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A 669Kb deletion in 17q23.2, encompassing *TBX2* and *TBX4* genes, in a girl with a moderate developmental delay without any other pertinent abnormality

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Microdeletion of the 17q23.1-q23.2 region recently emerged as a syndrome (OMIM#613355) based in a small number of cases with a common phenotype including mild-to-moderate developmental delay, heart defects, microcephaly, postnatal grow retardation, and hand, foot, and limb abnormalities. All patients reported to date present mild to moderate developmental delay, in particular speech delay, and half of them hearing loss.

The smallest overlapping region has approximately 2.2 Mb and includes the transcription factors *TBX2* and *TBX4* genes. These genes have been implicated in a number of developmental pathways, including those of the heart and limbs. The *TBX4* gene is also associated with the autosomal dominant small patella syndrome (SPS, OMIM 147891).

Here we report a 8 year-old girl with moderate developmental delay including learning disabilities. The test for Fragile X syndrome indicated an allele within the grey area (number of repeats ~50 CGG) inherited from her mother and probably not relevant.

Affymetrix Cytoscan HD chromosome microarray analysis was performed and a 669 Kb interstitial deletion was detected at 17q23.2 region, encompassing only five OMIM genes: *BCAS3*, *TBX2*, *TBX4*, *NACA2* and *BRIP1*. To our knowledge this is the smallest deletion described in this region. None of the genes present in the deleted region are known to be associated with developmental problems.

We compare our patient with the other similar reported cases, in order to add some increased value to the phenotype-genotype correlation of deletions in this region.