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# Macrocephaly-spastic paraplegia-dysmorphism syndrome

INSERM

## Source

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

*Macrocephaly-spastic paraplegia-dysmorphism syndrome. ORPHA:2429*

Macrocephaly-spastic paraplegia-dysmorphism syndrome is a rare syndrome of multiple congenital anomalies characterized by macrocephaly (of post-natal onset) with large anterior fontanelle, progressive complex spastic paraplegia, dysmorphic facial features (broad and high forehead, deeply set eyes, short philtrum with thin upper lip, large mouth and prominent incisors), seizures, and intellectual deficit of varying severity. Inheritance appears to be autosomal recessive.