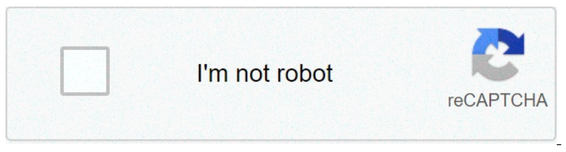

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Bruton Tyrosine kinase, genotype-phenotype correlation, X-Linked Agammaglobulinemia Priority: X-Linked Agamaglobulinemia (XLA) is characterized by absent or strongly reduced, low or detectable immunoglobulin levels, and clinically from extracellular bacterial infections that compromise mainly The respiratory tract. We aimed to analyze the clinical, immunological and genetic characteristics of 22 male children with XLA. Methods: Twenty-two children with XLA from 12 unrelated families were enrolled in this study. The clinical and demographic characteristics of patients, serum immunoglobulin levels, the percentage of B cells and btk gene mutations were examined retrospectively. 12 different mutations were identified in 22 patients from 12 non-confined families. The most frequent type of mutation was premature stop codon (33.3%). Ten mutations had been previously reported including three Missense mutations (C.1774T> C, C.1684C> T, C.83g> T), three premature stop codons (C.1558C> T, C.1573C> T, C.753g> a), two splice-site (C.683-1G> A, C.1567-12 1567-9Delttg) and two small nucleotide deletions (C.902-904 Delaag, C.179 181Delaga). Two new btk gene mutations were also presented and included a splice-site mutation (C.391 + 1G> C) and a premature mutation stop code (C.1243 1243Delg). You are on 12 mutations of the BTK gene they were in the SH1 domain, two in the pH domain, two in the SH3 domain and two in the SH2 domain. Three patients had a story of serious infection before the diagnosis. We do not identify a correlation between the gravity of clinical symptoms and genotype. Conclusions: Our results show that the mutations in the south-east of Turkey could be different from those of the rest of the world and molecular genetic tests are an important tool for the premature confirmed diagnosis of XLA. 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