The eye examination, the key diagnosis in a rare case of Wilson disease

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Abstract: We report of a 19-year-old man complaining of clumsy gait, drooling of saliva, difficulty in speech, and behavioral changes who underwent ophthalmic examination and was diagnosed with a rare case of Wilson disease: Kayser-Fleischer ring and unilateral hypertrophy of retinal pigment epithelium.

Key words: Hypertrophy of retinal pigment epithelium, Wilson disease, Kayser-Fleischer ring.

Introduction
Wilson disease is a rare (1/40000- 1/100000) autosomal recessive disorder, characterized by degenerative changes mainly in liver and brain.1 There are several aspects which make this disease particularly interesting and important for neurologists, gastroenterologists and pediatricians, mainly because of the drastically varied clinical presentation that leads to diagnostic difficulties.2 Diagnosis may be missed at early stages, and the disease will be progressive and fatal if it is left untreated. Therefore, applying clinical findings in early stages of treatment could prove to be a beneficial way to establish the diagnosis.

Case report
The consanguineous parents of a 19-year-old male watched their boy’s mood swings. He had recently shown aggressive behavior, anger and high sensitivity to voice. Their physician’s diagnosis was stomach reflux, but treatment exacerbated the reflux. The patient gradually developed Parkinson symptoms and signs (clumsy gait, drooling of saliva, difficulty in speech and behavioral changes) and was admitted in the internal ward. The liver CT-scan was within normal range and ultrasonography showed a suspicious lesion which had not been revealed via multiple liver biopsies for pathologic findings. The serum caeruloplasmin ratio was 13 mg/dl and 24-hour urine copper was 275 µg/day.

Appearance of initial signs of subnormal intelligence takes about six years until a fully developed classical central nervous system is found. The patient’s voice hoarseness, dysarthria, excessive salivation, shuffling gait with short steps, and eating and swallowing difficulty were signs which strengthened the suspicion.

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of Wilson disease. The other cranial nerves were normal without associating any other systemic findings. The disease remained undiagnosed during the first six years. Finally, the patient was visited by an internist in this hospital. A provisional diagnosis of Wilson disease was made and the case was investigated accordingly. The suspected diagnosis was Wilson disease and he began D-Penicillamine for disease management, which showed good therapeutic response. No abnormalities in other organs were found, but ophthalmic consultation revealed a greenish brown ring at the limbus in both eyes and hypertrophy of retinal pigment epithelium (Figs. 1, 2).

Discussion

Wilson disease must be suspected in patients who show unexplained and unjustified mood swings. The patient was suspected due to appearance of abnormal neurological symptoms and extra pyramidal manifestations. Although neurological manifestations are rare before the age of 20, this patient had earlier manifestations; hence ophthalmic findings mainly established the diagnosis. Hypertrophy of retinal pigment epithelium, in this case, could be an incidental finding and we are waiting to hear from other studies.

Although this is, to our knowledge, the first finding, it could be a significant one, because as described by Krajacic et al., retinal epithelial abnormality in mice was due to copper metabolism in Wilson disease.3

Patients having original clinical descriptions such as cirrhosis, neurological finding, Kayser-Fleischer rings and decreased serum ceruloplasmin are generally considered as having classic Wilson disease. However, about half of the patients diagnosed with liver disease, do not show these criteria and pose a challenge for establishment of diagnosis.4

Ronald F. Pfeiffer claims that the ophthalmic manifestation of Kayser-Fleischer rings is always present in persons with Wilson’s disease who have developed neurological or psychiatric dysfunction. Among other rare reported ocular manifestations was classic sunflower cataract in Wilson disease.5 Recognition of Kayser-Fleischer rings and retinal degeneration played a crucial role in the diagnosis of Wilson
disease by ophthalmic examination.\textsuperscript{6,7} Therefore, variable funduscopic findings such as congenital choroidal cloboma in the left fundus and normal right fundus\textsuperscript{8} and retinal degeneration can play a useful role in the diagnosis of Wilson disease, but this is yet to be confirmed.

References