Autosomal recessive inheritance of hereditary motor and sensory neuropathy with optic atrophy


16 Harding AE. From the syndrome of Charcot, Marie and Tooth to disorders of peripheral myelin proteins. Brain 1995;118:809-18.


HISTORICAL NOTES

Johann Jakob Wepfer (1620–95) and cerebral haemorrhage

The Swiss physician Wepfer showed that apoplexy is due to cerebral haemorrhage. In Historiae apoplectico-rum, published in 1658,1 is a detailed description of four cases, his first case having been studied in 1655:

"Johann Jacobus Reiter Kenzinga-Brisgojus, age about 45 years, with a slender build, endowed with yellow and curly hair, naturally strong; of honest parents, descended from the Consul. He acted as schoolmaster and at length was admitted to a most celebrated Monastery. He suffered from extensive gouty arthritis.

In the year 1655, the seventh day of November... he did much of everything—assisted the Most Reverend Lord Abbott in the carrying out of sacrifices; latter that day the Abbott by chance found him prostrate upon the ground, insensible to shouts, to shaking and pinching... I was summoned.

I arrived in half an hour, I saw him livid from pallor, deprived of all sensation and animate motion, with nostrils cold to the touch. His pulse at first strong, full, quick, soon afterwards weaker, smaller and more frequent, his breathing also most laborious, soon it became irregular, and many times appeared about to cease from within... At the tenth hour before midday his body was shaken albeit by a movement and much sputum white, viscid, tenacious passed from his mouth, but indeed no blood: after this more and more his strength began to weaken and his extremities to become more cold; The first hour after midday he ceased to live...

I opened the head: the skull removed and the dura mater being cut into pieces much blood flowed from the space between this and the thin meninges, copiously... Not truly had the blood collected solely at the base of the brain, but covered it all over the top both anteriorly and posteriorly, indeed it had forced itself into nearly all the windings of the brain, as many as there are: extravasated blood totalled two pounds. The whole brain, ventricles and surface were contaminated by blood in large amounts and crumbly... I was able to find no ruptured vein or artery. This however is certain, no external violent cause, be it a blow, be it a fall, was the cause of such ruptures of the blood vessels; to settle this point with his hair cut and skin washed off he showed not the slightest trace of any contusion whatever."

Historiae apoplecticorum also contains "The history of the sickness of Marcello Malpighi, the Pope's physician; with an account of the dissection of his corps". Wepfer was one of the earliest investigators of the cerebral vasculature. His book on diseases of the head provided an ample account of basilar artery migraine and its location in the brainstem, a description of a stroke in migraine in a child; and, he wrote of the visual auras and diagnosis of migraine. He also described trigeminal neuralgia localised to the maxillary nerve.

Born in Schaffhausen, Switzerland in 1620, son of the Councillor of the local Canton, Wepfer read medicine publishing for his degree in 1647 De palpitatione cordii, which stoutly supported Harvey's controversial ideas on the circulation of the blood. He studied in Basle, Strasbourg, and Italy, but returned to his native town where he built a successful practice. His renown spread widely, and his name was hallowed as "The Hippocrates of Helvetia."

A man of temperate habits, he was a most industrious scholar, devoted to reading and the scriptures. His son Johann Conrad and his grandson Georg Michael became physicians. His considerable library and writings were bought by the University of Leyden in 1774 for 400 gold florins.

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