SERVICE GUIDE

Clinical Variant Confirmation

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SVG2402CVC

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1.0 Overview

Advancements in Next Generation Sequencing (NGS), particularly whole exome and whole genome sequencing, have significantly enhanced the identification of clinically relevant genetic variants. However, to validate these findings, confirmation is crucial. Sanger sequencing, recognized as the gold standard for DNA sequence analysis, is ideal for verifying variant calls made on NGS platforms.

AGRF offers a clinical variant confirmation service with NATA accreditation to the clinical standard (ISO15189). By providing genomic coordinates and purified genomic DNA, we design and order primers, conduct amplification and sequencing of the variant. The resulting data is supplied for interpretation, a step that, for clinical use, should be performed by a qualified clinical pathologist.

2.0 Turnaround Time

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

3.0 Sample and Data Storage

Samples are stored with AGRF for 3 months after you receive your data. If you wish for your samples to be returned, you must discuss this with your account manager during quoting or contact us after you receive your data. At the completion of your project, we can either:

- Return your samples by courier at ambient (please ask your account manager for a quote).
- Return samples by courier with dry ice (please ask your account manager for a quote).

If we are not notified within the specified time frame, samples will be automatically discarded.

4.0 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)

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.

AGRF can organise dry ice shipment for your samples as part of your quoted services or you can use our free shipping between nodes once a week service. For information on this service go to Free Shipping.

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 Quality Statement

All clinical works carried out by AGRF follow the strict requirements of ISO15189. AGRF Ltd is accredited by the National Association of Testing Authorities (NATA) in the field of Medical Testing (Scope: Investigation of constitutional genetic variants - Diagnostic Testing. Whole exome sequencing studies for inherited (germline) DNA/RNA changes). Staff and analysis processes follow Standard Operating Procedures, which define responsibilities and quality checks to achieve reported standards. Compliance is monitored at regular reviews and during internal audits. The work is supervised by a person with relevant qualifications and checked while in progress and upon completion to ensure that it meets the necessary ISO15189 standards.