NSW Health Pathology

Policy

Incidental Findings for Genomics

NSWHP_PD_029

1. Purpose

This document sets out the requirements for reporting 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 more regions of the human genome undergo sequencing analysis.

3. Scope

The scope of this policy includes all laboratories performing massive parallel sequencing and is in accordance with the National Pathology Accreditation Advisory Council (NPAAC) Guidelines, which 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 includes but is not limited to unexpected familial relationships.

5. Policy Statement

5.1 Consultation Process

In the majority of cases, the reason for referral and the target genes to be reported will be clear. 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; acknowledging
   - 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 approved system that complies with operational procedures and audit capability.

5.2 Patient Consent to Receive Incidental Findings

Referrals for level 3 genomic testing as defined by NPAAC requirements must:

a) include a clear indication of the reason for the 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) have evidence of consent retained in a NSWH and/or a NSWHP records management system.

The laboratory must comply with NPAAC requirements in relation to:

a) Reducing the chance of identifying incidental findings; and
b) Ensuring testing and reporting comply with the laboratory reporting policy and patient consent.

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

Analysis of variants should be concordant with the reason for referral.

The laboratory must consult with the referring clinician/service to obtain written clarification on the genes that should be analysed and reported 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 they are reported. The level of curation may differ for variants not associated with the referral.

Variants categorised as incidental must not be reported without patient consent.

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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 following consultation with the referrer and/or clinical services.

7. Legal and Procedure Framework

National Pathology Accreditation Advisory Council (NPAAC) Guidelines – Requirements for medical testing for human genetic variation, Third Edition 2022.

8. Review

This procedure will be reviewed by 01/08/2027.

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 Choose at least one category	Assets & Infrastructure ☐; Clinical Care & Patient Safety ☒; Compliance ☒; Cyber Security ☐; Governance ☒; Information, Technology & Data ☐; Leadership & Management ☒; People & Culture ☐; Resilience ☐; Technological ☐

10. Further Information

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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	Procedure Author	Risk Rating	Sections Modified
1.0	23/11/2020	Clinical Governance Quality and Risk Committee	17/11/2020	Dr Cliff Meldrum	High	New Procedure
1.1	12/04/2021	Director Genomics	7/04/2021	Dr Cliff Meldrum	High	5.4 Typographic error, “must not” inserted into second paragraph
2.0	02/07/2024	Associate Director Genomics Director Clinical Services and CMIO	26/06/2024	Nicole Pearce	High	Updated references current to NPAAC guidelines. Deleted text in 5.2 covered by NPAAC guidelines.

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