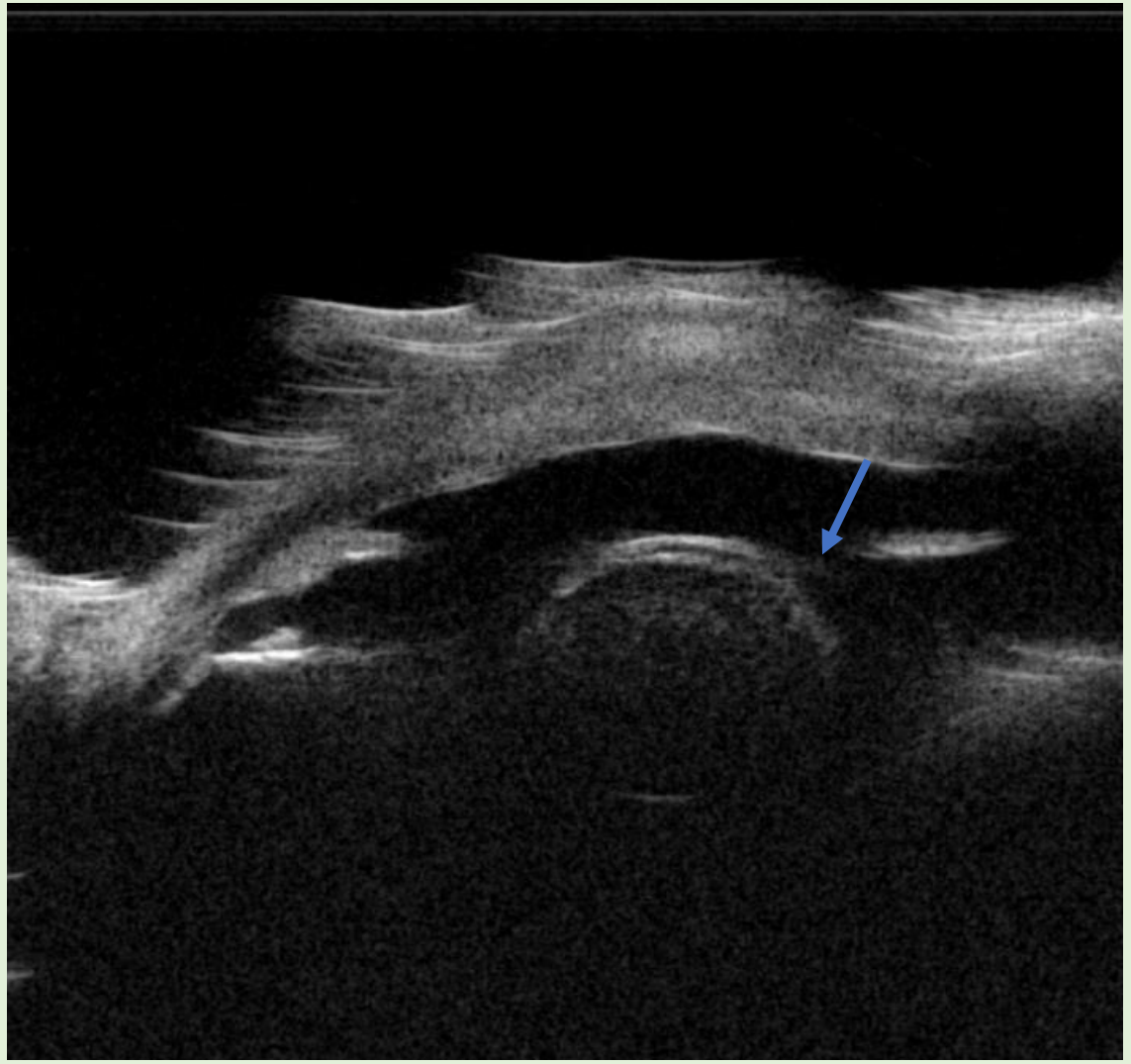
1 Trabeculotomía + 2 Goniotomía

EVOLUCIÓN



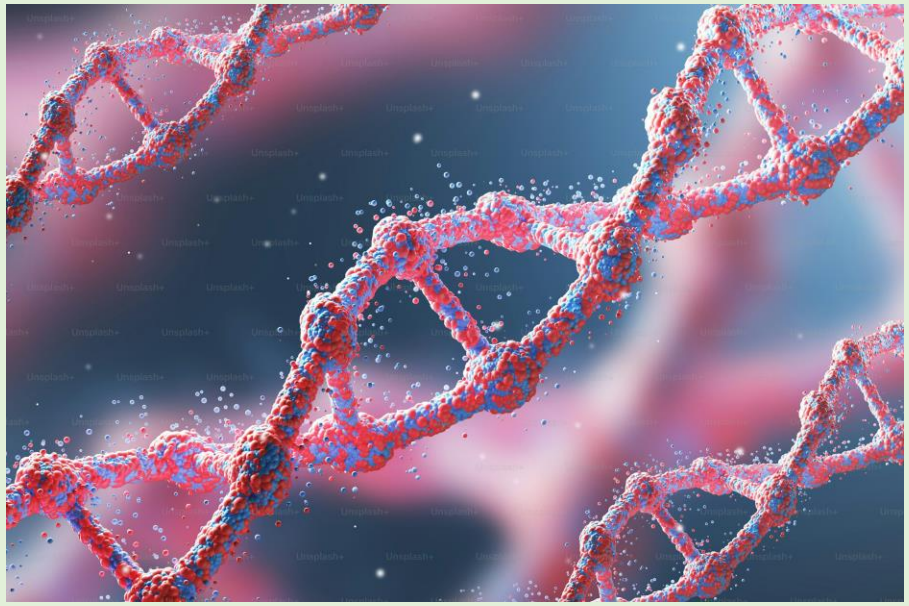
Queratoplastia penetrante



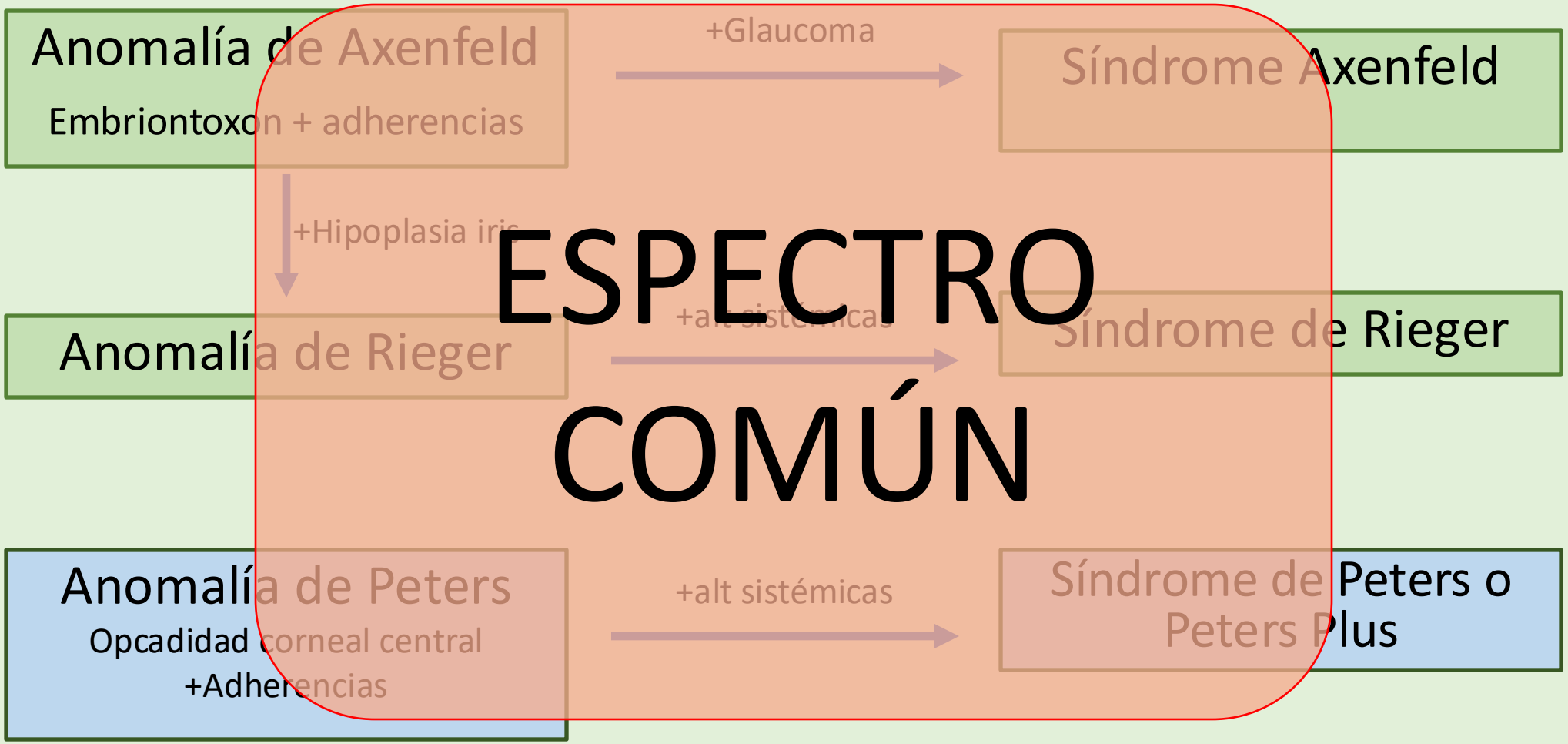


GENÉTICA

- No antecedentes oftalmológicos en la família.
- TEST GENÉTICO--> **mutación gen FOXC1,** en heterocigosis
- Ser131Phefs*51



OVERLAP ANOMALÍA AXENFELD-RIEGER Y PETERS



OVERLAP ANOMALÍA AXENFELD-RIEGER Y PETERS

- **Disgenesias de segmento anterior**
- **Crestopatías**
- Amplio fenotipo/**gravedad**

HETEROGENEIDAD GENETICA

Diferentes mutaciones en varios genes producen un mismo fenotipo

EXPRESIVIDAD VARIABLE

Una misma mutación de un gen produce diferentes fenotipos

2008

A novel mutation in the *FOXC1* gene in a family with Axenfeld–Rieger syndrome and Peters' anomaly

Weisschuh N, Wolf C, Wissinger B, Gramer E. A novel mutation in the *FOXC1* gene in a family with Axenfeld–Rieger syndrome and Peters' anomaly.
Clin Genet 2008: 74: 476–480. © Blackwell Munksgaard, 2008

N Weisschuh[a], C Wolf[a], B Wissinger[a] and E Gramer[b]

[a]Molecular Genetics Laboratory, Institute for Ophthalmic Research,

- Familia 5 miembros--> **mutación nonsense (Q120X) en el gen FOXC1 → 4 miembros S.Axenfeld-Rieger + 1 con anomalia de Peter's bilateral.**

1977

Case Reports > J Pediatr Ophthalmol. 1977 Mar-Apr;14(2):112-6.

Peters-Rieger's syndrome

K J Awan

PMID: 406376

El **primero en describir** la rara asociación entre anomalía de Peters y Riegers en un mismo paciente

1991

Heterogeneity in dominant anterior segment malformations

Gerd E Holmström, William P Reardon, Michael Baraitser, John S Elston, David S Taylor

Describen 2 familias con afectos Riegers + 1 miembro con Riegers y Peters

2003

A Family With Axenfeld–Rieger Syndrome and Peters Anomaly Caused by a Point Mutation (Phe112Ser) in the *FOXC1* Gene

ROBERT A. HONKANEN, MD, DARRYL Y. NISHIMURA, PHD, RUTH E. SWIDERSKI, PHD, STEVEN R. BENNETT, MD, SUNGPYO HONG, MD, YOUNG H. KWON, MD, PHD, EDWIN M. STONE, MD, PHD, VAL C. SHEFFIELD, MD, PHD, AND WALLACE L.M. ALWARD, MD

TABLE 2. Clinical Features of Affected Family Members*

	Cornea		Angle	Iris			Systemic	
Pedigree Number	Embryotoxon	Peters	Iris Processes	Hypoplasia	Corectopia	Glaucoma	Facial / Dental	Cardiac Anomalies
II:3	+	–	+	+	+	post-op	–	Aortic valve replacement
III:1	+	–	+	+	+	+	–	Congestive heart failure
III:3	+	–	+	–	–	–	+	none
IV:1	+	+	+	–	–	–	–	none
IV:2	+	–	+	–	–	–	+	none

*There are no columns for polycoria, umbilical abnormalities, or hypospadias because no family members demonstrated those signs.

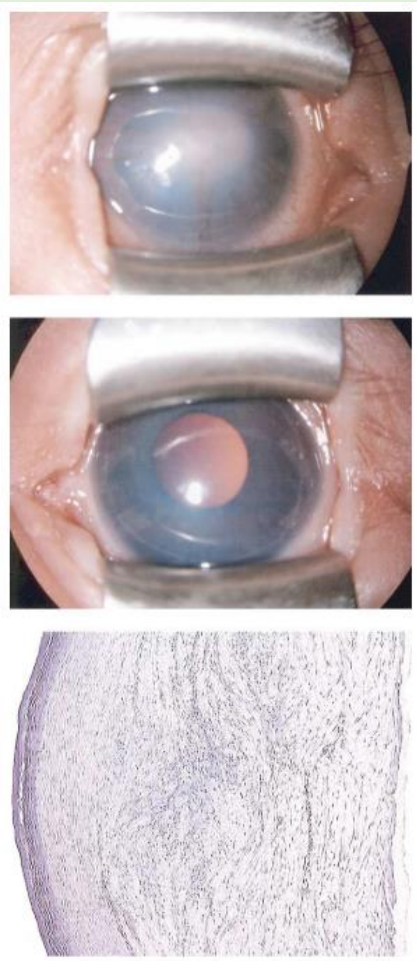


FIGURE 3. Patient IV-1 as an infant. (Top) Operating room photograph illustrating the corneal opacity in the right eye. (Middle) The left cornea was clear. (Bottom) Histopathology of the cornea of the right eye indicating the lack of endothelium and Descemet membrane, consistent with Peters anomaly.

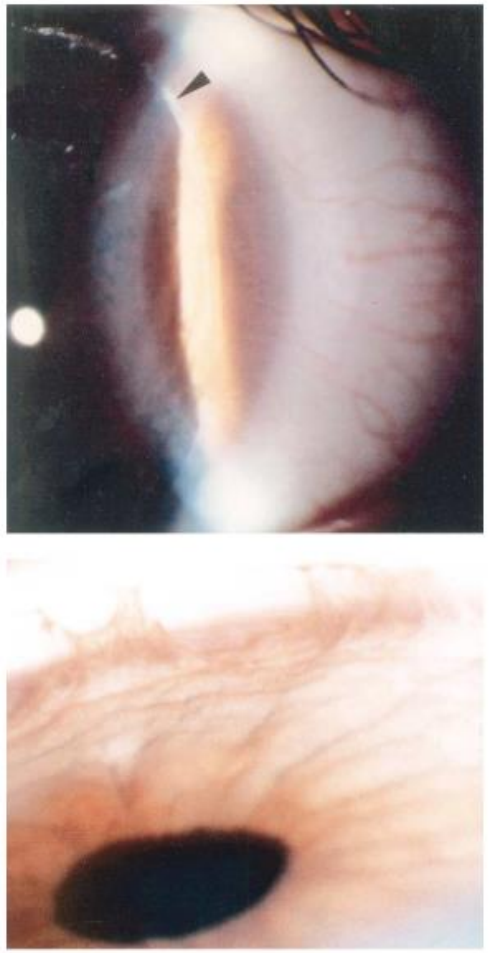


FIGURE 4. Patient IV-1 at age 13. (Top) Slit-lamp photograph of the posterior embryotoxon (arrow). (Bottom) Gonioscopic photograph of the iridocorneal angle demonstrating broad adhesions of the iris to the cornea.

Familia 10 miembros--> 5 disgnesia SA + **mutación F112S en el gen FOXC1 --> Diferentes fenotipos**
IV:1 Paciente con anomalía Rieger y Peters

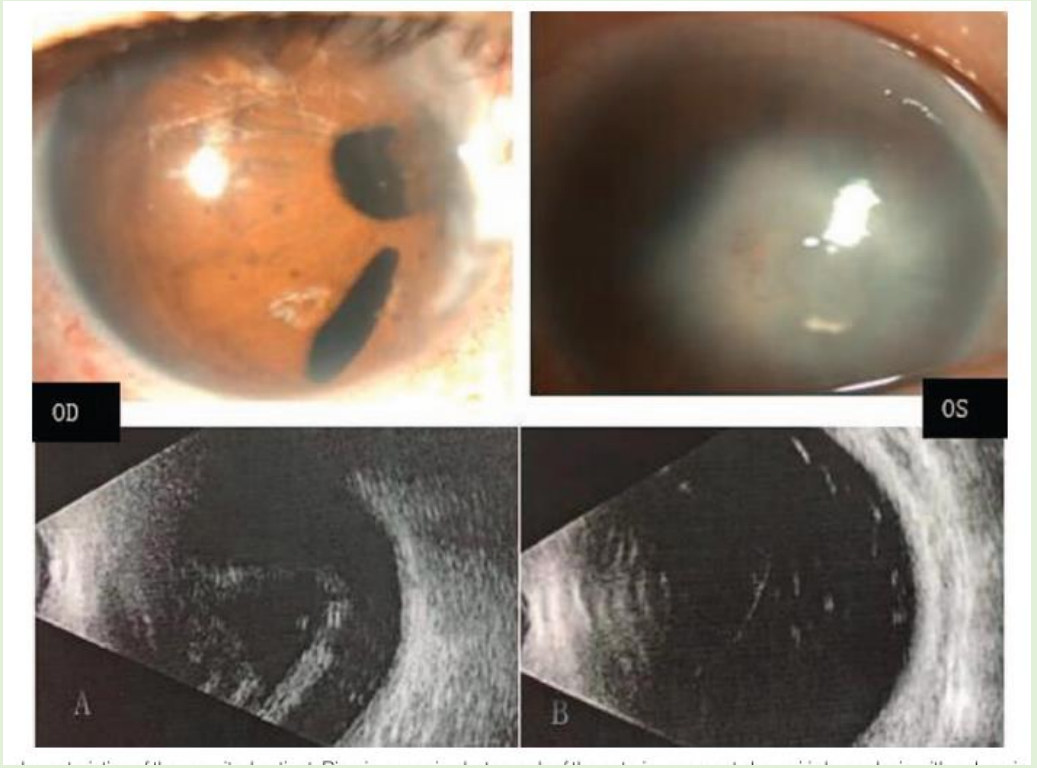
2022

Case report of the rare Peters' anomaly complicated with Axenfeld-Rieger syndrome

A case report and brief review of the literature

Yong Meng, MD[a], Guohua Lu, MD[b], Yang Xie, MD[b], Xincheng Sun, MD[b], Liqin Huang, MD[b,*]

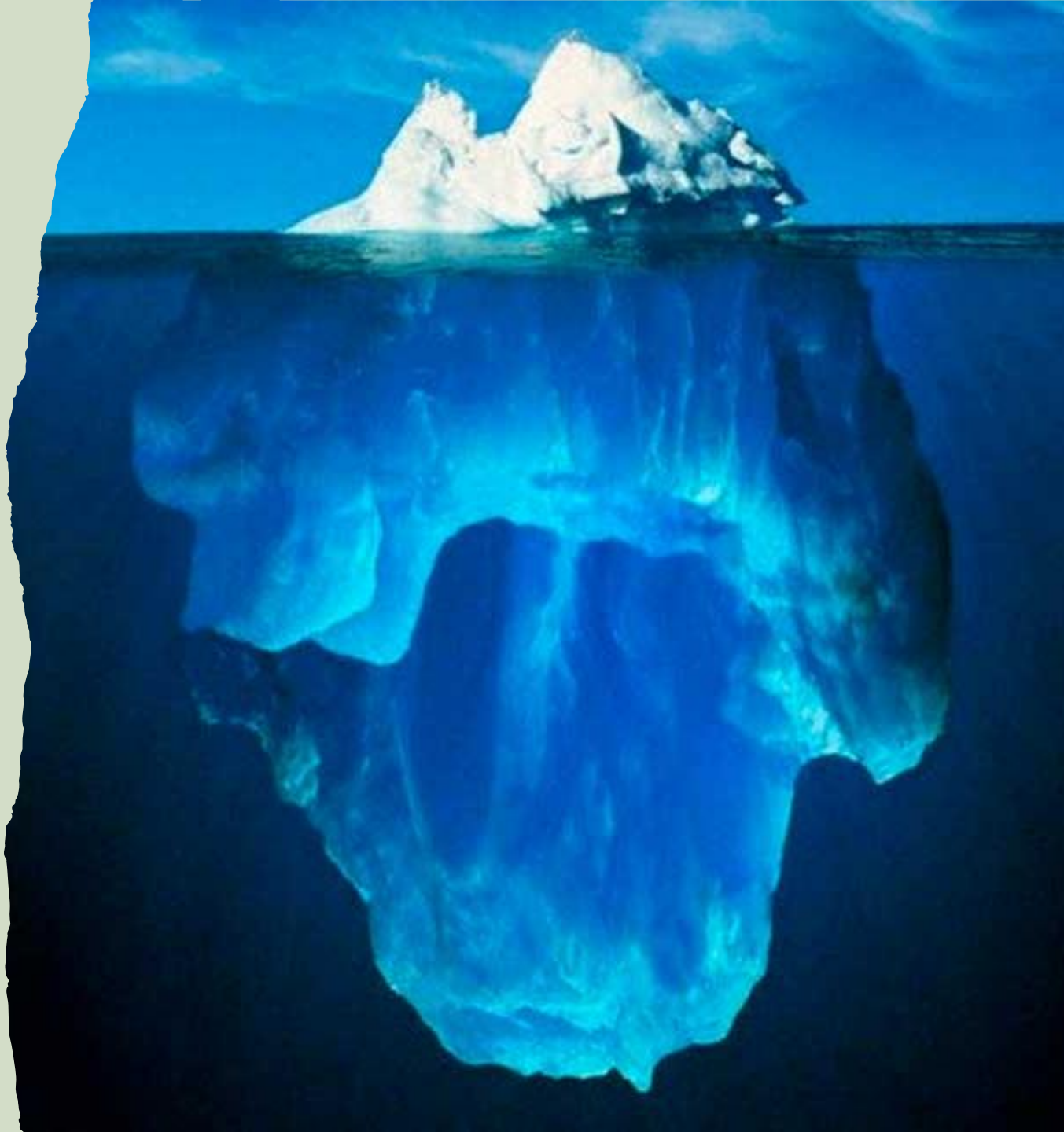
Paciente anomalía Riegers y Peters --> asoc catarata, DR, exotropia
Test genético -



Artículo	KJ Awan 1977	Gerd E Holmström 1991	Robert A.Honkanen 2003	Weisschuh 2008	Yong Meng 2022	Nuestro caso
Genética	-	-	Mutación **F112S en el gen FOXC1**	Mutación **nonsense (Q120X) en el gen FOXC1**	Test genético **negativo**	**Mutación de significado incierto** (Ser131Phefs*51) en el **gen FOXC1**
Clínica	Paciente con Rieger + Peters	2 familias con diferentes afectos de rieger y 1 miembro Rieger + Peters	Paciente con Rieger + Peters	Familia 4 Rieger + 1 Peters bilateral	Paciente con Rieger + Peters	Paciente con Rieger + Peters
Asoc oft	Cata polar, papila oblicua, hipoplasia N.O, ectopia mácula				Exotropia, nistagmus, catarata, DR, displasia foveal hipoplasia N.O	

CONCLUSIONES

- ✓ Overlap **Genotipo-fenotipo** disgenesias SA
- ✓ Mismo **espectro** de enfermedades
- ✓ Heterogeneidad genética y **expresividad variable**
- ✓ Mutación única de gen concreto (**FOXC1**): amplio espectro fenotipos



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