Silver Russell syndrome in a preterm girl with 8q12.1 deletion encompassing PLAG1.

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Abstract

Silver Russell syndrome (SRS) is a congenital disorder characterised by intrauterine growth retardation (IUGR), feeding difficulties and postnatal growth retardation. In a small number of cases *PLAG1* variants have been described (OMIM #618907). *PLAG1* haploinsufficiency decreases IGF2 expression and produces a Silver Russell syndrome like phenotype. Here, we describe the phenotype and molecular features of a 26 months girl with clinical features of SRS and a de novo 2.1 Mb deletion encompassing PLAG1 is reported in association with clinical features suggestive of SRS.

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