Reduced expression by SETBP1 haploinsufficiency causes developmental and expressive language delay indicating a phenotype distinct from Schinzel-Giedion syndrome

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Mutations of the SET binding protein 1 gene (SETBP1) on 18q12.3 have recently been reported to cause Schinzel-Giedion syndrome (SGS). As rare 18q interstitial deletions affecting multiple genes including SETBP1 correlate with a milder phenotype, including minor physical anomalies and developmental and expressive speech delay, mutations in SETBP1 are thought to result in a gain-of-function or a dominant-negative effect. However, the consequence of the SETBP1 loss-of-function has not yet been well described.
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