# ATYPICAL PHENOTYPE OF OPITZ GBBB SYNDROME CAUSED BY A NOVEL MID1 VARIATION

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Purpose: Opitz GBBB syndrome is a genetic condition that affects midline structures manly characterized by hypertelorism, cleft lip/palate and hypospadias. MID1 variations have been described as etiology of recessive X-linked form. Here we described a male with atypical phenotype of Opitz GBBB syndrome. Case report: A 20-years-old male, singleton of a nonconsanguineous couple, was born at term through cesarean section after an unremarkable gestation. Presented feeding difficulties and episodes of pneumonia in childhood. Neuromotor development was normal. Physical examination at 15 years old revealed atypical phenotype of Opitz GBBB syndrome consisting of small head, brachycephaly, arched eyebrows, small and low set ears, large neck and low posterior hairline, besides the common signs of the condition (hypertelorism, cleft lip/palate and hypospadias). Molecular analysis of MID1 gene detected 1bp-insertion (c.1637_1638insT) lead to frameshift and create a premature stop codon at the SPRY domain (p.Ile547Hisfs*20). Discussion: The majority of Opitz GBBB-causing variations are clustered at the C-terminus of the MID1 gene corresponding to the coiled-coil region and the B30.2 domain. In previous studies, B30.2 domain was proposed as the major cause of the phenotype, while genotype-phenotype correlation was not found. Conclusion: These atypical features observed in this patient can be due to stochastic events or influenced by other genes, or even expand the phenotype spectrum of Opitz GBBB syndrome.