Resource Summary Report

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BTKbase

RRID:SCR_013101
Type: Tool

Proper Citation

BTKbase (RRID:SCR_013101)

Resource Information

URL: http://bioinf.uta.fi/BTKbase/

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Description: A mutation registry for X-linked agammaglobulinemia (XLA). BTKbase lists mutation entries of 1,111 patients from 973 unrelated families showing 602 unique molecular events. Agammaglobulinemia is characterized by failure to produce mature B lymphocyte cells and is associated with a failure of Ig heavy chain rearrangement. Two thirds of cases are familial, and one third of cases are believed to arise from new mutations. Mutations of the BTK gene are found in approximately 80% of patients with agammaglobulinemia. The localization of the mutations on the gene and protein for BTK can be analyzed by clicking sequences on the web pages. It includes a mutation browser, which gives users access to mutations in Bruton tyrosine kinase (BTK) protein sequences, and XLA fact file, and forms for users to submit mutation to the dataset.

Synonyms: BTKbase

Resource Type: data or information resource, database

Keywords: bruton tyrosine kinase, xla, x-linked agammaglobulinemia

Related Condition: Agammaglobulinemia

Resource Name: BTKbase

Resource ID: SCR_013101

Alternate IDs: nif-0000-02625

Record Creation Time: 20220129T080314+0000

Record Last Update: 20260904T000341+0000

Ratings and Alerts

No rating or validation information has been found for BTKbase.

No alerts have been found for BTKbase.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 14 mentions in open access literature.

Listed below are recent publications. The full list is available at kravitz2 .

Kraft MT, et al. (2021) Identification of 22 novel BTK gene variants in B cell deficiency with hypogammaglobulinemia. Clinical immunology (Orlando, Fla.), 229, 108788.

Mohammad T, et al. (2020) Impact of amino acid substitution in the kinase domain of Bruton tyrosine kinase and its association with X-linked agammaglobulinemia. International journal of biological macromolecules, 164, 2399.

Wist M, et al. (2020) Noncatalytic Bruton's tyrosine kinase activates PLC?2 variants mediating ibrutinib resistance in human chronic lymphocytic leukemia cells. The Journal of biological chemistry, 295(17), 5717.

Chen XF, et al. (2016) Clinical characteristics and genetic profiles of 174 patients with X-linked agammaglobulinemia: Report from Shanghai, China (2000-2015). Medicine, 95(32), e4544.

Teocchi MA, et al. (2015) BTK mutations selectively regulate BTK expression and upregulate monocyte XBP1 mRNA in XLA patients. Immunity, inflammation and disease, 3(3), 171.

Boushaki S, et al. (2015) Prevalence of BTK mutations in male Algerian patterns with agammaglobulinemia and severe B cell lymphopenia. Clinical immunology (Orlando, Fla.), 161(2), 286.

Debost-Legrand A, et al. (2013) A new mutation that predicted a drastic alteration of the BTK protein function. Gene, 527(1), 426.

Lim LM, et al. (2013) Atypical X-linked agammaglobulinaemia caused by a novel BTK mutation in a selective immunoglobulin M deficiency patient. BMC pediatrics, 13, 150.

Thompson ER, et al. (2012) Exome sequencing identifies rare deleterious mutations in DNA repair genes FANCC and BLM as potential breast cancer susceptibility alleles. PLoS genetics, 8(9), e1002894.

Küntzer J, et al. (2010) Human variation databases. Database : the journal of biological databases and curation, 2010, baq015.

Tóth B, et al. (2009) Genetic and demographic features of X-linked agammaglobulinemia in Eastern and Central Europe: a cohort study. Molecular immunology, 46(10), 2140.

Lopez-Herrera G, et al. (2008) Characterization of Bruton's tyrosine kinase mutations in Mexican patients with X-linked agammaglobulinemia. Molecular immunology, 45(4), 1094.

Kristufek D, et al. (2007) Characterization of novel Bruton's tyrosine kinase gene mutations in Central European patients with agammaglobulinemia. Molecular immunology, 44(7), 1639.

Galperin MY, et al. (2005) The Molecular Biology Database Collection: 2005 update. Nucleic acids research, 33(Database issue), D5.