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Observational Study

Potential protein–phenotype correlation in 3 LRBA-deficient patients

Wen-Juan Tang, Wen-Hui Hu, Ying Huang, Bing-Bing Wu, Xiao-Min Peng, Xiao-Wen Zhai, Xiao-Wen Qian, Zi-Qing Ye, Hai-Jiao Xia, Jie Wu, Jie-Ru Shi

Abstract

BACKGROUND

Patients with LPS-responsive beige-like anchor protein (LRBA) deficiency have a

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However, CD4+ CD25+ FoxP3+ CD127- cells were significantly decreased in **LRBA-deficient patients** compared with those of HCs, particularly in **patients** with autoimmunity. There was a negative **correlation** between the frequencies of CD4+ CD25+ FoxP3+ CD127- cells and Th1-like Th17 cells in **LRBA-deficient patients**, and an overlapping phenotype of ...

Cited by: 10 **Author:** Gholamreza Azizi, Gholamreza Azizi, Abba...
Publish Year: 2018

Atypical Manifestation of LRBA Deficiency with ... 3 mins read

https://academic.oup.com/ibdjournal/article/21/1/40/4604228

Dec 04, 2014 · **LRBA-deficient patients** present with heterogeneity of clinical symptoms with no clear genotype to phenotype **correlation**. Common features of **LRBA-deficient patients** are quantitative and/or qualitative B-cell defects as well as autoimmunity. 26 , – 28 Nine of 11 published **patients** to date present with autoimmune IBD-like manifestations. 26 , 27 ...

Cited by: 72 **Author:** Nina Kathrin Serwas, Aydan Kansu, Elisang...
Publish Year: 2015

[PDF] Novel compound heterozygous stop-gain mutations of LRBA ...

https://onlinelibrary.wiley.com/doi/pdf/10.1002/mgg3.1216

et al., 2012). **LRBA deficiency** is a rare autosomal recessive disorder caused by biallelic mutations in the

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LPS-responsive beige-like anchor (LRBA) deficiency is an autosomal recessive primary immunodeficiency disorder, OMIM (#614700). LRBA deficiency **patients** suffer from variable…

Cited by: 1 **Author:** Safa Meshaal, Rabab El Hawary, Rana A…

Publish Year: 2020

The extended phenotype of LPS-responsive beige-like ...

https://www.sciencedirect.com/science/article/pii/S0091674915013652

Jan 01, 2016 · This diagnostic protocol allows us to classify our **patients** in **3** different groups: (1) possible LRBA deficiency, including all **patients** with the clinical suspicion of LRBA deficiency from…

Cited by: 196 **Author:** Laura Gámez-Díaz, Dietrich August, Poli…

Publish Year: 2016

LRBA deficiency

Rare Genetic Disorder

LRBA deficiency is a rare genetic disorder of the immune system. This disorder is caused by a mutation in the gene LRBA. LRBA stands for “lipopolysaccharide-responsive and beige-like anchor protein”. This condition is characterized by autoimmunity, lymphoproliferation, and immune deficiency. It was first described by Gabriela Lopez-Herrera from University College London in 2012. Investigators in the laboratory of Dr. Michael Lenardo at National Institute of Allergy and Infectious Diseases, the National Institutes of Health

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https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5022363

Sep 14, 2016 · Keywords: autoimmune thyroiditis, lipopolysaccharide responsive beige-like **anchor** gene, autoimmune enteropathy, stem cell transplantation, genotype–phenotype **correlation** Introduction A...

Cited by: 13 **Author:** Shahrzad Bakhtiar, Frank Ruemmele, Fabie...
Publish Year: 2016

The extended phenotype of LPS-responsive beige-like anchor ...

https://www.sciencedirect.com/science/article/pii/S0091674915013652

Jan 01, 2016 · LPS-responsive beige-like **anchor** protein (LRBA) is a member of the PH-BEACH-WD40 (pleckstrin homology-beige and Chediak-Higashi-tryptophan aspartic acid dipeptide) protein family, whic...

Cited by: 196 **Author:** Laura Gámez-Díaz, Dietrich August, Polina ...
Publish Year: 2016

The extended phenotype of LPS-responsive beige-like anchor ...

https://www.sciencedirect.com/science/article/abs/pii/S0091674915013652

LPS-responsive beige-like **anchor** protein **(LRBA)** deficiency is a primary immunodeficiency caused by biallelic mutations in LRBA that abolish LRBA protein expression. Objective We sought to report the...

Cited by: 196 **Author:** Laura Gámez-Díaz, Dietrich August, Polina ...
Publish Year: 2016

[PDF] Atypical Manifestation of LPS-Responsive Beige-Like Anchor ...

https://core.ac.uk/download/pdf/82835349.pdf

affected **patients**. Keywords: autoimmune thyroiditis, lipopolysaccharide responsive beige-like **anchor** gene, autoimmune enteropathy, stem cell transplantation, genotype–phenotype **correlation**....

987 - Gene ResultLRBA LPS responsive beige-like anchor ...

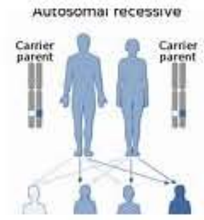
https://www.ncbi.nlm.nih.gov/gene/987

May 09, 2021 · Case Report: **potential** causative role of LRBA gene mutations in juvenile arthritis. Among 2 brothers homozygous for LPS responsive beige-like **anchor** protein (LRBA) mutation, one developed...

Clinical, Immunologic, and Molecular Spectrum of Patients ...

LPS-responsive beige-like anchor protein deficiency

Rare Genetic Condition



LPS-responsive beige-like anchor protein deficiency is a rare genetic condition caused by the absence of LPS-responsive beige-like anchor protein.

Wikipedia

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LRBA deficiency (Rare Genetic Disorder)

LRBA deficiency is a rare genetic disorder of the immune system. This disorder is caused by a mutation in the gene LRBA. LRBA stands for "lipo...