# MRI IN A CASE OF HALLERVORDEN SPATZ DISEASE

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## SUMMARY

The cranial magnetic resonance imaging (MRI) of a 12-year-old child with a clinic presentation suggestive of Hallervorden-Spatz disease (HSD) is described. Progressive dystonia, dysarthria and pyramidal signs were present for 2 years. The presence of both increased and decreased signals in globus pallidus was consistent with the pathology of the disease: loss of myelin and increased iron deposition in the basal ganglia.

**Key Words :** Extrapyramidal disorder, Hallervorden-Spatz disease, magnetic resonance imaging.

## INTRODUCTION

The disease was first described by Hallervorden and Spatz in 1922. Inherited as an autosomal recessive trait, the onset is in childhood or adolesance. Extrapyramidal signs such as dystonia, rigidity and choreathetosis are combined with pyramidal signs. As the movement disorder slowly progresses, the child becomes dystonic, dysarthric and finally completely mute and disabled. A certain degree of dementia is eventually added. Optic atrophy is also reported in some cases. The distinctive pathology of Hallervorden-Spatz disease (HSD), i.e. deposition of iron and demyelination in basal ganglia cannot be corroborated by any of the present laboratory test (1). Radioisotope scanning and CT have been used as diagnostic aids. However, magnetic resonance imaging (MRI) is more effective in demonstrating the iron deposits in basal ganglia, and the characteristic clinical findings combined with MRI make an antemortem diagnosis possible (2-7).

## CASE REPORT

The patient was a 12-year-old child born of consanguineous parents. Family history revealed no neurological abnormality. His birth, motor and mental development were reported to be normal.

At age 10, he had first complained of difficulty in writing. He had generalized tonic-clonic seizures and was using diphenylhydantoin. A CT scan performed during that period showed bilateral small hyperdense regions in the basal ganglia. The patient was first seen at age 11, one year after the onset of the symptoms. Choreiform movements of the fingers and toes more prominent on the right side of body, and tremor and occasional dystonic movements of the right hand were also present. There was a slight weakness of the flexion movements of the fingers in the left hand. Deep tendon reflexes were brisk and plantar responses were extansor. IQ was 105. Ophthalmologic examination revealed no abnormality. Optic atrophy, retinopathy, Kayser-Fleischer ring were absent. Laboratory examination including blood smear, electrolytes, ceruloplasmin, parat hormon, liver function tests, creatin clerence, urinary amino acids were all normal. EEG was also normal. Hyperdense regions in globi pallidi persisted on the second CT scan.

During the follow-up period, dystonic movements slowly progressed. Lips, tongue and the left hand were eventually involved. 10 months after his first hospitalization, at age 12, there was an acute and dramatic progression of symptoms. Severe axial and appendicular dystonia made walking impossible. Grimacing was present. Speech and swallowing were completely interrupted. Administration of diazepam and baclofen relieved the symptoms considerably.

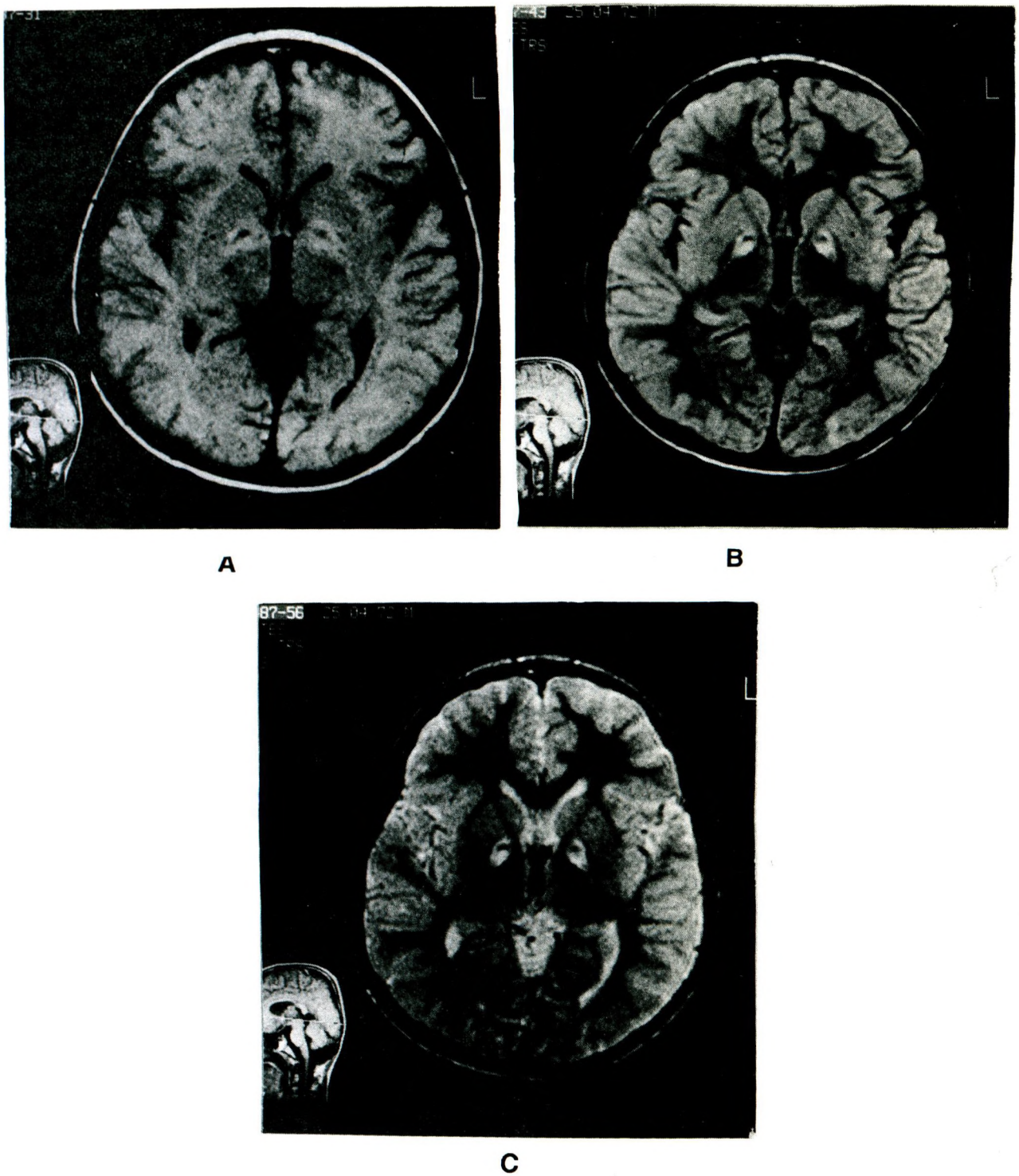


Fig. 1. (A) T1-weighted axial MR image (TR=500 msec TE=38 msec) show decreased signal intensity in the globus pallidus (B) intermediate (TR=2100 msec TE=38 msec) (C) T2-weighted axial MR image (TR=2100 msec TE=110 msec) show the presence of both increased and decreased signal in the globus pallidus.

Walking and speech improved slowly. Dystonia of the right hand, slight dysarthria and grimacing and occasional dystonia remained. During the last 10 months the symptoms showed a very slow progression and the disease followed a rather stationary course.

**MRI Results :** During the patient's second hospitalization, at age 12, MRI was performed using a 0.2 tesla Unit (HITACHI). SE technique was employed : T1 (TR=500msec TE=38 msec) intermediated (TR=2100msec TE=38msec), T2 (TR=2100 msec TE=110 msec). A slice thickness of 7.5 mm was used with a 256x256 matrix size.

There was a small central hypointense region in the globus pallidus on T1, intermediate and T2 images. Rest of globus pallidus appeared hyperintense on T1, intermediate and T2 weighted images (Fig.1).

## DISCUSSION

The clinical presentation of the patient and absence of any pathological laboratory findings are suggestive of HSD. The hyperdensity in the basal ganglia is similar to some CT findings reported previously and presumably corresponds to the accumulation of iron and calcium. Hypodensity in the globus pallidus, diffuse atrophy and normal CT in HSD are reported as well (4, 5, 8-10).

There are also very few reports on MRI in HSD. Findings of "decreased signal intensity" in the globus pallidus, best seen on T2-weighted images are reported (2-7). In a pathologically proven case this is shown to correspond to iron deposition. Small, hyperintense regions were also seen and they were shown to represent demyelination (4).

The opposite is true for our case. Hyperintensity is "prominent" and "peripheral", while signal decrease is "small" and "central". MRI of our patient was obtained 2 years after the onset of the symptoms. Most of previous MRI findings in HSD belong to patients with much longer durations of the disease and repeat MRI's do not show any difference. Galucci and friends report a young case at a rather earlier stage of the disease, and hypointensity is shown to increase over the years (11). We think the signal decrease in our patient will also become prominent, as the, disease evolves. As for the central site of the signal decrease, it is also most likely due to the stage of the disease, and therefore may have no further implications. However the role of imaging at a low field,strength -if there is any- on such an alteration should also be investigated.
In conclusion, we believe MRI follow up this patient may allow us to follow the pathological changes during the life time.

Besides repeat MRI's with a higher Tesla unit may also make it possible to compare the effects of iron in MRI at different field strengths.

## REFERENCES


1. *Adams R, Victor M. (eds) Hallervorden-Spatz disease. In: Principals of neurology 4th edition. Singapore: Mc Graw Hill Inf Services Company 1989: 804-805.*
2. *Littrup PJ, Gebarski SS. MR imaging of Hallervorden-Spatz disease. J Comput Assist Tomogr 1985; 9: 491-493.*
3. *Drayer B, Burger S, Darwin M, Riederer S, Hefkens R, Johnson G. Magnetic resonance imaging of brain iron. J Comput Assist Tomogr 1986; 7: 373-380.*
4. *Rutledge N, Hilal S, Silver J, Defendini R, Fahn S. Study of movement disorders and brain iron by MR. AJNR 1987; 8: 397-411.*
5. *Schaeffert DA, Johnson SD, Johnson PC, Burton P, Drayer BP. Magnetic resonance imaging in pathologically proven Hallervorden-Spatz disease. Neurology 1989; 39: 440-442.*
6. *Mutoh K, Okuno T, Masatoshi I et al. MR imaging of a group 1 case of Hallervorden-Spatz disease. J Comput Assist Tomogr 1988; 12: 851-853.*
7. *Tanfani G, Mascalchi M, Dal Pozzo GC-Taverni N, Saia A, Trevison C. MR imaging in a case of Hallervorden-Spatz disease. J Comput Assist Tomogr 1987; 11: 1057-1058.*
8. *Bolthauser E, Lang W, Janzer R et al. Computed tomography in Hallervorden-Spatz disease. Neuropediatrics 1986; 18: 81-83.*
9. *Dooling EC, Richardson EP, Davis KR. Computed tomography in Hallervorden-Spatz disease. Neurology 1980; 30: 1128-1130.*
10. *Tennison MB, Bouldwin TW, Whaley RA. Mineralization of basal ganglia detected by CT in Hallervorden-Spatz syndrome. Neurology 1988; 38: 154-155.*
11. *Galluci M, Cordana F, Arachi M, Splendiani A, Bozzao A, Passariello R. Follow-up MR studies in Hallervorden-Spatz disease. J Comput Assist Tomogr 1990; 14: 118-120.*