WILSON'S DISEASE (HEPATOLENTICULAR DEGENERATION) MEDICAL APPENDIX

DEFINITION

1. Wilson's disease is characterised by an accumulation of copper especially in the liver, brain, kidneys and cornea.

CLINICAL MANIFESTATIONS

2. Although it may present at any age, most patients are first seen between the ages of 8 and 25. Presenting manifestations are very variable. Most commonly they are hepatic, neurological or psychological.

3. **Hepatic** symptoms and signs may be acute but more commonly they are gradual. Cirrhosis ultimately develops. **Neurological** signs include dystonic features affecting the face or upper limbs, an unsteady gait and tremor or loss of coordination of fine movements. As the disease progresses, dysarthria, rigidity, drooling and titubation may develop. **Psychological** symptoms include intellectual deterioration, personality change and unstable behaviour. **Rare manifestations** include cholelithiasis, haemolytic crisis, arthropathy, renal calculi and the Fanconi syndrome.

4. Although the diagnosis can be made clinically, it is confirmed by the identification of an abnormality of copper transport and storage.

5. Kayser Fleischer rings in the eyes are diagnostic but are not present in all cases. They consist of golden brown or greenish rings or arcs due to the deposition of copper in the cornea.

AETIOLOGY

6. It is a genetic disorder, being inherited as an autosomal recessive trait. Excretion of copper by the bile (the normal route) is impaired and copper accumulates in the body. The primary genetic defect has not been identified. Without treatment using a chelating agent the disease is progressive. Otherwise environmental factors do not influence the course of the disease.

CONCLUSION

7. Wilson's disease is a hereditary disorder of copper metabolism causally unrelated to external factors, whose progress, in the absence of treatment, is unrelated to environmental factors.

REFERENCES

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