Case report:

HALLERVORDEN - SPATZ DISEASE

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Case No1:

- 15 year old girl.
- Since about 1 year and a half:
  - Weakness of the right hemi-body.
  - Increasing rigidity of the right arm and slowing of voluntary movement.
  - Dysarthria
  - Mental deterioration
- Hematological exam: normal.
- Abdominal echography: normal.
- EMG: Normal.

→ Clinical suspicion of the white matter disease → MRI
Conclusion: Metabolic disorder affecting deep gray matter.

→ HALLERVORDEN-SPATZ DISEASE
Case No2:

- 10 year old girl
- Not go to school?
- Hypertonicity and rigidity of 4 limbs progressive from 1 month.
- Hematological and biochemical exam: normal
  - Suspicion: WM disease
  - MRI of the head.
Diagnosis:
HALLERVORDEN-SPATZ DISEASE
HALLERVORDEN-SPATZ DISEASE

- Hallervorden and Spatz: 1924
- Neurodegeneration with brain iron accumulation type 1" (NBIA-1)
- Inheritance: chromosome 20 - 20p12.3-13
- Mutation \textit{PANK2} gene
- Abnormal iron accumulation in the brain
Onset in the second decade. Die in the second or third decade.

Progressive dementia, corticospinal signs and extrapyramidal signs

No biochemical markers have been found in HSD
Diagnostic criteria: Swaiman, 1991

All of the obligate findings and
At least 2 of the corroborative findings
None of the exclusionary factors
The obligate findings

- Onset during the first 2 decades of life
- Progression of signs and symptoms
- Evidence of extrapyramidal dysfunction including one or more of the following: dystonia, rigidity, choreoathetosis
The corroborative findings

- Corticospinal tract involvement
- Progressive intellectual impairment
- Retinitis pigmentosa and/or optic atrophy
- Seizures
- Positive family history consistent with autosomal recessive inheritance
- Hypointense areas on MRI involving the basal ganglia
- Abnormal cytosomes in circulating lymphocytes and/or sea-blue histiocytes in bone marrow
The exclusionary factors

- Abnormal ceruloplasmin levels and/or abnormalities in copper metabolism
- Presence of overt neuronal ceroid lipofuscinosis as demonstrated by severe visual impairment and/or seizures that are difficult to control
- Predominant epileptic symptoms
- Severe retinal degeneration or visual impairment preceding other symptoms
- Presence of familial history of Huntington chorea and/or other autosomal dominantly inherited neuromovement disorders
- Presence of caudate atrophy on imaging studies
- Deficiency of hexosaminidase A
- Deficiency of ganglioside monosialic acid-1 (GM1)-galactosidase
- Nonprogressive course
- Absence of extrapyramidal signs
Imaging study:

- CT imaging is not very helpful
- MRI:
  - Hypointensity in the globus pallidus on T2WI
  - Foci of hyperintensity on T2WI represent destruction and gliosis.
  
  "eye of tiger" sign.

- "Dif diagnosis: oculodigital dental dysplasia:
  Whitte matter (++)"
`Eye of tiger` sign
### Anatomic distribution of some diseases affecting basal ganglia


<table>
<thead>
<tr>
<th>Diagnosis</th>
<th>Globus palladus</th>
<th>caudate</th>
<th>Putamen</th>
<th>WM</th>
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</thead>
<tbody>
<tr>
<td>Acute</td>
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<tr>
<td>Chronic:</td>
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<tr>
<td>Mitochondrial disorder</td>
<td>+</td>
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<tr>
<td>Canavan’s disease</td>
<td>+</td>
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<td>++</td>
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<tr>
<td>GM2 gangliosidoses</td>
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<td>++</td>
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<tr>
<td>Juvenile Huntington’s</td>
<td>-</td>
<td>++</td>
<td>++</td>
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<tr>
<td><strong>Wilson’s</strong></td>
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<tr>
<td>Glutaric aciduria type I</td>
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<tr>
<td>Glutaric aciduria type II</td>
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<tr>
<td>Methylmalonic acidemia</td>
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<td><strong>Hallervorden-Spatz</strong></td>
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<tr>
<td>Huntington’s</td>
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“The finding of T2 shortening in the globi palladi during the second decade should raise strong suspicion for the diagnosis of Hallervorden-Spatz disease rather than any of the many disorders affecting the basal ganglia”.
Treatment

- Treatment remains directed toward symptomatic findings.
- Systemic chelating agents such as desferrioxamine have not proved beneficial.
References

- Emedicine