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able to kill CD48+ lymphomas expressing sufficient amounts of the ligands of these activating receptors.

Conclusions: This study sheds new light on the XLP1 immunological defect and on the crosstalk of inhibitory 2B4 with triggering NK receptors.

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A new IL-2RG gene mutation in an x-linked SCID identified through TREC/KREC screening: a case report

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Severe combined immunodeficiencies (SCID) represent a rare group of primary immunodeficiency disorders (PIDs), which will be fatal in early childhood due to development of common and opportunistic infections. Owing to a high number of unknown mutations in related genes especially in the exons of IL-2RG gene, new cases are intermittently reported around the world. Here, we found a new mutation of the common gamma chain in an Iranian X-SCID newborn.

The patient was a 6-day old boy with a family history of PID who was referred to the Immunology, Asthma and Allergy Research Institute (IAARI) for immunological evaluation. The child was screened using a molecular-based analysis that measures the number of copies of T cell receptor excision circles (TRECs) and kappa-deleting recombination excision circles (KRECs) using a multiplex quantitative Real Time PCR. To confirm the results, a complete immunological evaluation and IL2RG (interleukin 2 receptor, gamma) gene sequencing were performed (both molecular assessment and gene sequencing were done in Leipzig, Germany).

The results of the multiplex quantitative real time PCR showed undetectable TREC but a high level of KREC copy numbers. Flow cytometric analysis of lymphocyte subsets also showed low numbers of T and NK cells, but elevated number of B cells. We subsequently found a novel substitution in the IL2RG encoding gene (X-SCID): c.675 C>A, leading to p.225 Ser>Arg. Based on the functional analysis using PolyPhen, the mutation is predicted to be damaging. The patient was thus diagnosed as a T- B+ NK- X-linked SCID.

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