

This is a repository copy of *P4HB recurrent missense mutation causing Cole-Carpenter syndrome*.

White Rose Research Online URL for this paper: http://eprints.whiterose.ac.uk/125577/

Version: Supplemental Material

## Article:

Balasubramanian, M. orcid.org/0000-0003-1488-3695, Padidela, R., Pollitt, R.C. orcid.org/0000-0002-8864-397X et al. (6 more authors) (2018) P4HB recurrent missense mutation causing Cole-Carpenter syndrome. Journal of Medical Genetics, 55 (3). pp. 158-165. ISSN 0022-2593

https://doi.org/10.1136/jmedgenet-2017-104899

## Reuse

Items deposited in White Rose Research Online are protected by copyright, with all rights reserved unless indicated otherwise. They may be downloaded and/or printed for private study, or other acts as permitted by national copyright laws. The publisher or other rights holders may allow further reproduction and re-use of the full text version. This is indicated by the licence information on the White Rose Research Online record for the item.

## **Takedown**

If you consider content in White Rose Research Online to be in breach of UK law, please notify us by emailing eprints@whiterose.ac.uk including the URL of the record and the reason for the withdrawal request.



Figure 1a-d



3-years



C



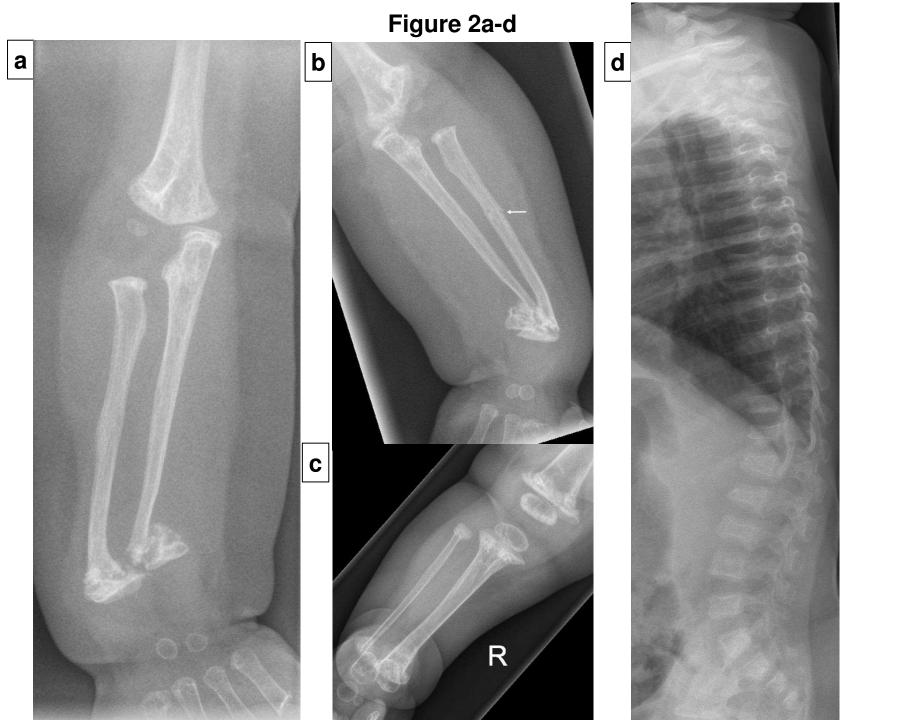


Figure 2e-f

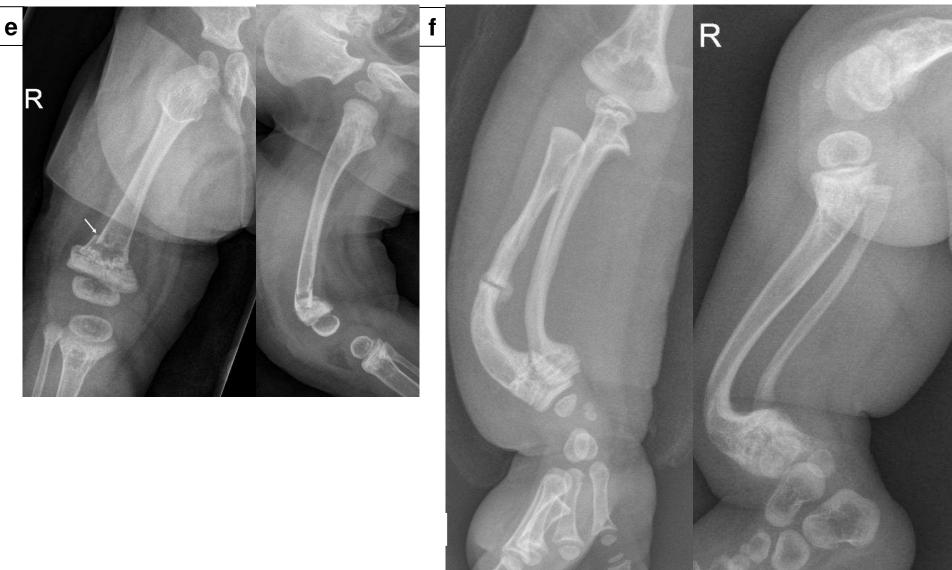
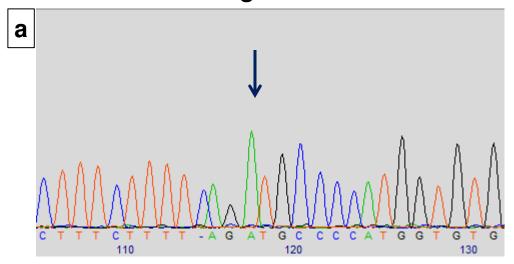


Figure 3



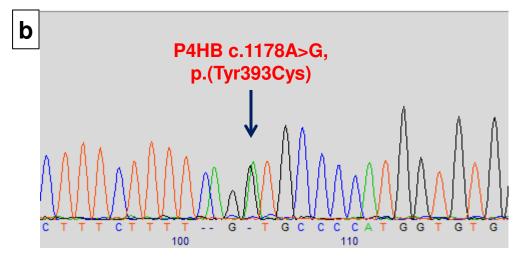
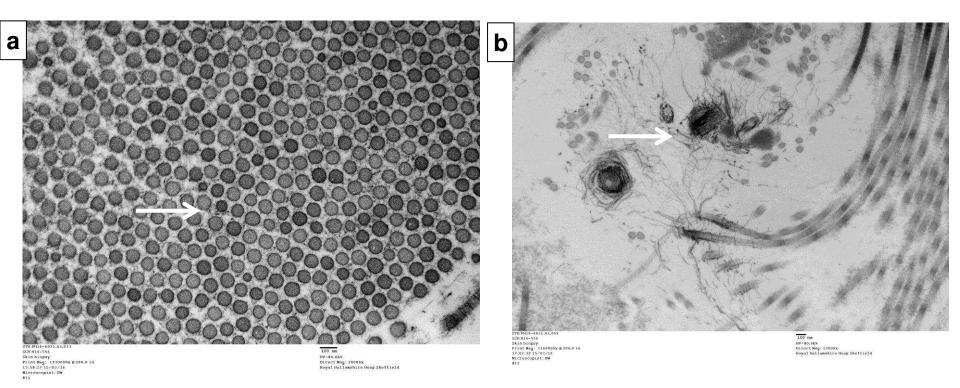
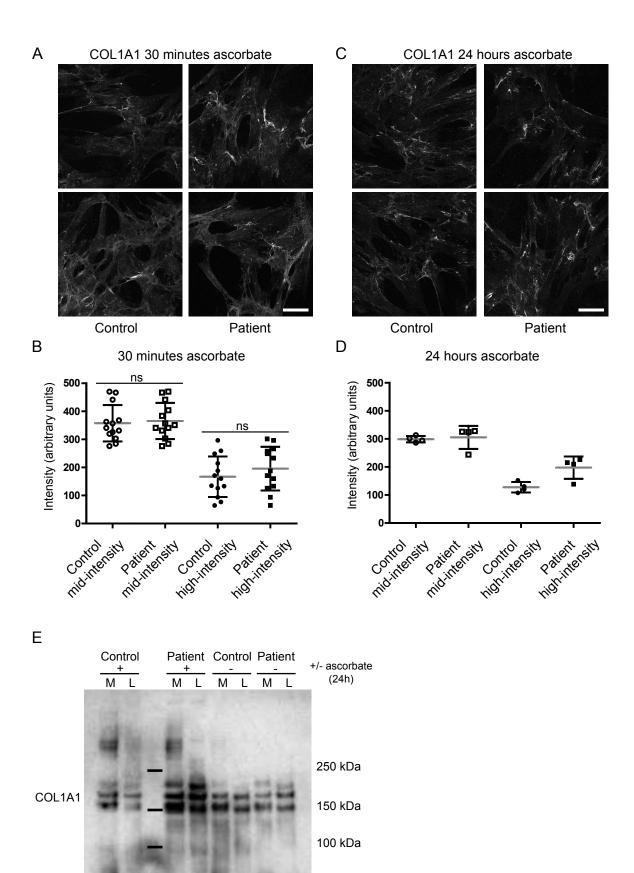


Figure 4





37 kDa

GAPDH