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# Pontocerebellar Hypoplasia Type 2

National Cancer Institute

## Source

National Cancer Institute. *Pontocerebellar Hypoplasia Type 2*. NCI Thesaurus. Code C124057.

A rare brain developmental disorder caused by mutations in the TSEN54, TSEN2, TSEN34, or SEPSECS gene. The pons and cerebellum are the brain structures that are more severely affected. It is characterized by microcephaly, lack of voluntary motor skills, dysphagia, inability to communicate, abnormal patterns of movement, and spasticity.