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Mechanisms of Disease

## Role of *TBX1* in human del22q11.2 syndrome

Hisato Yagi MS <sup>a</sup> ... Dr Rumiko Matsuoka MD <sup>a, b</sup>

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### Summary

#### Background

Del22q11.2 syndrome is the most frequent known chromosomal microdeletion syndrome, with an incidence of 1 in 4000–5000 livebirths. It is characterised by a 3-Mb deletion on chromosome 22q11.2, cardiac abnormalities, T-cell deficits, cleft palate facial anomalies, and hypocalcaemia. At least 30 genes have been mapped to the deleted region. However, the association of these genes with the cause of this syndrome is not clearly understood.

#### Methods

To test for the chromosomal deletion at 22q11.2, we did fluorescence in-situ hybridisation analysis with ten probes on 22q11.2 in 235 unrelated patients with clinically diagnosed del22q11.2 syndrome. To investigate mutations in the coding sequence of *TBX1*, we did direct sequencing in 12 patients from the same cohort. In all patients,

*TBX1*, we also did genetic analysis in 13 patients from ten families who have the 22q11.2 syndrome phenotype but no detectable deletion of 22q11.2.

## Findings

96% (225 of 235) of patients had a defined 1.5–3-Mb deletion at 22q11.2. We identified three mutations of *TBX1* in two unrelated patients without the 22q11.2 deletion—one with sporadic conotruncal anomaly face syndrome/velocardiofacial syndrome and one with sporadic DiGeorge's syndrome—and in three patients from a family with conotruncal anomaly face syndrome/velocardiofacial syndrome. We did not record these three mutations in 555 healthy controls (1110 chromosomes;  $p < 0.0001$ ).

## Interpretation

Our results suggest that the *TBX1* mutation is responsible for five major phenotypes in del22q11.2 syndrome. Therefore, we conclude that *TBX1* is a major genetic determinant of the del22q11.2 syndrome.



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