Corneal Dystrophies and Simulating Lesions

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Case-Based Questions (please see pager 3 for answers)

1.	What is the Category 1 Corneal Dystrophy according to the IC3D Clasification?	
	a.	A well-defined corneal dystrophy in which the gene has been mapped and identified and the specific mutations are known
	b.	A well-defined corneal dystrophy that has been mapped to one or more specific chromosomal loci, but the gene(s) remains to be identified
	c.	A well-defined corneal dystrophy in which the disorder has not yet been mapped to a chromosomal locus
	d.	This category is reserved for a suspected, new, or previously documented corneal dystrophy, although the evidence for it, being a distinct entity is not yet convincing

2.	What gene is responsible for pathogenesis of lattice, granular, Reis–Bücklers, and Thiel–Behnke corneal dystrophies?	
	a.	*TACSTD2*
	b.	*TGFBI*
	c.	*UBIAD1*
	d.	*CHST6*

3.	An elderly male presents with recent onset of bilateral progressive corneal stromal deposits resembling granular dystrophy. Histopathologic evaluation of the cornea demonstrates amorphous eosinophilic deposits that stain with Masson trichrome and PAS stains and do not stain with Congo red stain. What is a likely diagnosis?	
	a.	Adult-onset lattice corneal dystrophy
	b.	Atypical granular corneal dystrophy
	c.	Paraproteinemic keratopathy

Scroll to page 3 for answers.

Answers

Question 1 Correct answer and rationale: **a. A well-defined corneal dystrophy in which the gene has been mapped and identified and the specific mutations are known**

The International Classification of Corneal Dystrophies (IC3D) groups dystrophies in 4 categories based on the level of evidence supporting existence of a given corneal dystrophy depending on how substantive was the knowledge of its clinical, pathological, and genetic basis. The category 1 dystrophy definition is outlined in choice a), category 2 (choice b), category 3 (choice c), category 4 (choice d).

Question 2 Correct answer and rationale: **b. *TGFBI***

TGFBI gene mutations are responsible for lattice, granular, Reis–Bücklers, and Thiel–Behnke corneal dystrophies. *TACSTD2* mutations are associated with gelatinous droplike dystrophy, *UBIAD1* mutations with Schnyder dystrophy, and *CHST6* mutations with macular dystrophy.

Question 3 Correct answer and rationale: **c) Paraproteinemic keratopathy.**

Paraproteinemic keratopaty (immunoglobulin corneal deposition) can present with deposits that mimic adult-onset corneal dystrophy. These deposits can occur in any layer of the cornea, are generally not as sharply demarcated as hyaline deposits of granular dystrophy, stain with Masson trichrome like granular dystrophy, stain with PAS like lattice dystrophy, do not stain with Congo red, and do not demonstrate birefringence.