Human *Taz* Gene Variants Database
Submission Information

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

- 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 send this information to:

Iris L. Gonzalez, PhD, Senior Research Scientist, Molecular Diagnostics Laboratory, A. I. duPont Hospital for Children, Wilmington, DE.