Empowering Australian Genomics

SERVICE GUIDE

Clinical Variant Confirmation

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1.0 Overview

Next generation sequencing (NGS) technologies and the advent of whole exome and whole genome sequencing has significantly improved the detection of clinically relevant genetic variants. With this comes the need to confirm the presence of variants of diagnostic importance. Sanger sequencing is widely considered the gold standard technology for DNA sequence analysis and is therefore ideal for confirming variant calls made on NGS platforms.

AGRF's clinical variant confirmation service can provide confirmation of any variant using Sanger sequencing. Our service is NATA accredited to a clinical standard (ISO15189). Simply provide the genomic coordinates of your variant/s and submit your purified genomic DNA; and we will design and order primers, amplify and sequence the variant. Data is then supplied for your interpretation – for clinical use this must be performed by a qualified clinical pathologist.

2.0 Quality Assurance

All aspects of this service follow strict requirements of ISO15189: 2013. AGRF Ltd is accredited as a Medical Testing laboratory according to the ISO15189: 2013 standard by the National Association of Testing Authorities (NATA). Compliance is monitored at regular reviews and internal audits. The work is supervised by an AGRF staff member with relevant qualifications and checked while in progress and upon completion to ensure the necessary standards are met.

3.0 Sample and Data Storage

Samples are stored at AGRF for three months after you receive your data. If you wish for your samples to be returned, you must let us know at the time when you are submitting your samples. At the completion of sequencing, we will return your samples by post, using an Australia Post Express Post satchel at ambient temperature. If required, we can return samples using dry ice, which will incur a $150 charge. If we do not hear back from you by the date specified in your data delivery email, we will discard your samples.

AGRF maintains an archive of client data, however, clients are advised to back up their data themselves. Data will remain available for download for 30 days, prior to archive. If you require past data, please contact AGRF. Please note that charges will apply for restoring files to the server for data more than six months old.

4.0 Turnaround Time

The turnaround time is 3-4 weeks from receipt of samples.

5.0 Sample Submission Requirements

5.1 Sample Preparation

Samples should be submitted as purified genomic DNA, at 10ng/ul in a 10ul volume.
Submit your samples to AGRF Melbourne for processing (samples may be sent ambient).

5.2 Online Submission

- In the client portal, select 'Clinical Variant Confirmation' from the service dropdown menu
- Enter your submission format (tube or plate)
- Complete and upload the template file/s
- Submit the form and print the submission receipt to be included with your sample package

5.3 Packing of Samples

Samples can be shipped at room temperature via express post or courier, or delivered in person. To prevent leakage in transit please use parafilm to seal tubes, and ensure plates are heat-sealed or sealed with strip caps.

Post/send/deliver samples to the addresses below:

Physical address (courier)
ATTN Sanger Sequencing Team
AGRF
VCCC Loading Dock
14 Flemington Road
North Melbourne, VIC 3051

Postal Address (mail)
ATTN Sanger Sequencing Team
AGRF
Level 13, Victorian Comprehensive Cancer Centre
305 Grattan Street
Melbourne, VIC 3000

6.0 Data Output

Each batch submitted will receive the following files for each sample processed:

- The raw electropherogram file (sample.ab1)
- The sequence file (sample.seq), the sequence represented in a text format
- The FASTA file (sample.fa), reads are quality trimmed and the sequence represented in a text format
- A BLAST of the trimmed FASTA file (sample.bn), this text file comprises the top 10 hits against the NCBI GenBank database (sample.bn)