Case Report

Kayser-Fleischer Rings in a Patient With Basal Cell Carcinoma: Fortuitous Diagnosis of Presymptomatic Wilson’s Disease

DAVID G. BRODLAND, M.D., Department of Dermatology; GEORGE B. BARTLEY, M.D., Department of Ophthalmology

Kayser-Fleischer rings were detected during a routine ophthalmologic workup in a 25-year-old man with basal cell carcinoma of the eyelid. Although the importance of this association is uncertain, the routine preoperative evaluation of basal cell carcinoma led to the presymptomatic diagnosis of Wilson’s disease. These disorders are discussed, as are the benefits of a multidisciplinary approach for the diagnosis and management of medical problems.

Wilson’s disease (hepatolenticular degeneration), a rare inherited metabolic disorder, is characterized by the accumulation of copper predominantly in the liver and brain; this process leads to the primary clinical manifestations of cirrhosis and a wide spectrum of degenerative neurologic symptoms. Deposition of sulfur-rich copper granules within the cornea results in Kayser-Fleischer rings, which are highly suggestive of Wilson’s disease. Cutaneous manifestations are rare. Hyperpigmentation of the legs, azure blue lunulae, and a single episode of a blistering eruption have been encountered in Wilson’s disease. No associated cutaneous neoplasms have been noted. Herein we describe a case of periocular basal cell carcinoma in a young adult man in whom assessment for removal of the tumor and repair of the eyelid led to the fortuitous diagnosis of Wilson’s disease.

REPORT OF CASE

A 25-year-old fair-complexioned man came to the Mayo Department of Dermatology 12 months after he had noted the onset of a papule (6 by 4 mm) on his right lower eyelid. No prior history of skin cancer was elicited; he reported that he was otherwise in good health. Mohs’ micrographic surgery was planned, and an ophthalmologist was consulted for potential reconstruction of the eyelid after removal of the tumor. Slit-lamp biomicroscopy performed as part of a routine ophthalmic examination revealed a prominent ring of corneal pigment in each eye, consistent with Kayser-Fleischer rings. Sunflower cataracts were not present, and no further ocular abnormalities were noted.

The basal cell carcinoma was subsequently extirpated without complication. The patient was then further assessed for Wilson’s disease. A review of his medical history indicated that, 2½ years previously, routine blood studies had shown mildly increased results of liver function tests, which initially were thought to be attributed to ingestion of alcohol. Despite abstention from alcohol, results of repeated studies were unchanged. An evaluation for infectious hepatitis was negative. No underlying cause was determined, and the patient did not return for further assessment at that time.

During the subsequent medical examination, laboratory tests yielded the following abnormal results: alkaline phosphatase, 375 U/liter (normal, 98 to 251); aspartate aminotransferase, 70 U/liter (normal, 12 to 31); creatinine, 1.4 mg/dl (normal, 0.8 to 1.2); total protein, 6.2 g/dl (normal, 6.3 to 7.9); and albumin, 3.2 g/dl (normal, 3.5 to 5.0). The concentration of copper in the serum was low (0.20 μg/ml; normal, 0.75 to 1.45), as was the ceruloplasmin level (2.4 mg/dl; normal, 22.9 to 43.1). The content of copper in the urine was substantially increased (3,235 μg per specimen; normal, 15 to 60). The prothrombin time was increased (14.6 seconds), and the patient had mild thrombocytopenia (platelet count, 155,000/mm³). Because the prothrombin time was uncorrectable with intramuscularly administered vitamin K, a radioactive metabolic study for copper was done instead of a liver biopsy. This study showed that the...
patient was unable to incorporate copper and form ceruloplasmin; these findings were thought to be consistent with homozygous Wilson’s disease. Results of a neurologic examination were normal. The family had no history of disorders of the liver or central nervous system or psychiatric disease.

A regimen of trientine hydrochloride (250 mg four times a day) was initiated, and a low-copper diet was begun. Subsequent studies showed the concentration of copper slowly returning toward normal. Six months after Wilson’s disease was diagnosed, the patient was asymptomatic. (The patient’s siblings are also being assessed for the disease.)

DISCUSSION

Wilson’s disease is a rare disorder that predominantly affects young persons in the first or second decade of life. Basal cell carcinoma is the most common cutaneous cancer, but it occurs most frequently in older persons. Although basal cell carcinoma is known to occur in young adults and even in children, it rarely does. In our patient, the concurrent presence of Wilson’s disease and basal cell carcinoma was probably coincidental. No clinical association between Wilson’s disease and malignant involvement has been reported. Likewise, no evidence exists that copper is carcinogenic or cocarcinogenic. In fact, some investigators suggest that deposition of copper may actually be protective against hepatocellular carcinoma in patients with Wilson’s disease. Deposition of copper is known to exert a toxic effect on many organs, including the hepatic and neural tissue, kidney, skeletal system, parathyroid glands, and components of the integument. The histologic observation of copper within and around Descemet’s membrane of the cornea is notable. No copper was found in the epidermis of patients with Wilson’s disease although increased levels have been noted in fibroblasts. In our patient, a rhodanine stain for copper in the skin was negative.

The benefit of an interdisciplinary approach to patient care is exemplified in the current case by the diagnosis of a potentially debilitating or even fatal disease in its presymptomatic stage as a result of a thorough routine ophthalmologic examination. This report also illustrates the importance of a high index of suspicion when unexplainable liver abnormalities are noted in a young person.

CONCLUSION

A cause-and-effect relationship between basal cell carcinoma and Wilson’s disease is only speculative. A closer inspection of the effects of deposition of copper on the epidermis, however, is warranted because of the known toxic effects of copper on other organs.

REFERENCES