

# Diagnostic Mutation Data Base

## Frequently Asked Questions

- Q1. I am a registered user, but I am not able to login?**  
**A.** DMuDB login allows username in lowercase only, username is registered email address, and if you use capital letters you will not be able to login. If you still have problems contact [support@dmudb.net](mailto:support@dmudb.net)
- Q2. If I forget my password, how do I retrieve it?**  
**A.** Contact [support@dmudb.net](mailto:support@dmudb.net) . You will receive a new password, with that password you can login and change it.
- Q3. What is the difference between manual submission and bulk submission?**  
**A.** Manual submission allows adding one referral at a time, bulk submission allows for adding of more than one referral at a time. Bulk submission operation is restricted to the admin only, so users need to send this data to the admin. The DMuDB admin can submit more than one variant data at a time on behalf of the submitter. The submitter can then login and approve data. Contact admin at [support@dmudb.net](mailto:support@dmudb.net) for further details.
- Q4. How do I upload my mutation spreadsheet with mutation data?**  
**A.** It is not possible for user to upload spreadsheets directly. Please see previous question for details.
- Q5. How can I add new disease/gene/reference sequence?**  
**A.** Users can't add new genes, diseases or reference sequences. These operations are restricted to admin only. Please contact [support@dmudb.net](mailto:support@dmudb.net) to add new disease/gene/reference sequence.
- Q6. What sequences are allowed for DMuDB data?**  
**A.** In general we use sequences from NCBI (RefSeq) reference sequences database.
- Q7. In what format data need to be sent and what is the mandatory information to be send?**  
**A.** Excel spreadsheet is preferable. It should contain details of Patient's labID/sampleID, labname, type of dna sample, diagnostic test, variant in HGVS format, disease, gene (OMIM numbers) and reference sequence used. A sample spreadsheet can be downloaded from [http://www.ngml.org.uk/Manchester/Downloads/Informatics/DMuDB\\_Sample\\_Spreadsheet.xls](http://www.ngml.org.uk/Manchester/Downloads/Informatics/DMuDB_Sample_Spreadsheet.xls) .
- Q8. In what format do I present variant information?**

- A.** Variant information need to be in HGVS format only. For more details see HGVS guidelines  
<http://www.hgvs.org/mutnomen/recs-DNA.html> .
- Q9. How is pathogenicity classified in DMuDB?**
- A.** Current pathogenicity values in DMuDB are Unclassified, Pathogenic, Non-pathogenic and Not Known. Use Unclassified if it was not possible to report an interpretation, and use Not Known if you do not know what classification was reported.
- Q10. How many variants a referral can have?**
- A.** There is no limit on the number of variants a referral can have. Generally it is useful to put all the variants of a referral together.
- Q11. What identification detail of the patient am I required to submit?**
- A.** Patient identity information like name and date of birth is not allowed.
- Q12. How can I edit Patient details?**
- A.** Patient details like sampleID and year of birth once entered they are not editable by user.
- Q13. How to anonymise patient data?**
- A.** DMuDB uses encrypted labID/sampleID, it don't need any patient identification information. You need to anonymise labID/sampleID through DMudB anonymising software ( available at <http://ngri.manchester.ac.uk/anonymise/> ) and screencast of anonymising procedure is available at <http://ngri.manchester.ac.uk/screencasts/anonymise-patientId/>