A novel deletion mutation in IL2RG gene results in X-linked severe combined immunodeficiency with an atypical phenotype

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#	Paper	IF	Citations
13	Family History of Early Infant Death Correlates with Earlier Age at Diagnosis But Not Shorter Time to Diagnosis for Severe Combined Immunodeficiency. *Frontiers in Immunology*, **2017**, 8, 808	8.4	21
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11	The IL-2RG R328X nonsense mutation allows partial STAT-5 phosphorylation and defines a critical region involved in the leaky-SCID phenotype. *Clinical and Experimental Immunology*, **2020**, 200, 61-72	6.2	1
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6	Case Report: A Novel IL2RG Frame-Restoring Rescue Mutation Mimics Early T Cell Engraftment Following Haploidentical Hematopoietic Stem Cell Transplantation in a Patient With X-SCID. *Frontiers in Immunology*, **2021**, 12, 644687	8.4	
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3	Gene analysis of seven cases of primary immunodeficiency. *Translational Pediatrics*, **2020**, 9, 117-125	4.2	1
2	Induced Pluripotent Stem Cell Meets Severe Combined Immunodeficiency. *Cell Journal*, **2020**, 22, 1-10	2.4	3
1	Somatic Reversion of a Novel Mutation Resulting in Atypical X-Linked Combined Immunodeficiency.. *Genes*, **2021**, 13,	4.2	0