Policy

Incidental Findings for Genomics

NSWHP_PD_029

1. Purpose

This document sets out the requirements for the reporting of Incidental Findings identified by Genomic testing within NSW Health Pathology.

2. Background

Incidental findings are becoming more prevalent as genomic testing moves from single gene testing to large gene panels and even exome and genome testing.

The likelihood of identifying an incidental finding increases significantly as the regions of the human genome targeted for sequencing analysis are expanded

3. Scope

The scope for this policy includes all laboratories undergoing massively parallel sequencing beyond small targeted panels. The policy is available to all referring services.

The scope excludes Forensic and Analytical Science Services (FASS).

4. Definitions

Incidental finding: A finding of medical significance that is present in the genomic data but is unrelated to the reason for referral. This definition is independent of whether the finding is in a gene that is systematically searched or not.

5. Policy Statement

5.1. Consultation Process

In the majority of referred cases the reason for referral and the requested target genes to be reported will be clear. However, in all cases where there is ambiguity as to the gene variants to be reported a consultation process between the laboratory and clinical referral service must occur.

This consultation process must:

a) occur prior to the laboratory issuing a report;
b) occur between the laboratory, their clinical service and the referring clinician;
c) determine whether a specific incidental finding should be reported; taking into account

- patient consent
- variant quality and robustness
- clinical value of the finding, and
- significance to other family members.

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d) Be recorded in an appropriate system such as the site-specific LIS or a separately maintained record system for this purpose that complies with local site-specific operational procedures and audit capability if required.

5.2. Patient Consent to Receive Incidental Findings

All referrals for genomic testing (implying any individual undergoing massively parallel sequencing) must:

a) include a clear indication of the reason for referral
b) be accompanied by a consent form that covers the reporting of incidental findings; or indication on the request form that consent is on file; and
c) evidence of consent having been obtained must be retained in a NSWH and/or a NSWHP records management system.

The laboratory must comply with NPAAC Massively Parallel Sequencing Guidelines[1] in relation to:

a) Reducing the chance of identifying incidental findings; and
b) Ensuring that testing and reporting complies with patient consent.

If an incidental finding is identified, the laboratory must:

a) Conduct the consultation process again; and
b) Determine whether the variant identified is truly an incidental finding or is within the reason for referral.

5.3. Analysis of Variants Detected by Gene Panels/Whole Exome Sequencing/Whole Genome Sequencing

Analysis of variants should be restricted to those associated with the referral in accordance with NPAAC Guidelines[1].

The laboratory must consult with the referring clinician/service to obtain written clarification on the genes that should be analysed and reported in the following circumstances:

a) Analysis of genes beyond the scope of the referral; or
b) In the case of ambiguity.

5.4. Storage of Detected/Curated Incidental Findings

All variants must be stored in a NSW Health Pathology database regardless of whether the variant is reported or not. The level of curation may differ for variants not associated with the referral.

Variants categorised as incidental findings that are stored in the NSW Health Pathology database must not be reported without patient consent.

6. Roles and Responsibilities

The responsibility for determining whether a variant is an incidental finding lies with the clinical scientist or genetic pathologist responsible for reporting the test through consultation with the referrer and/or clinical services where appropriate.

7. Legal and Policy Framework

[1] *NPAAC Guidelines – Requirements for human medical genome testing utilising massively parallel sequencing (First Edition 2017).*

8. Review

This policy will be reviewed by 18/11/2022

9. Risk

Risk Statement	If incidental findings are reported without consent due to non-compliance with this policy the consequences could result in harm to the patient.
Risk Category	Clinical Care and Patient Safety, Health of the Population

10. Further Information

For further information, please contact:

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11. Version History

The approval and amendment history for this document must be listed in the following table.

Version No	Effective Date	Approved By	Approval Date	Policy Author	Risk Rating	Sections Modified
1.0	23/11/2020	Clinical Governance Quality and Risk Committee	17/11/2020	Dr Cliff Meldrum	High	New Policy
1.1	12/04/2021	Director Genomics	7/04/2021	Dr Cliff Meldrum	High	5.4 Typographic error, “must not” inserted into second paragraph