Human TAFFAZIN Gene Variants Database Submission Information

Please provide the following information for adding your mutation(s) to the database:

- Use reference sequence NM_000116 (11 exons).
- Mutation position, counting from AUG in the mRNA: Specific nucleotide change (substitution, deletion, insertion). F.ex., c.51G>A, or c.53_54delCC, or c.157dupC.
- Mutation effect at protein level (f.ex.: p.Gly197Arg, or p.Tyr51*, or p.Leu53Argfs*79). Please use the 3 letter abbreviation for amino acids to prevent confusion with nucleotides.
- If a splice mutation, indicate the position in the intron counting from nearest exon. F.ex., c.110-2A>G
- How do we cite your contribution in the Reference/attribution list? f.ex., Direct submission, Jane Doe, Ph.D., Hospital / Laboratory name. If the case has been published, please provide us with the literature citation and how the subject is identified in the citation (f.ex., case #3, or case JL).
- If you know that the mutation is de novo, please state so. Also include if you know in whom the mutation originated (mother, grandmother, grandfather).
- If you have tested cardiolipin/MLCL levels, please tell us the result so it can be included in the proper column (column F).
- Any supporting information? mRNA, familial segregation, etc.
- If you have found variants in other relevant genes, please add this information as there is a separate column for these in the database (column J). F.ex., variant in MYH7, in DMD, etc.
- If you have found any benign variants, please also tell us about it to add them to the appropriate section at the end of the database.

Please email your submissions to the Variants Database Manager, Dr. Prasoon Agarwal: variantsdb@barthstrong.org