



## Analysis of *GATA1* mutations and leukemogenesis in newborns with Down syndrome

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**ABSTRACT.** It has been reported that patients with Down syndrome (DS) frequently develop transient myeloproliferative disorder (TMD) and less commonly myeloid leukemia in DS (ML-DS). We examined the pathogenetic relationship of these conditions with somatic mutations of the *GATA1* gene in children with both TMD and ML-DS. To determine the incidence of *GATA1* mutations in a cohort of DS patients and the applicability of these mutations as a clonal marker to detect minimal residual disease, we screened 198 samples of 169 patients with DS for mutations in *GATA1* exon 2 by direct sequencing. Novel mutations were detected in four of the 169 DS patients (2 with TMD and 2 with ML-DS). We examined spontaneous remission and response to therapy in TMD and ML-DS patients and concluded that

these mutations can be used as stable markers in PCR analysis to monitor these events.

**Key words:** Down syndrome; Transient myeloproliferative disorder; *GATA1* mutation; Myeloid leukemia in DS; Minimal residual disease