Preface

Congenital limb malformations are among the most frequent congenital malformations in humans caused by genetic mutations or teratogenic effects resulting either in abnormal, loss of, or additional skeletal elements with a frequency of about 1 in 500 live births for upper limbs. Up to 18% of all children with a congenital limb malformation die before the age of 6 years, usually because of associated, more serious organ malformations and/or dysfunctions. Despite this wide prevalence of limb abnormalities, to date only, 84 genes have been associated with syndromes that include limb defects, 15 of which have described polydactyly. Present study aimed to identify genetic cause underlying congenital limb malformations in Indian population. In this study syndromic and non-syndromic polydactyly, syndactyly and Split-hand foot malformation (SHFM) were mainly focused. Involvement of GLI3 in syndromic and non-syndromic polydactyly, HOXD13 and GJA1 in syndromic and non-syndromic syndactyly and TP63 in limb malformation especially SHFM with/without oral cleft were explored. Cytogenetic analysis of familial SHFM and sporadic frontonasal dysplasia with limb malformation was also studied to understand the contribution of chromosomal imbalance toward congenital limb malformation. In order to identify gene expression profiling during human limb development whole transcriptome analysis of human limb bud from aborted embryo was also studied.