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# IthaGenes Submission Form

(please return the completed form to [petrosk@cing.ac.cy](mailto:petrosk@cing.ac.cy))

**A. Submitter’s details**

**(to be included in ITHANET Organisations & Experts)**

|  |  |
| --- | --- |
| **Name** |  |
| **Surname** |  |
| **Title** |  |
| **Expertise / research interests** |  |
| **Address** |  |
| **E-mail address** |  |
| **ORCID, if available**  **(**[**http://orcid.org/**](http://orcid.org/)**)** |  |
| **ResearcherID, if available**  **(**[**http://www.researcherid.com/**](http://www.researcherid.com/) **)** |  |
|  |  |
| **Organisation’s name** |  |
| **Organisation’s brief description** |  |
| **Organisation’s address** |  |
| **Organisation’s website** |  |

**Note:**

The information you provide for a specific novel mutation will appear on the IthaGenes website and your submission will be acknowledged.

**B. Mutation details**

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| --- | --- |
| **Parameter** | **Information** |
| **Functionality** | Causative / Modifier / Neutral |
| **Common name**  **(e.g. IVS I-110 G>A or Hb S)** |  |
| **HGVS name**  **(e.g. HBB:c.20A>T)** |  |
| **Gene(s)**  **(e.g. HBB)** |  |
| **Genomic location**  **(e.g. chr11:g.5227002)** | Please provide location on human assembly GRCh38/hg38, or specify otherwise |
| **Ethnic origin** | (any type of ethnicity description of local or international relevance) |
| **Inheritance** | Recessive / dominant / quantitative trait / Other (please specify) |
| **Phenotypic information** | Any information on the phenotype, e.g.   * Allele phenotype (e.g. beta+, alpha0 etc.) * Haematology / biochemistry * Protein stability and oxygen affinity * Associations with clinical compications |
| **dbSNP rs#**  **(if any)** |  |
| **Publication**  **(PubMed ID; if any)** |  |
| **Other comments** | (Any other information that are important for the description of the variation) |

**C. Comments/ suggestions about IthaGenes or the ITHANET Portal in general**

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**THANK YOU FOR YOUR CONTRIBUTION**