

ABSTRACT

Non-syndromic cleft palate (CP) is a complex disorder with variable phenotypes, largely attributed to the interactions of multiple genes and environment. Numerous genes have been reported to be involved in disorder and each gene potentially having certain effects on the phenotypes. One of them, *TBX22* gene located on X chromosome was studied in the current work. To investigate whether mutations in *TBX22* play a part in the development of non-syndromic CP in the Pakistani population, in the current study we performed mutation analysis in coding region of four exons (Exon1, Exon3, Exon4, and Exon8) on the basis of study performed in Thai population (Suphapeetiporn *et al.*, 2007). Four novel mutations were identified in four different Thai patients. In the current investigation, we included 8 unrelated Pakistani patients with CP and 2 unaffected normal subjects to find out the above said mutations from Thai population. We identified two novel mutations in two unrelated patients. All mutations were not detected in normal subjects. One of the mutations was a heterozygous condition at 673 C>A Exon 4 found in two different patients (Patient ID: D1b and 3DE), leading to a non-conservative amino acid substitution from proline to glutamine (P180→Q). Mutation 1117 G>T Exon 8 found in same patient ID 3DE, involving non-conservative amino acid substitution from serine to isoleucine (S328I). Our study indicates that *TBX22* mutations might be responsible for the non-syndromic CP in Pakistani population. Further study is needed to include other genes studied in different populations in the world responsible for non-syndromic CP.