Tagged results:

Move the mouse over the entity to display the HPO id.

The clinical features of Angelman syndrome (AS) comprise severe mental retardation, postnatal microcephaly, macrostomia and prognathia, absence of speech, ataxia, and a happy disposition. We report on seven patients who lack most of these features, but presented with obesity, muscular hypotonia and mild mental retardation. Based on the latter findings, the patients were initially suspected of having Prader-Willi syndrome. DNA methylation analysis of SNRPN and D15S63, however, revealed an AS pattern, ie the maternal band was faint or absent. Cytogenetic studies and microsatellite analysis demonstrated apparently normal chromosomes 15 of biparental inheritance. We conclude that these patients have an imprinting defect and a previously unrecognised form of AS. The mild phenotype may be explained by an incomplete imprinting defect or by cellular mosaicism.

Phenobert Results

HPO_id	Given Term
HP:0010864	severe mental retardation
HP:0005484	postnatal microcephaly
HP:0000154	macrostomia
HP:0000303	prognathia
HP:0001344	absence speech
HP:0001251	ataxia
HP:0100024	happy disposition
HP:0001513	obesity
HP:0001252	muscular hypotonia
HP:0001256	mild mental retardation

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Phenotagger Results

HPO_id	Given Term
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