Unblinded by the Lights

A 24-year-old man was transferred to the hospital for evaluation of jaundice and abdominal pain. He had a lifelong history of severe photosensitivity and wore long-sleeved shirts and sunscreen to limit sunburn. During the evaluation for abdominal pain, an initial upper endoscopy was negative. However, postendoscopy melena prompted a second look, which revealed multiple mucosal burns caused by the light source of the endoscope during the initial procedure (Panel A). Burns also developed on his fingers from the light source of a pulse oximeter (Panel B). A workup revealed elevated red-cell free protoporphyrin levels and elevated levels of porphyrin in the stool and urine. A diagnosis of erythropoietic protoporphyria was confirmed with genetic testing. Burns are caused by photoactivation of protoporphyrin deposits in tissues. This patient also has severe liver disease due to hepatic protoporphyrin deposition. He is being evaluated for combined liver and bone marrow transplantation.

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