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**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) (2017) P4HB recurrent missense mutation causing Cole-Carpenter syndrome. Journal of Medical Genetics. ISSN 0022-2593

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Figure 1a-d

6-months

3-years
Figure 3

(a) 

(b) P4HB c.1178A>G, p.(Tyr393Cys)
Patient
mid-intensity
Control
mid-intensity
Patient
mid-intensity
Control
mid-intensity
Patient
high-intensity
Control
high-intensity
Control
mid-intensity
Patient
high-intensity

30 minutes ascorbate 24 hours ascorbate

Intensity (arbitrary units)

COL1A1 30 minutes ascorbate
COL1A1 24 hours ascorbate

A
B
C
D
E

GAPDH

COL1A1

250 kDa
150 kDa
100 kDa
37 kDa