

Corrigendum: genotype–phenotype association analysis reveals new pathogenic facto...

[Health & Medicine](#)



**ASSIGN
BUSTER**

A Corrigendum on

[Genotype-Phenotype Association Analysis Reveals New Pathogenic Factors for Osteogenesis Imperfecta Disease](#)

by Shi J, Ren M, Jia J, Tang M, Guo Y, Ni X and Shi T. (2019). *Front. Pharmacol.* 10: 1200. doi: [10.3389/fphar.2019.01200](https://doi.org/10.3389/fphar.2019.01200)

There is an error in the Funding statement. The correct number for “Beihang University & Capital Medical University Advanced Innovation Center for Big Data-Based Precision Medicine Plan” is “BHME-201804.”

Additionally, in the original article, there was one error. In the Discussion section, the locations of the four candidate pathogenic variations in *COL1A1* were incorrectly mapped to the major ligand-binding region (MLBR3).

A correction has been made to the Discussion, Paragraph five:

“To validate the pathogenicity of the candidate variations in *COL1A1*, we checked the specificity of their locations (positions of the four candidate mutations: 1094 and 1097). Evidence from the protein families database (Pfam) ([El-Gebali et al., 2019](#)) demonstrate that the locations of all four variations belong to the collagen triple helix region (PF01391: Collagen triple helix repeat (1079–1137)). Structurally, different abnormalities in the collagen helix are associated with the identity of the residue replacing Gly ([Bryan et al., 2011](#); [Qiu et al., 2018](#)), which also influence the

severity of OI patients (residues replacing Gly of four candidate
<https://assignbuster.com/corrigendum-genotypephenotype-association-analysis-reveals-new-pathogenic-factors-for-osteogenesis-imperfecta-disease/>

mutations: Asp, Arg, and Ser). Through the statistical analysis on the location of Glysubstitution mutations in a large number of OI patients, Beck et al. found that all Gly→Asp in the $\alpha 1(I)$ chain led to OI type II (perinatal lethal form) ([Beck et al., 2000](#)). In addition, the study of the impact of various Gly replacements discovered that the three replaced form (Gly→Arg, Gly→Ser, and Gly→Cys) had a stronger association with OI lethality than the other replaced forms ([Beck et al., 2000](#)). In all, these conclusions indicate that the four candidate mutations of *COL1A1* we identified are highly likely to cause lethal OI phenotypes.”

Due to the error outlined above, the citations for “ [Di Lullo et al., 2002](#) ” and “ [Xiao et al., 2015](#) ” have been removed from the reference list, and “ [El-Gebali et al., 2019](#) ” has been cited instead.

The authors apologize for these errors and state that these do not change the scientific conclusions of the article in any way. The original article has been updated.

References

Beck, K., Chan, V. C., Shenoy, N., Kirkpatrick, A., Ramshaw, J. A., Brodsky, B. (2000). Destabilization of osteogenesis imperfecta collagen-like model peptides correlates with the identity of the residue replacing glycine. *Proc. Natl. Acad. Sci. U. S. A.* 97 (8), 4273–4278. doi: 10. 1073/pnas. 070050097

[PubMed Abstract](#) | [CrossRef Full Text](#) | [Google Scholar](#)

<https://assignbuster.com/corrigendum-genotypephenotype-association-analysis-reveals-new-pathogenic-factors-for-osteogenesis-imperfecta-disease/>

Bryan, M. A., Cheng, H., Brodsky, B. (2011). Sequence environment of mutation affects stability and folding in collagen model peptides of osteogenesis imperfecta. *Biopolymers* 96 (1), 4–13. doi: 10. 1002/bip. 21432

[PubMed Abstract](#) | [CrossRef Full Text](#) | [Google Scholar](#)

Di Lullo, G. A., Sweeney, S. M., Korkko, J., Ala-Kokko, L., San Antonio, J. D. (2002). Mapping the ligand-binding sites and disease-associated mutations on the most abundant protein in the human, type I collagen. *J. Biol. Chem.* 277 (6), 4223–4231. doi: 10. 1074/jbc. M110709200

[PubMed Abstract](#) | [CrossRef Full Text](#) | [Google Scholar](#)

El-Gebali, S., Mistry, J., Bateman, A., Eddy, S. R., Luciani, A., Potter, S. C., et al. (2019). The Pfam protein families database in 2019. *Nucleic Acids Res.* 47 (D1), D427–D432. doi: 10. 1093/nar/gky995

[PubMed Abstract](#) | [CrossRef Full Text](#) | [Google Scholar](#)

Qiu, Y., Mekkat, A., Yu, H., Yigit, S., Hamaia, S., Farndale, R. W., et al. (2018). Collagen Gly missense mutations: effect of residue identity on collagen structure and integrin binding. *J. Struct. Biol.* 203 (3), 255–262. doi: 10. 1016/j. jsb. 2018. 05. 003

[PubMed Abstract](#) | [CrossRef Full Text](#) | [Google Scholar](#)

Xiao, J., Yang, Z., Sun, X., Addabbo, R., Baum, J. (2015). Local amino acid sequence patterns dominate the heterogeneous phenotype for the collagen

<https://assignbuster.com/corrigendum-genotypephenotype-association-analysis-reveals-new-pathogenic-factors-for-osteogenesis-imperfecta-disease/>

connective tissue disease osteogenesis imperfecta resulting from Gly mutations. *J. Struct. Biol.* 192 (1), 127-137. doi: 10.1016/j.jsb.2015.05.002

[PubMed Abstract](#) | [CrossRef Full Text](#) | [Google Scholar](#)