

DMuDB Newsletter

Contact us for user support and data submission

support@dmudb.net

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[latest data](#) | [system updates](#) | [laboratory focus](#)

New data added in the last 2 months

MSH6, MSH2, MLH1	Cheshire and Merseyside Regional Genetics Laboratory (Liverpool), UK
MLH1, MSH2	Manchester Regional Genetics Laboratory, UK
PMP22, MPZ	Bristol Genetics Laboratory, UK
BRCA1, BRCA2	London SE Thames Regional Genetics Laboratory (Guy's Hospital), UK
GJB1	Nottingham Regional Genetics Service, UK
BRCA1, BRCA2	London SW Thames Regional Genetics Laboratory (St Georges), UK
RYR2	Manchester Regional Genetics Laboratory, UK
MFN2	Bristol Genetics Laboratory, UK
APC	Cardiff Regional Genetics Laboratory, UK
EXT1	Laboratory for Diagnostic Genome Analysis, Leiden, Netherlands

DMuDB news

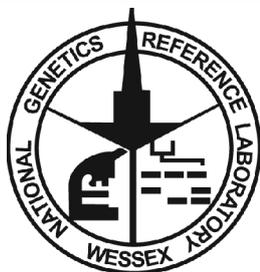
Since our last newsletter we have opened DMuDB subscription to labs outside of the UK and have had a great response! We now have 52 international labs subscribing to DMuDB in just three months, including labs from as far afield as China, Canada and New Zealand. We would like to take this opportunity to welcome all new members to the DMuDB community and we look forward to working with them as they use the database and begin submitting data.

The DMuDB Team is dedicated to working with users to build up the collection of data within DMuDB so that it is an increasingly useful resource for all involved. We base our data collection activities on the needs and suggestions of our users, so please get in touch to let us know of any particular genes for which you would like to see more data. There has recently been particular interest in data for CFTR, CYP21A2, F8 and HFE, so we have been contacting labs to identify new data that could be submitted for these genes. So far we have had an excellent response and hope that all labs will eventually submit their relevant data.

If you would like to submit data for these or any other genes and have questions about how to do so, please don't hesitate to contact us at support@dmudb.net.

Your lab, your voice

Wessex Regional Genetics Laboratory — John Harvey



Wessex Regional Genetics Laboratory

Address for samples to be sent:
Salisbury NHS Foundation Trust
Wessex Regional Genetics Laboratory
Salisbury District Hospital
Salisbury, Wiltshire
SP2 8BJ
UK

To contact the lab:
Tel +44 (0)1722 429080

Director, Head of Service: Prof Nick Cross
ncpc@soton.ac.uk

Head of Molecular and Service Development: Dr John Harvey
john.f.harvey@salisbury.nhs.uk

The Wessex Regional Genetics Laboratory (www.wrgl.org.uk) at Salisbury District Hospital performs genetic analysis for a local population of approximately 2.5 - 3 million in the NHS South region and also undertakes specialised tests for patients throughout the UK and overseas. The WRGL has established a large repertoire of cytogenetic, molecular cytogenetic and molecular genetic tests for both constitutional abnormalities and somatically acquired mutations in cancer. Across the service we received in excess of 16,000 diagnostic referrals in 2010/11, making us one of the larger NHS regional genetics diagnostics services.

An up-to-date list of our services, turn around times and relevant referral forms can be accessed at our web site above. In particular we would like to highlight our High Throughput Screening Facility, which focuses on cost-effective mutation scanning of large genes (e.g. BRCA1, BRCA2, NF1, FBN1) and our array CGH laboratory which undertakes in excess of 2,000 front line tests for individuals with developmental disorders. We will introduce next generation sequencing service in 2012 based on the Illumina platform that will help us to screen expanded panels of genes in a highly cost effective manner.

The WRGL also has a strong research component, conducted partly within the NHS and partly under the auspices of the University of Southampton. In particular our research into the pathogenesis of haematological malignancies and imprinting is competitive at an international level.

In total the WRGL has 90 staff, occupies 1,200m² of space and is fully equipped to meet future challenges in genetic diagnostics. We have full CPA accreditation, are a member of the UK Genetic Testing Network (UKGTN) and participate in international initiatives such as Techgene, Eurogentest and European LeukaemiaNet.

New genes and diseases set up in DMuDB

Multiple Exostoses Type 1 (EXT1)

Multiple Exostoses Type 2 (EXT2)

If you have data on any of these genes, please get in touch (support@dmudb.net) to discuss submitting them to DMuDB.

DMuDB tips

Searching for variants within DMuDB can sometimes be problematic, especially if the variant name is complex. The Universal Browser (<https://ngri.manchester.ac.uk/Browser/>) was developed alongside DMuDB in order to provide a more user-friendly way of browsing the database. In addition to accessing DMuDB variants, the Browser will also display variants from multiple other databases in one view, making it easy to check for the presence of your variant in many databases at once.



DMuDB Data Alerts

If you would like to hear about new data available in DMuDB, and new genes that have been set up, on a regular basis, then sign up to receive DMuDB Data Alerts. Simply email support@dmudb.net with 'Subscribe DMuDB alerts' in the subject line and you will receive a simple text email with details of new data submissions, on a weekly basis.

Data collection

The DMuDB Team works hard to encourage laboratories to participate in the DMuDB project by submitting data to the database. We are always happy to help and support labs to collate and submit their data, and will keep in contact throughout the process to ensure the data are uploaded to DMuDB correctly.

Data collection is driven to a large extent by user demand—so please contact us to let us know which genes your laboratory provides services for, and what your priorities for data collection are.

support@dmudb.net

DMuDB news continued...

Somatic mutation data

Recent updates to DMuDB have allowed us to store somatic mutations in the database. This facility has been developed as part of a UK project that aims to establish a repository of EGFR test data. The storage of patient treatment and response data is also possible. At present EGFR test data is being collected from UK laboratories, however it is possible to store other somatic mutation data, e.g., KRAS, KIT, PDGFRA, etc.

Data upload developments

Bulk data submissions on Excel spreadsheets can now be uploaded directly to the secure DMuDB server using the DMuDB 'Modules' tab, once you are logged into the system. DMuDB curators are notified of new uploads and can then retrieve the files and proceed with formatting and uploading your data as normal. As only DMuDB curators have access to the files that you upload, this is the most secure way to send data to the DMuDB teams and avoids the security risks associated with sending files by email.

We have also been working with a number of popular laboratory software developers to implement DMuDB-specific data sharing features that could simplify the data submission process for users. The latest version of Alamut (www.interactive-biosoftware.com) now provides an improved 'Export to DMuDB' option, which enables users to export their data into a spreadsheet that is ready-formatted for upload to DMuDB. We have also been working with our local implementation of the STARLIMS system to integrate a similar data export functionality. We hope that, once finalized, this module will be available for roll-out to other laboratories with STARLIMS implementations.

Want to appear in our next newsletter?

If you would like to feature in our 'Your Lab, Your Voice' section, send a short article about your laboratory (approx. 250 words) plus a picture of the building, or a logo, to support@dmudb.net and you could be in our next newsletter! Send your information to us as soon as possible as only one lab can be selected each issue.

Bioinformatics training courses

In partnership with NGRL Manchester, Nowgen (www.nowgen.org.uk) runs a number of training courses for healthcare professionals, focussed on bioinformatics for diagnostic genetics. For more information, or to register your interest, contact bookings@nowgen.org.uk.

Proposed course dates for 2012 are:

<i>Bioinformatics for Genetic Scientists</i>	
28 Feb - 1 Mar 2012	2 - 4 Oct 2012
22 - 24 May 2012	27 - 29 Nov 2012

Bioinformatics for Clinical Geneticists
16 - 17 Oct 2012