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X-linked macrocytic dyserythropoietic anemia in females with an *ALAS2* mutation

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Corrigendum

Original citation: J Clin Invest. 2015;125(4):1665–1669. https://doi.org/10.1172/JCI78619 Citation for this corrigendum: J Clin Invest. 2020;130(1):552. https://doi.org/10.1172/JCI132538 The amino acid substitution for the ALAS2 mutation was incorrectly noted in the original article. The correct designation is ALAS2 Y365H. The correct sentences and figure part are below. Abstract: We determined that this mutation (Y365H) impairs binding of the essential cofactor pyridoxal 5′-phosphate to ALAS2, resulting in destabilization of the enzyme and consequent loss of function. Results and Discussion: This A-to-G variant was found at position 55042086 on the X chromosome (hg19 coordinates), resulting in a coding change of Y365H in the ALAS2 protein (Figure 1F and Supplemental Figure 1). By modeling this novel ALAS2 Y365H mutation in the structure of the Rhodobacter capsulatus homolog, we noted that Y365 fits within a hydrophobic core critical for binding the essential cofactor pyridoxal 5′-phosphate (PLP) (Figure 2A and ref. 16). Together, these findings indicate that the Y365H mutation markedly impairs PLP binding, which may account for some or all of the substantially reduced stability of the ALAS2 enzyme. Figure 2 legend: Severe LOF with the ALAS2 Y365H mutation and lack of highly skewed X inactivation in female mutation carriers. (A) Model of ALAS2 shows PLP highlighted in blue and the Y or H amino acid at position 365 highlighted in red. The authors regret the […]

Retraction

Human α1 type IV collagen NC1 domain exhibits distinct antiangiogenic activity mediated by α1β1 integrin

Akulapalli Sudhakar, Pia Nyberg, Venkateshwar G. Keshamouni, Arjuna P. Mannam, Jian Li, Hikaru Sugimoto, Dominic Cosgrove, and Raghu Kalluri

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The Office of Research Integrity recently notified the *JCI* of falsified data reported in this article. Therefore, the *JCI* is retracting the article.

Corrigendum

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We determined that this mutation (Y365H) impairs binding of the essential cofactor pyridoxal 5′-phosphate to ALAS2, resulting in destabilization of the enzyme and consequent loss of function.

Results and Discussion:
This A-to-G variant was found at position 55042086 on the X chromosome (hg19 coordinates), resulting in a coding change of Y365H in the ALAS2 protein (Figure 1F and Supplemental Figure 1).

By modeling this novel ALAS2 Y365H mutation in the structure of the *Rhodobacter capsulatus* homolog, we noted that Y365 fits within a hydrophobic core critical for binding the essential cofactor pyridoxal 5′-phosphate (PLP) (Figure 2A and ref. 16).

Together, these findings indicate that the Y365H mutation markedly impairs PLP binding, which may account for some or all of the substantially reduced stability of the ALAS2 enzyme.

Figure 2 legend:
Severe LOF with the ALAS2 Y365H mutation and lack of highly skewed X inactivation in female mutation carriers. (**A**) Model of ALAS2 shows PLP highlighted in blue and the Y or H amino acid at position 365 highlighted in red.

The authors regret the errors.