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[https://www.cell.com/ajhg/fulltext/S0002-9297\(14\)00480-7](https://www.cell.com/ajhg/fulltext/S0002-9297(14)00480-7) ▾

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Publish Year: 2015

[PDF] Defects of CRB2 Cause Steroid-Resistant Nephrotic ...

<https://core.ac.uk/download/pdf/82829487.pdf>

REPORT Defects of CRB2 Cause Steroid-Resistant Nephrotic Syndrome Lwaki Ebarasi,1 ,2 12Shazia Ashraf,3 Agnieszka Bierzynska,4 Heon Yung Gee,3 Hugh J. McCarthy,4 Sijetlana Lovric, 3Carolyn E. Sadowski, Werner Pabst, 3Virginia Vega-Warner,5 Humphrey Fang, Ania Koziell,6 Michael A. Simpson,7 Ismail Dursun,8 Erkin Serdaroglu,9 Shawn Levy,10 Moin A. Saleem,4 Friedhelm Hildebrandt,3 ,11 ...

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Recent advances in high-throughput sequencing for clinical genetic testing have revealed novel disease-causing genes, such as Crumbs homolog 2 (CRB2) for early-onset steroid-resistant nephrotic syndrome (SRNS). We report the long-term clinicopathologic observation of a Japanese female patient with SRNS caused by a newly identified compound heterozygous mutation of CRB2 (p.Arg628Cys and ...
Author: Shojiro Watanabe, Tomomi Aizawa, H... Publish Year: 2018

CRB2 crumbs cell polarity complex component 2 [(human)]

<https://www.ncbi.nlm.nih.gov/gene/286204>

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(PDF) A case report of CRB2 mutation identified in a ...

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Recessive CRB2 mutations were recently reported to cause both steroid resistant nephrotic syndrome and prenatal onset ventriculomegaly with kidney disease. We report two ...

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CRB2 mutation in two siblings with steroid-resistant nephrotic syndrome: Report of two cases and literature review

Jing Lu, Yan-Nan Guo, Li-Qun Dong

Abstract

BACKGROUND

CRB2 is a recently discovered gene that is closely related to the maintenance of the normal polarity of podocytes, and its mutation can directly lead to steroid-resistant nephrotic syndrome (SRNS). However, there is a lack of a summary of the characteristics of NS caused by these CRB2 mutations.

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<https://www.ncbi.nlm.nih.gov/pubmed/29473663>

Recent advances in high-throughput sequencing for clinical genetic testing have revealed novel disease-causing genes, such as **Crumbs homolog 2 (CRB2)** for early-onset steroid-resistant nephrotic syndrome (SRNS). We report the long-term clinicopathologic observation of a Japanese female patient with SRNS caused by a newly identified compound heterozygous **mutation** of CRB2 (p.Arg628Cys and ...

Author: Shojiro Watanabe, Tomomi Aizawa, H... Publish Year: 2018

CRB2 crumbs cell polarity complex component 2 [(human)]

<https://www.ncbi.nlm.nih.gov/gene/286204>

Dec 13, 2020 · Case Report: steroid-resistant **nephrotic syndrome** caused by a novel **Crumbs homolog 2 mutation**. These findings demonstrate that Crb2 abnormalities caused by these **mutations** are the mechanism of steroid-resistant **nephrotic syndrome**; Clinically, CRB2 should be assessed when ciliopathy is suspected, especially in Ashkenazi Jews, where we found that ...

Expansion of Phenotype and Genotypic Data in CRB2-related ...

<https://pubmed.ncbi.nlm.nih.gov/27004616>

All reported patients have been homozygotes or compound heterozygotes for sequence variants in the **Crumbs, Drosophila, Homolog of, 2 (CRB2)** genes. Variants affecting CRB2 function have also been identified in four families with **steroid resistant nephrotic syndrome**, but without any other known systemic findings.

Cited by: 19 Author: Ryan E Lamont, Wen-Hann Tan, A Miche...

Publish Year: 2016

Long-term clinicopathologic observation in a case of ...

<https://onlinelibrary.wiley.com/doi/abs/10.1111/nep.13244>

Recent advances in high-throughput sequencing for clinical genetic testing have revealed novel disease-causing genes, such as **Crumbs homolog 2 (CRB2)** for early-onset **steroid-resistant nephrotic syndrome (SRNS)**. We report the long-term clinicopathologic observation of a Japanese female patient with SRNS caused by a newly identified compound heterozygous **mutation** of CRB2 ...

Author: Shojiro Watanabe, Tomomi Aizawa, H... Publish Year: 2018

Expanding the phenotype of CRB2 mutations - A new ...

<https://www.researchprotocols.org/2022/2/e3010>