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1: [Am J Med Genet.](#) 2001 Nov 1;103(4):320-5.

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Molecularly defined interstitial tandem duplication 6p case with mild manifestations.

[Ng D](#), [Mowrey P](#), [Ragoussis J](#), [Mirza G](#), [Coll E](#), [Di Fazio MP](#), [Turner C](#), [Levin SW](#).

Heritable Disorders Branch, NICHD, National Institutes of Health, Bethesda, Maryland, USA.

An interstitial tandem duplication of 6p21.1-p22.2 was found in a girl at 11 months of age when she was evaluated for developmental delay. Previous cases reported with partial 6p duplication usually have involved terminal duplications, with breakpoints ranging from 6p11 to 6p25. Our patient exhibits a milder phenotype compared to the previously reported cases in the literature. Features that she has in common with the other cases include craniofacial anomalies, such as broad nasal bridge and bulbous tip, thin lips, incomplete development of the scapha helix bilaterally, mild spastic paraparesis of the lower extremities, gross motor delay, and mild cognitive delays. Copyright 2001 Wiley-Liss, Inc.

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