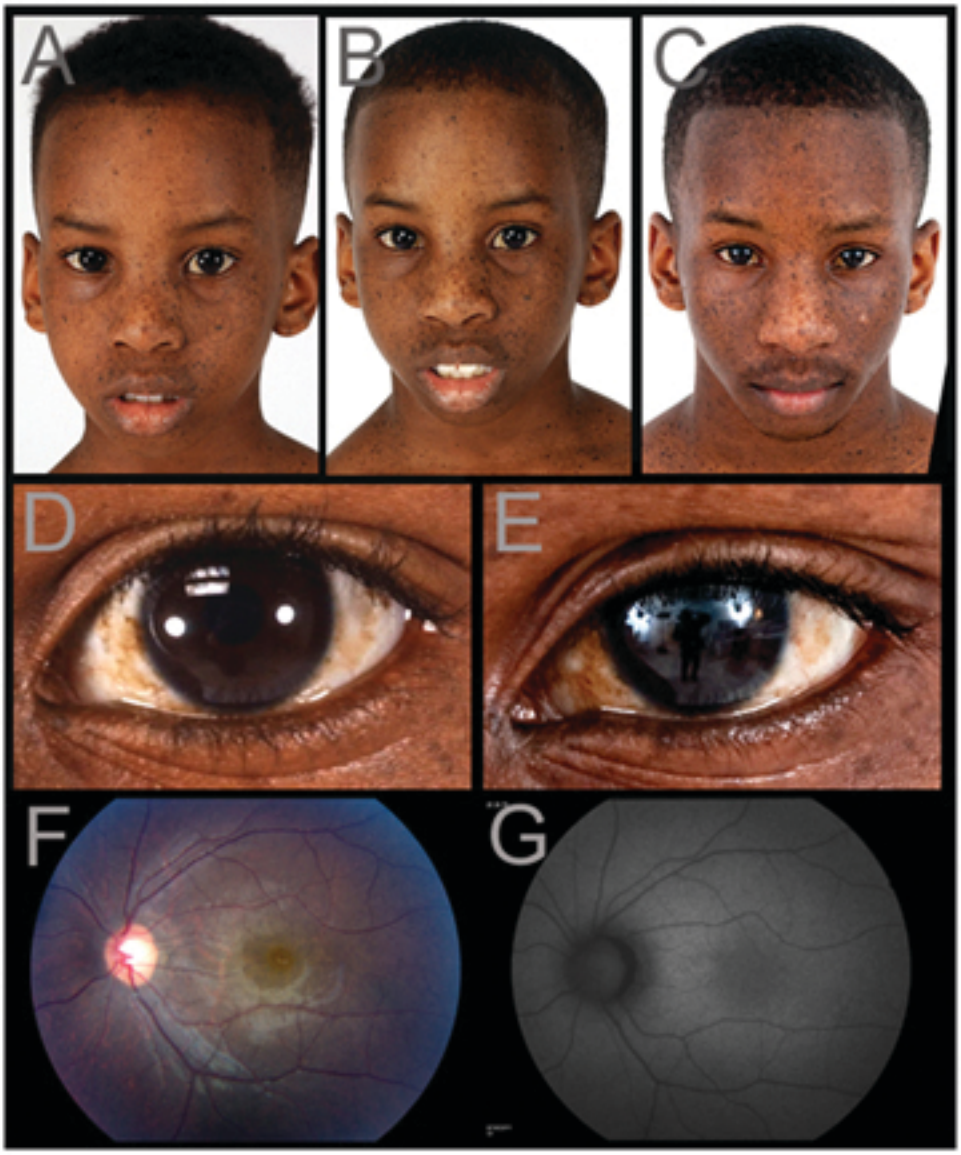

Figure 4: Recent re-classification of a patient from Xeroderma pigmentosum (XP) to XP/Cockayne syndrome complex via information obtained during an ophthalmic exam.

All photos in this figure are Case 7, African American XP-G patient (XP218BE), shown at **(A)** age 10, **(B)** age 13 and **(C)** age 17 years. Conjunctival melanosis OS (left eye) is shown at ages 10 **(D)** and 17 **(E)**. Until visit at age 17, this patient had been classified as XP, however subtle fundus abnormalities observed during that examination (macular granularity with a few yellowish dots and mild arteriolar attenuation) led to further testing. Fundus image **(F)** and fundus autofluorescence image **(G)** from patient at age 17. Static autofluorescence imaging shows diffuse central hyperautofluorescence and is suggestive of increased macular lipofuscin accumulation, given the patient's age. Full-field electroretinogram (ERG) was abnormal (e.g., dark-adapted maximum combined response a wave amplitude = 125-146 μV [normal > 188 μV], b wave amplitude = 236-279 μV [normal > 373 μV], and cone flicker amplitude 52-63 μV = [normal >101 μV]), suggestive of early cone-rod degeneration. The patient also had corneal thickening and specular microscopy abnormalities.