Appendix 1: Expanded Case 4 history, Figure 1, panel D.

Case 4, in Figure 1D, is 46-year old NHW patient XP1BE.[1-3] She died at age 49 of uterine cancer.[1] She had a history of multiple melanoma and non-melanoma skin cancers: the first appeared at age 2. The patient's first NEI ophthalmic exam was at age 19. At that point she had already had an ocular surface squamous cell carcinoma and two basal cell carcinomas surgically removed from the left lids. Due to these surgeries, her eyelids were tight, and she had ectropion of her left lower lid. Her best-corrected visual acuity was 20/80 in the right eye (OD) and 20/300 left eye (OS) due to stromal opacification. By age 28, her vision had deteriorated to 20/400 in each eye (OU), and she underwent corneal transplant OS. Following the transplant the patient had some visual improvement to 20/80 OS, but difficulty with conjunctival injection, blepharitis, and superficial staphylococcus infection of the cornea. She had two conjunctival squamous cell carcinomas removed OD at the age of 30, and a recurrence at age 32 led to complete orbital exenteration OD. At age 35 the patient had a squamous cell carcinoma removed from the cornea OS, and a separate mass described as a fleshy, papillomatous lesion over most of cornea was biopsied and shown to be benign epithelial cells. The ocular surface squamous cell carcinoma recurred at age 36, and the left orbit was exenterated at that time.[4]

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