Diagnosis of paediatric Huntington’s disease relies on imaging findings

**Introduction**

Huntington’s disease is an autosomally inherited neurodegenerative disorder clinically characterised by disturbances of movement, mentality 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’.

**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). 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.

**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**