

Open Peer Review on Qeios

## Angelman Syndrome

National Cancer Institute

## Source

National Cancer Institute. <u>Angelman Syndrome</u>. NCI Thesaurus. Code C75462.

A genetic syndrome characterized by mental retardation, speech impairment, microcephaly, ataxia, and seizures. The majority of cases result from deletions on the long arm of chromosome 15. A minority of cases result from mutations in the UBE3A gene.

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