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10

Name of Journal: *World Journal of Clinical Cases***Manuscript NO:** 54880**Manuscript Type:** CASE REPORT

Novel deletion mutation in Burton's tyrosine kinase results in X-linked agammaglobulinemia: A case report

Xiao-Mei Hu, Ke Yuan, Hong Chen, Chun Chen, Yan-Lan Fang, Jian-Fang Zhu, Li Liang, Chun-Lin Wang

Abstract

BACKGROUND

X-linked agammaglobulinemia is a primary immunodeficiency disease caused by mutations of the Burton's tyrosine kinase (*BTK*) gene. We found a

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Sep 27, 2013 · **Mutations** in the **gene** coding for Bruton's tyrosine kinase (BTK) have been identified as the cause of XLA. Most affected patients exhibit a marked reduction of **serum immunoglobulins**, mature B cells, and an increased susceptibility to recurrent **bacterial infections**.

Cited by: 6

Author: Lee-Moay Lim, Jer-Ming Chang, I-Fang W...

Publish Year: 2013

[PDF] CASE REPORT Open Access Atypical X-linked ...

<https://bmcpediatr.biomedcentral.com/track/pdf/10.1186/1471-2431-13-150?site=...>

CASE REPORT Open Access Atypical X-linked agammaglobulinaemia caused by a **novel BTK mutation** in a selective immunoglobulin M deficiency patient Lee-Moay Lim¹, Jer-Ming Chang^{2,6}, I-Fang Wang³, Wei-Chiao Chang^{4,5}, Daw-Yang Hwang^{1*} and Hung-Chun Chen^{1,6} Abstract

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Author: Lee-Moay Lim, Jer-Ming Chang, I-Fang W...

Publish Year: 2013

Delayed diagnosis of X-linked agammaglobulinaemia in a ...

<https://bmcneurol.biomedcentral.com/articles/10.1186/s12883-019-1536-7> ▾

Dec 12, 2019 · X-linked **agammaglobulinemia** (XLA) is an immunodeficiency disease caused by **mutations** in the **gene** coding for BTK, leading to failure to produce mature B lymphocytes []. Patients with XLA are subject to recurrent severe bacterial infections from early age and severely reduced B cell and immunoglobulin levels.

Author: Ya-Ni Zhang, Yuan-Yuan Gao, Si-Da Y... Publish Year: 2019

A novel BTK gene mutation creates a de-novo splice site in ...

<https://www.sciencedirect.com/science/article/pii/S0378111915001547>

Apr 15, 2015 · Bruton's tyrosine kinase (BTK), encoded by the BTK gene, is a cytoplasmic protein critical in B cell development. **Mutations** in the BTK gene cause X-linked agammaglobulinemia (XLA), a primary immunodeficiency with characteristically low or absent B cells and antibodies.

Cited by: 1

Author: Chai Teng Chear, Adiratna Mat Ripen, Sh...

Publish Year: 2015

Analysis of Btk Mutations in Patients with X-Linked ...

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Jun 10, 2014 · Discovery of **Bruton's tyrosine kinase** (BTK) **mutations** as the cause for **X-linked** agammaglobulinemia was a milestone in understanding the genetic basis of primary immunodeficiencies. Since then, studies have highlighted the critical role of this enzyme in **B-cell development** and function, and particularly in **B-cell receptor signaling**.

Cited by: 80

Author: Sabine Ponader, Jan A. Burger

Publish Year: 2014

Metastatic colorectal cancer and severe hypocalcemia ...

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6739925>

X-linked agammaglobulinemia (XLA) is a primary immunodeficiency disorder caused by **germline mutations** in the **Bruton tyrosine kinase** (BTK) gene on **X** chromosome. These **mutations** disturb **B-cell development**, decrease immunoglobulin levels, increase susceptibility to infection or neoplasms, and increase the risk of developing **colorectal cancer** (CRC).

Cited by: 2

Author: Mingming Li, Wei Chen, Xiaomeng Sun, ...

Publish Year: 2019

Naturally occurring Bruton's tyrosine kinase mutations ...

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Cited by: 2

Author: Mingming Li, Wei Chen, Xiaomeng Sun, ...

Publish Year: 2019

Identification of mutations in the Bruton's tyrosine ...

<https://www.nature.com/articles/jhg200356>

May 24, 2003 · **Mutations** in the **Bruton's tyrosine kinase** (BTK) **gene** are responsible for **X-linked** agammaglobulinemia (XLA). We identified **BTK mutations in six patients** with presumed XLA from unrelated Korean...

Cited by: 9

Author: Eun Kyeong Jo, Yue Wang, Hirokazu Kan...

Publish Year: 2003

Agammaglobulinemia

Medical Condition



A primary immunodeficiency disorder that presents with reduced or lack of gamma globulin proteins.

📅 Very rare (Fewer than 20,000 cases per year in US)

🧪 Requires lab test or imaging

👨‍⚕️ Treatment from medical professional advised

🕒 Can be lifelong

The main cause is the mutation of Bruton's tyrosine kinase (Btk) gene on the X-chromosome leading to blockage of B cell development. Characterized by recurrent respiratory and enteroviral infections. Treatment is by the administration of pooled gamma globulin every 3-4 weeks.

Symptoms