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1: [Am J Med Genet.](#) 1999 Nov 19;87(2):139-42.

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Rare interstitial deletion (2)(p11.2p13) in a child with pericentric inversion (2)(p11.2q13) of paternal origin.

[Lacbawan FL](#), [White BJ](#), [Anguiano A](#), [Rigdon DT](#), [Ball KD](#), [Bromage GB](#), [Yang X](#), [DiFazio MP](#), [Levin SW](#).

Medical Genetics Branch, NHGRI, the National Institutes of Health, Bethesda, Maryland, USA.

An unbalanced 46,XY,der(2)del(2)(p11.2p13) inv(2)(p11.2q13) karyotype was found in a phenotypically abnormal child with a de novo interstitial deletion of band 2p12 associated with an inv(2)(p11.2q13) inherited from the father. The inv(2) is generally considered a benign familial variant without significant reproductive consequences. However, our findings led us to consider a previously proposed mechanism of unequal meiotic crossing over at the base of a parental inversion loop, which could lead to either a deletion or duplication of a segment adjacent to the inverted region in the offspring. This phenomenon has been reported in other inversions of chromosomes 1, 7, 13, 15, and 17 and may explain the origin of the deletion in our patient. Although repetitive sequences might be present around such inversions, which could predispose to de novo deletions independently of the inversion, current evidence including this case favors a proposed causal relationship between the parental inversion and the deletion in the child. Our review and results suggest there could be a small risk for a related imbalance to couples with an inv(2)(p11.2q13). For del(2)(p11.2p13), which is rare, a more distinct phenotype has been proposed herein. Our patient shared several findings with the three previously published cases, namely the broad nasal bridge, abnormal ears, high-arched palate, psychomotor retardation, and micrognathia. However, our patient also had sensorineural hearing loss and significant hypotonia, which have not been previously reported, thereby expanding our understanding of this rare deletion. *Am. J. Med. Genet.* 87:139-142, 1999. Published 1999 Wiley-Liss, Inc.

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