

CORRECTION

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Correction to: Identification of novel mutations in congenital afibrinogenemia patients and molecular modeling of missense mutations in Pakistani population

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Correction to: *Thromb J* (2017) 15:24
<https://doi.org/10.1186/s12959-017-0143-3>

Following the publication of this article [1], the authors noted the following typographical errors:

- 1) Affiliation 3 should read “University of Sheffield, Sheffield, United Kingdom” and Affiliations 6, 7, 8 and 9 were unnecessary duplicates
- 2) In the abstract the sentence “Ten patients had mutations in FGA followed by three mutations in FGB and three mutations in FGG, respectively” should be “Ten patients had mutations in FGA followed by four mutations in FGB and two mutations in FGG, respectively.”
- 3) In the Results section the following three sentences:

“In FGA gene, eight mutations were identified as novel and the remaining two were reported mutations. Eight novel mutations include five missense, one nonsense and two frameshift mutations including homozygous and a compound heterozygous frameshift mutation. The two nonsense mutations in FGA are reported in literature. There is one more mutation with reported status in proband (C3). This patient had compound heterozygous mutation with frameshift as novel mutation and nonsense as reported. We identified three mutations in FGB including one novel missense mutation (C9) and two homozygous nonsense mutations reported in siblings. The FGG gene mutations are the rarest of all three fibrinogen genes. We detected three novel mutations

including two similar nonsense mutations in siblings and one frameshift mutation in unrelated proband in different exons of FGG gene (Table 1).”

Should be:

“In FGA gene, seven mutations were identified as novel and the remaining three were reported mutations. Seven novel mutations include five missense and two frameshift mutations including homozygous and a compound heterozygous frameshift mutation. The three nonsense mutations in FGA are reported in literature. There is one more mutation with reported status in proband (C3). This patient had compound heterozygous mutation with frameshift as novel mutation and nonsense as reported. We identified four mutations in FGB including one novel missense mutation (C9), two homozygous nonsense mutations reported in siblings and one frameshift mutation (C12). The FGG gene mutations are the rarest of all three fibrinogen genes. We detected two novel similar nonsense mutations in siblings (Table 1).”

- 4) There are a number of errors in Tables 1 and 2. The corrected versions are provided in this Correction article with the corrections given in bold.
- 5) Frameshift mutation (p.Gln282Thr fsx83*) and (p. Lys (AAA) 48Arg fs9*) are the novel compound heterozygous mutations which have manifested deletions along with frameshift defects” should in fact read “Frameshift mutations (p.Thr283Arg fs138*) and (p. Lys (AAA) 48Arg fs9*) are the novel compound heterozygous mutations which have manifested deletions along with frameshift defects.

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Table 1 Genotypic expression of mutations in fibrinogen gene (*FGA*, *FGB* & *FGG*)

| IP # | Gene | Exon | Mutation | Amino Acid change | Zygosity | Mutation type | Reported/Novel |
|------------------|------------|----------|----------------------------|----------------------------|-----------------------|--------------------|-----------------------|
| C1 | FGA | 1 | c.24C > A | p.Cys8* | Homozygous | Nonsense | Ref #23 € |
| C2 | | 2 | c.143_144 del AA | p.Lys (AAA)48Arg fs9* | Compound Heterozygous | Frame shift | Novel mutation |
| C3 | | 5 | c.846delG | p.Thr 283Arg fs138* | Compound Heterozygous | Frame shift | Novel mutation |
| | | 4 | c.385C > T | p.Arg129* | Homozygous | Nonsense | Ref #24 € |
| C4 | | 4 | c.385 C > T | p.Arg129* | Homozygous | Nonsense | Ref #24 € |
| C5 | | 5 | c.598C > T | p.Gln200* | Homozygous | Nonsense | Ref 27* |
| C6 | | 5 | c.904C > G | p.Pro302Ala | Homozygous | Missense | Novel mutation |
| C7 | | 5 | c.913A > G | p.Thr 305 Ala | Homozygous | Missense | Novel mutation |
| C8 | | 5 | c.992A > G | p.Thr331Ala | Homozygous | Missense | Novel mutation |
| C9 ⁱ | | 5 | c.992A > G | p.Thr331Ala | Homozygous | Missense | Novel mutation |
| C10 | | 5 | c.973A > G | p.Ser325Gly | Homozygous | Missense | Novel mutation |
| C11A | FGB | 2 | c.141 > T | p.Arg47* | Homozygous | Nonsense | Ref # 25 € |
| C11B | | 2 | c.141C > T | p.Arg47* | Homozygous | Nonsense | Ref # 25 € |
| C9 ⁱⁱ | | 8 | c.1294T > A | p.Trp 432Arg | Homozygous | Missense | Novel mutation |
| C12 | | 2 | c.118_124dupTTCTTCA | TTCTTCA | Homozygous | Frame shift | Novel mutation |
| C13A | FGG | 4 | c.361A > T | p.Lys121* | Homozygous | Nonsense | Novel mutation |
| C13B | | 4 | c.361A > T | Lys121* | Homozygous | Nonsense | Novel mutation |

Identified novel and reported mutations in three genes of fibrinogen. The letter A and B with patient code designate the sibling status, **i & ii shows mutation identified in same patient but in different genes**, € (reported mutation) c (complimentary deoxyribonucleic acid), A (adenine), T (thymine), C (cytosine), G (guanine), Lys (lysine), Arg (arginine), Tyr (tyrosine), Pro (proline), Trp (tryptophan), Thr (threonine), Gln (glycine), Cys = cystine, fs = frame shift, * stop codon number, *FGA* (fibrinogen Aα-chain gene), *FGB* (fibrinogen Bβ-chain gene), *FGG* (fibrinogen Gγ-chain gene).

Table 2 Assessment of coagulation markers and bleeding scores with consanguinity/ethnicity

| IP# | Fibrinogen Level (g/l) | Thrombin Time (Sec) | Prothrombin Time (Sec) | Activated partial thromboplastin Time (aPTT) (Sec) | Bleeding Score | Consanguinity | Interfamilial Relation | Ethnic Origin |
|------|------------------------|---------------------|------------------------|--|----------------|---------------|------------------------|---------------|
| *C1 | 0.01 | 23 | > 120 | > 180 | 20 | positive | Unrelated | NA |
| C2 | 0.02 | 24 | > 120 | > 180 | 21 | positive | Unrelated | Punjabi |
| C3 | 0 | 33 | > 120 | > 180 | 22 | positive | Unrelated | Punjabi |
| C4 | 0.1 | 24 | > 120 | > 180 | 17 | positive | Unrelated | Urdu Speaking |
| C5 | 0.02 | 31 | > 120 | > 180 | 20 | positive | Unrelated | Sindhi |
| C6 | 0.01 | 25 | > 120 | > 180 | 20 | positive | Unrelated | Urdu speaking |
| C7 | 0.02 | 29 | > 120 | > 180 | 22 | positive | Unrelated | Sindhi |
| C8 | 0.0 | 30 | > 120 | > 180 | 20 | positive | Unrelated | Sindhi |
| C9 | 0.0 | 32 | > 120 | > 180 | 22 | positive | Unrelated | Punjabi |
| C10 | 0.01 | 25 | > 120 | > 180 | 16 | positive | Unrelated | Punjabi |
| C11A | 0.02 | 28 | > 120 | > 180 | 18 | positive | ** | Punjabi |
| C11B | 0.01 | 24 | > 120 | > 180 | 16 | positive | | Punjabi |
| C12 | 0.0 | 30 | > 120 | > 180 | 21 | positive | Unrelated | Punjabi |
| C13A | 0.02 | 24 | > 120 | > 180 | 20 | positive | ** | Punjabi |
| C13B | 0.01 | 25 | > 120 | > 180 | 21 | positive | | Punjabi |

Shows the individual test values of PT, aPTT and fibrinogen (Clauss Method), consanguinity and the relationship status. Bleeding score calculated, Tostetto et al [26]. ** Siblings, NA = not available, s (seconds). The fibrinogen levels in all patients were found to be equal to or lower than 0.1g/l (Normal Range 2-4 g/dl), PT more than 120 s (Normal Range 9-11 s) aPTT more than 180 s (Normal Range 24-27 s) and prolonged thrombin time (normal range 10-13 s). Ethnicity explains the frequency of majorly affected, thickly populated and largest province of Pakistan (Punjab).

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Received: 6 March 2019 Accepted: 6 March 2019

Published online: 04 April 2019

Reference

1. Naz A, et al. Identification of novel mutations in congenital afibrinogenemia patients and molecular modeling of missense mutations in Pakistani population. *Thromb J*. 2017;15:24. <https://doi.org/10.1186/s12959-017-0143-3>.