

Accepted Manuscript

Male X-chromosome Mosaicism leading to Carrier Phenotype and inheritance of Chronic Granulomatous Disease

Stephanie Harris, MD, Helen Braggins, Karin van Leeuwen, MSc, Kimberly Gilmour, PhD, Matthew S. Buckland, MD PhD, Dirk Roos, PhD, David M. Lowe, MD PhD

PII: S2213-2198(18)30044-8

DOI: [10.1016/j.jaip.2018.01.017](https://doi.org/10.1016/j.jaip.2018.01.017)

Reference: JAIP 1498

To appear in: *The Journal of Allergy and Clinical Immunology: In Practice*

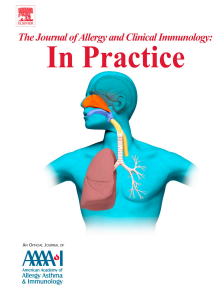
Received Date: 1 December 2017

Revised Date: 21 January 2018

Accepted Date: 26 January 2018

Please cite this article as: Harris S, Braggins H, van Leeuwen K, Gilmour K, Buckland MS, Roos D, Lowe DM, Male X-chromosome Mosaicism leading to Carrier Phenotype and inheritance of Chronic Granulomatous Disease, *The Journal of Allergy and Clinical Immunology: In Practice* (2018), doi: 10.1016/j.jaip.2018.01.017.

This is a PDF file of an unedited manuscript that has been accepted for publication. As a service to our customers we are providing this early version of the manuscript. The manuscript will undergo copyediting, typesetting, and review of the resulting proof before it is published in its final form. Please note that during the production process errors may be discovered which could affect the content, and all legal disclaimers that apply to the journal pertain.



Male X-chromosome Mosaicism leading to Carrier Phenotype and inheritance of Chronic Granulomatous Disease

Stephanie Harris MD¹, Helen Braggins², Karin van Leeuwen MSc³, Kimberly Gilmour PhD², Matthew S Buckland MD PhD^{2,4,5}, Dirk Roos PhD³, David M Lowe MD PhD^{4,5}

¹ Department of Infectious Diseases, Royal Free London NHS Foundation Trust, London, UK

² Department of Immunology, Great Ormond Street Hospital, London, UK

³ Sanquin Research, Amsterdam, the Netherlands

⁴ Institute of Immunity and Transplantation, University College London, London, UK

⁵ Department of Immunology, Royal Free London NHS Foundation Trust, London, UK

Corresponding author: Dr David Lowe
Institute of Immunity and Transplantation
University College London
Royal Free Campus
Pond Street
London
NW3 2QG
United Kingdom

d.lowe@ucl.ac.uk

+44 (0)207 794 0500

Funding: Nil

Word count: 946 words

Clinical implications: We present the first description of a male with X-chromosome mosaicism who has clinical features of the X-linked CGD carrier state and passed a *CYBB* mutation to a daughter and subsequently affected grandson: clinicians should be aware of this possibility.

Conflicts of Interest: The authors declare no conflicts of interest.

Download English Version:

<https://daneshyari.com/en/article/8963786>

Download Persian Version:

<https://daneshyari.com/article/8963786>

[Daneshyari.com](https://daneshyari.com)