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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) ▾

Nephrotic syndrome (NS), the association of gross proteinuria, hypoalbuminaemia, edema, and hyperlipidemia, can be clinically divided into steroid-sensitive (SSNS) and steroid-resistant (SRNS) forms. SRNS regularly progresses to end-stage renal failure. By homozygosity mapping and whole exome sequencing, we here identify recessive mutations in Crumbs homolog 2 (CRB2) in four different ...

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Publish Year: 2015

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REPORT Defects of CRB2 Cause Steroid-Resistant Nephrotic Syndrome Lwaki Ebarasi,1,2,12 Shazia 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

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[\(PDF\) A case report of CRB2 mutation identified in a ...](#)<https://www.researchgate.net/publication/327669029>

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

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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>

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

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