

Open Peer Review on Qeios

## Hypomyelination with atrophy of basal ganglia and cerebellum

**INSERM** 

## Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base.

Hypomyelination with atrophy of basal ganglia and cerebellum. ORPHA:139441

Hypomyelination with atrophy of basal ganglia and cerebellum (H-ABC) is characterized by slowly progressive spasticity, extrapyramidal movement disorders (dystonia, choreoathetosis and rigidity), cerebellar ataxia, moderate to severe cognitive deficit, and anarthria/dysarthria.

Qeios ID: ELN9VC · https://doi.org/10.32388/ELN9VC