Tagged results:

Move the mouse over the entity to display the HPO id. Phenotagger results are highlighted. PhenoBERT results are underlined.

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.

Results

HPO ID	Given Term	Official Term
HP:0010864	severe mental retardation	intellectual disability, severe
HP:0005484	postnatal microcephaly	postnatal microcephaly
HP:0000154	macrostomia	wide mouth
HP:0000303	prognathia	mandibular prognathia
HP:0001344	absence speech	absent speech
HP:0001251	ataxia	ataxia
HP:0100024	happy disposition	conspicuously happy disposition
HP:0001513	obesity	obesity
HP:0001252	muscular hypotonia	muscular hypotonia
HP:0001256	mild mental retardation	intellectual disability, mild