Diagnosis of paediatric Huntington's disease relies on imaging findings

Savvas Andronikou
MB BCh, FCRad(D), FRCR (Lond)

Conrad Nel
MB ChB

Department of Paediatric Radiology
Red Cross War Memorial Children's Hospital
University of Cape Town and Institute of Child Health

Introduction

Huntington's disease is an autosomally inherited neurodegenerative disorder clinically characterised by disturbances of movement, mentation and behaviour. Onset is typically in the 5th and 6th decade and symptoms can sometimes occur in an affected child before appearing in the affected parent, who is most likely to be the father. Five per cent of patients present under the age of 14 years. Juvenile presentation differs from that of adults, who usually show abnormal movements; in children there may be rigidity, dysarthria, progressive mental deterioration and convulsions. Convulsions occur frequently but late in the course of the disease. This should be borne in mind when investigating a child with 'epilepsy'.[1]

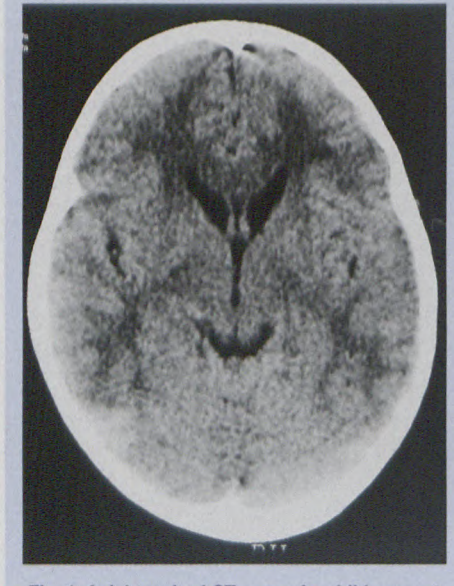

Fig. 1. Axial cerebral CT scan of a child presenting with seizures.
Level of the quadrigeminal plate cistern.

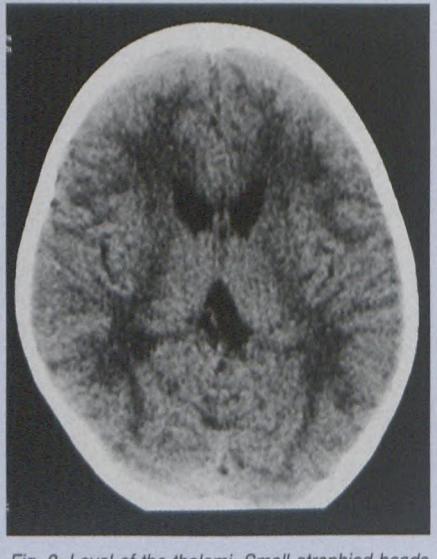

Fig. 2. Level of the thalami. Small atrophied heads of the caudate nuclei with resultant squaring and expansion of the frontal horns giving the 'box-like' appearance. These features are diagnostic of Huntington's disease.

Pathology and radiology

Both computed tomography (CT) and magnetic resonance imaging (MRI) can show striking basal ganglia atrophy (typically volume loss of the head of the caudate nucleus).[1,2] Secondary focal enlargement of the frontal horns of the lateral ventricles (the lateral walls are convex towards the shrunken caudate nuclei) results in their progressively squared, 'box-like' appearance. This is best appreciated on coronal images but these are not performed as a routine on CT scanning and therefore the radiologist must be able to appreciate this sign on axial images (Figs 1 and 2). A ratio of maximum width of the frontal horns to intercaudate distance < 1.6 is considered diagnostic. Increased and decreased putaminal signal intensities on MRI have been reported. Also common are associated atrophy of the cerebellum and brain stem.[1,2]

Conclusion

Because the above imaging findings may be encountered during imaging for 'epilepsy' without clinical suspicion for Huntington's disease, and because the parental disease may not have manifested clinically as yet, it is of utmost importance that the radiologist be familiar with the characteristic imaging findings. A rapid accurate imaging diagnosis will allow for genetic counselling prior to the child reaching reproductive maturity.

References

1. Osborn AG. *Diagnostic Neuroradiology.* St Louis: Mosby, 1994: 743-744.
2. Kirkwood RJ. *Essentials of Neuroimaging.* New York: Churchill Livingstone, 1990: 316.