Abnormal MRI signal in the rigid form of Huntington's disease

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Abstract
Eighteen patients with Huntington's disease (HD) were examined with MRI. Eleven had the common hyperkinetic form, seven had the rigid variant. All seven patients with rigid HD had increased signal intensity in the neostriatum in intermediate and T2-weighted images. Only one hyperkinetic HD had similar findings. In all the other cases, signal abnormalities were questionable or absent. Histological differences that account for differences in signal intensity may therefore lie in the caudate nucleus and putamen. Age of onset may be important, since the rigid patients were younger. Follow up studies may help in understanding these signal differences.

Huntington's disease (HD) is a disabling degenerative disease with autosomal dominant inheritance, clinically characterised by involuntary hyperkinesia, affective-emotional disturbances and intellectual deterioration, usually beginning between 35 and 45 years of age. HD clinical features may be variable and an atypical form mainly characterised by plastic rigidity and a Parkinsonian-like posture has been described.[1] This "rigid" variant of HD is more common in young patients, but similar cases have also been observed in adults. The classic hyperkinetic HD may often evolve toward an akinetic state.[1]

Gross pathological changes include diffuse cerebral atrophy and, more characteristically, atrophy of the striatum. The head of the caudate nucleus shrinks considerably and the profile of the frontal horn of the lateral ventricle therefore becomes flattened. The putamen is also reduced in size, and, less commonly and to a lesser degree, the pallidum may be affected.[2]

Atrophy of the head of the caudate nucleus with enlargement of the frontal horn, the most characteristic aspect of HD, was demonstrated in vivo even by pneumoencephalography. It is demonstrated by CT and MRI. Density changes have not been reported on CT, nor has abnormal increased signal intensity been reported on MRI studies.[3][4] MRI is better for demonstrating, in transverse and in coronal sections, the atrophy of the head of the caudate nucleus. No signal abnormalities are mentioned in a series of 12 patients with HD by Kido *et al*;[3] decreased signal intensity in the neostriatum in T2-weighted images at 1·5 T was described by Rutledge *et al* in four cases, consistent with increased iron content.[4]

We have recently observed high signal intensity in T2-weighted images in cases with the rigid variant of HD. We report a series of patients with HD, in which we compare the signal abnormalities of the cases with the rigid variant versus the cases with the classic hyperkinetic form.

Methods
We reviewed a series of 18 patients with HD. Eleven patients had the typical hyperkinetic form and seven had the rigid variant. All the patients had a family history of HD, as proved by examination of other affected members of the family and a review of medical records. Diagnosis of rigid HD was made according to Bruyn and Went's criteria, when a definite rigidity was observed. To our knowledge, no necropsy had been performed on other members of the affected families.

Of the 11 hyperkinetic HD patients, seven were males, and four females. Age ranged from 17 to 63 years (mean 45·2 years); disease duration ranged from one to 14 years (mean 5·1 years).

Of the seven rigid HD patients, three were males, four females. Age ranged from 14 to 47 years (mean 28·8 years). Disease duration ranged from three to nine years (mean 5·7 years). These patients presented rigidity and bradykinesia. In three subjects, postural tremor was observed; in four cases, mild hyperkinesia involved face and distal segments of the limbs.

Physical disability was estimated by the Shoulson and Fahn rating scale.[5] Mean (SD) disability rating was 6·42 (3·55) for the rigid form, and 7·09 (3·20) for the hyperkinetic form.

All patients were studied with MRI 0·5 T. One patient with the rigid form was also examined with a 1·5 T unit. All the examinations included sagittal T1-weighted images and transverse SE intermediate and T2-weighted images. Occasionally T1-weighted images in coronal or transverse planes were available.

Results
All patients presented with atrophy, both diffuse and focal, particularly involving the head of the caudate nucleus (fig 1). No differences

Figure 1 HD, hyperkinetic form. (A) Severe atrophy of the head of the caudate nucleus (arrowheads) in coronal section in a 42 year old male. (B) Atrophy of the putamen (arrowheads) in transverse section in a 57 year old female. No signal abnormalities in both cases (SE 2100/50); compare the signal intensity of the neostriatum with that of the insular cortex.
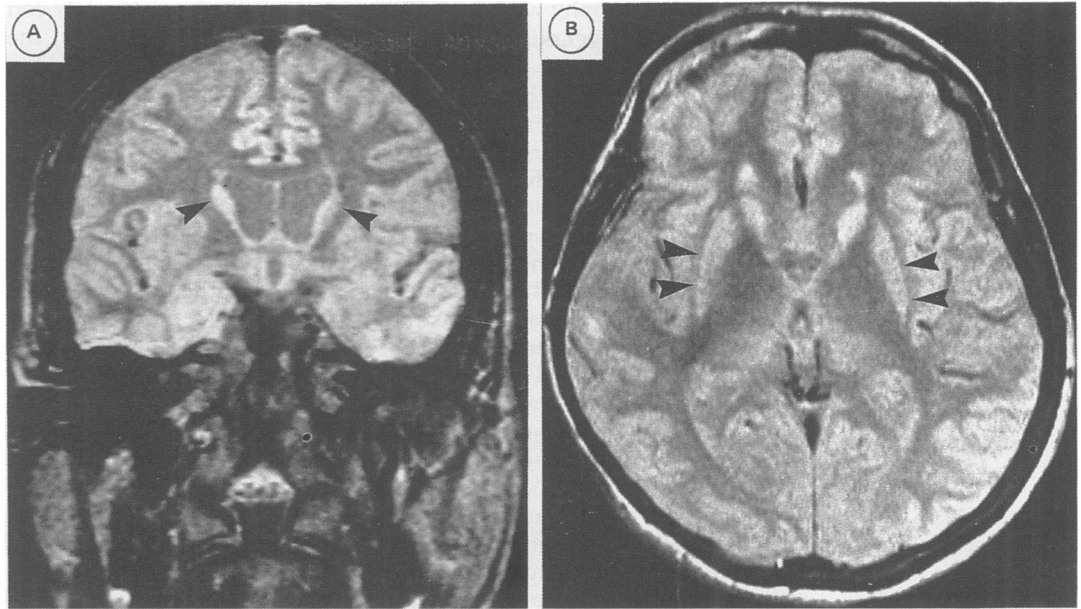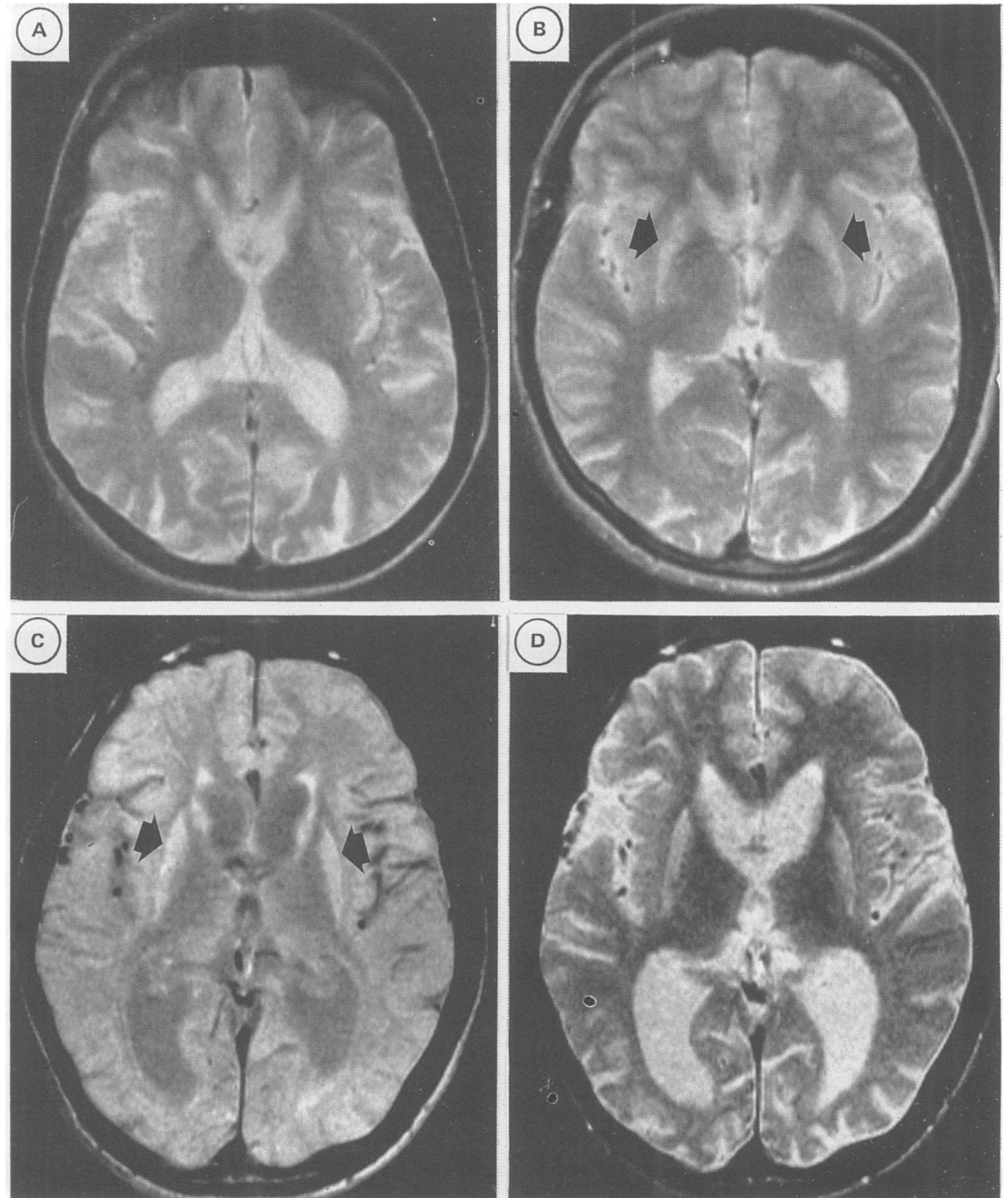

Figure 2 (A) Hyperkinetic HD in a 43 year old female. No abnormal signal intensity in T2-weighted image (SE 1933/100). (B) Rigid HD in a 23 year old female. Slightly increased signal intensity in T2-weighted image (SE 2100/100), (arrows). (C) and (D) Intermediate and T2-weighted images (SE 2100/50, 100) in a 28 year old male with rigid HD; increased signal intensity in the neostriatum (arrows) is more evident.

reported that the forms of HD with juvenile onset are associated with a more marked cell loss and atrophy of the neostriatum.[11]

In conclusion, the signal hyperintensity in the neostriatum described here is not exclusively observed in the rigid form of HD, but seems to be a characteristic of this variant; it is very rare in the classic hyperkinetic form.

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