1. Purpose

This document outlines the process for the return of Incidental Findings that are found in the process of testing the biospecimen for research. An Incidental Finding is a research finding that has a health implication for the participant or their genetic relatives.

2. Scope

This policy is only relevant for collections that are under the custodianship of NSW Health Statewide Biobank (NSWHSB); it applies to NSWHSB staff and researchers curating, coordinating or investigating collections under the custodianship of NSWHSB.

Consenting to biobanking means consenting to receiving any Incidental Finding that has been found by the researcher while they are doing their research. Only findings that are significant, clinically actionable and confirmed will be returned to the participant (see Section 3 for more information on these definitions).

The return of Incidental Findings involves the re-identification of the participant. Re-identification will be done by the NSWHSB by joining up the information from the researcher with that held by the NSWHSB, and does not involve using MBS/PBS data.

3. Definitions

- **ACMG:** American College of Medical Genetics and Genomics
- **Biospecimen Aliquot Specific Number:** A unique number which identifies the biospecimen aliquot
- **Biospecimen metadata:** The types of data linked to the biospecimen that may be associated with the collection and subsequently requested by researchers
- **Biospecimen related data:** Data directly associating biospecimen metadata (e.g. biospecimen storage information) with participant data (e.g. name, address)
- **CHeReL:** Centre for Health Record Linkage
- **Ethically Defensible Plan (EDP):** An EDP describes the management of disclosure to participants of any Incidental Findings that have been discovered during the research
- **Human Genetics Society of Australasia (HGSA):** A forum for the various disciplines collected under the title of Human Genetics
- **Medically actionable finding:** Incidental Findings are research findings which have health implications for the participant or their genetic relatives. Only those findings which meet each of the following criteria will be returned:
  - **Significant:** The finding indicates a life-threatening condition
  - **Clinically actionable:** There are specific and established therapeutic

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1 Only biospecimen collections housed within the NSWHSB will be linked to MBS/PBS data.
interventions or other available actions

- **Confirmed:** The finding has been checked and confirmed as far as reasonably possible in a research context to be accurate and/or valid.

To promote standardised reporting of actionable information from clinical genomic sequencing, the American College of Genetics and Genomics (ACMG) has published a minimum list of genetic conditions to be reported as clinically actionable or secondary findings (Appendix B).

- **Material Transfer Agreement (MTA):** A binding legal agreement setting out the conditions of transfer and use between a biobank and the researcher(s) receiving the biospecimens and data

- **MBS/PBS:** Medicare Benefits Scheme/Pharmaceutical Benefits Scheme

- **National Association of Testing Authorities (NATA):** Provider of assessment, accreditation and training services to laboratories and technical facilities

- **Nominated contact/clinician (NC):** The clinician nominated within the research project’s Ethically Defensible Plan who will be responsible for evaluating possible findings and managing their return

- **NSWHSB:** NSW Health Statewide Biobank

### 4. Policy Statement

The directions below accompany a process flow diagram shown as Appendix A.

#### 4.1 Development of Ethically Defensible Plan (EDP)

4.1.1 Researchers and/or NSWHSB will develop an EDP in accordance with the principles outlined in the National Statement on Ethical Conduct in Human Research 2007.

#### 4.2 Researcher applications to the NSWHSB

4.2.1 Researchers are to be informed of the criteria that apply to potential Incidental Findings that arise during their research, i.e. any finding which is clinically diagnosable through a NATA accredited laboratory and treatable (see Appendix B).

4.2.2 As part of their Material Transfer Agreement, researchers must agree to notify the NSWHSB of any Incidental Findings.

4.2.3 Biospecimens and associated data will be distributed to researchers without identifying details. A Biospecimen Aliquot Specific Number will be supplied for each biospecimen aliquot. This is for two purposes:

- to allow comparison of individual participant biospecimens/data within the laboratory setting whilst retaining privacy; and
- to allow for re-identification of participants by the NSWHSB in the event of discovery of a medically actionable Incidental Finding.
4.3 Discovery of Incidental Finding

4.3.1 Incidental Findings can arise from either of the following research approaches:

- direct analysis of individual biospecimens; or
- re-analysis of previous research outputs (such as Whole Genome Sequences) that arose from individual biospecimens.

4.3.2 Datasets to be linked by CHeReL (e.g. NSW Health administrative datasets, MBS/PBS datasets) will be held separately to the NSWHSB, and distributed to researchers without identifying details. These datasets have been made available to CHeReL under a managed disclosure process.

4.3.3 Incidental Finding information will not arise from analysis of MBS/PBS data. The purpose of making MBS/PBS data available to researchers is to complement/enhance the breadth and quality of information available to researchers; however, the information contained within the MBS/PBS datasets cannot and will not form the basis of any Incidental Findings on individual biobank participants.

4.3.4 For the duration of their research, researchers will remain aware of their responsibility to notify the NSWHSB of Incidental Findings of which they become aware.

4.3.5 Researchers will make every attempt to ensure that any Incidental Finding has been checked and confirmed as accurate and/or valid. This checking will include reviewing biospecimen/sample handling procedures, contamination errors, checks on instrument/test accuracy, and reproducibility within the research laboratory.

4.4 Notification of an Incidental Finding from researcher to NSWHSB

4.4.1 Following research confirmation of validity and accuracy, the researcher will notify the NSWHSB by completing and emailing NSWHSB_F_0001 Return of Incidental Findings, a copy of which can be obtained from the NSWHSB website (Reference 6.6). The researcher will provide the following information:

- the Biospecimen Aliquot Specific Number;
- a detailed description of the medically actionable finding;
- the reasons that the researcher identified the Incidental Finding as one that should be returned to a participant;
- the chain of custody/storage that has occurred following researcher receipt of the biospecimen;
- the method/s used to identify the Incidental Finding (information that reveals intellectual property from the research is not required);
- the method/s used to confirm the Incidental Finding; and
whether there is residual biospecimen that could be used for confirmation in an accredited laboratory.

4.5 Decision on whether a potential Incidental Finding is a confirmed Incidental Finding.

4.5.1 The information that is provided by the researcher to the NSWHSB will be assessed by the NSWHSB Clinical Research Director who will confirm that it is an Incidental Finding and should be reported back to the participant.

4.5.2 As required, the NSWHSB will arrange for the relevant residual anonymised biospecimen to be transported from the researcher to an accredited laboratory, or a matching aliquot housed in the NSWHSB to a NATA accredited laboratory. The confirmatory specimen should be identified to the testing laboratory only by its Biospecimen Aliquot Specific Number.

4.6 NSWHSB re-identification of participant.

4.6.1 Participant re-identification for the purposes of assessing and returning Incidental Findings will occur in limited circumstances where a potential Incidental Finding has been confirmed.

4.6.2 The re-identification will be limited to the participant information held by the NSWHSB and the custodian of the biospecimen collection (i.e. biospecimen related data).

4.6.3 NSWHSB will confirm that the information supplied by the researcher aligns with the biospecimen related data stored by NSWHSB.

4.6.4 NSWHSB will re-identify the participant without disclosing the identity to the researcher.

4.6.5 NSWHSB will compile a dossier containing:

- the name, date of birth and contact details of the participant;
- a copy of their NSWHSB consent form;
- biospecimen related data that the NSWHSB holds;
- contact details of the researcher, nominated clinician, and any other relevant clinicians (e.g., the appropriate Head of Clinical Services or Genetic Services);
- the availability of any duplicate and/or residual biospecimen aliquots that could be used for confirmation of the medically actionable finding in an accredited laboratory; and
- the nature of the Incidental Finding.

4.6.6 Re-identification of MBS/PBS data is not required for the return of Incidental Findings.
4.7 Communication

4.7.1 NSWHSB will contact the participant’s nominated contacts and/or clinician to notify them that their patient’s biospecimen has been associated with a confirmed Incidental Finding. The following people will be contacted in this order:

i. Nominated clinician and/or consenting clinician
ii. Participant’s GP
iii. Participant themselves by the NSWHSB Clinical Research Director

4.7.2 Contact will also be made with genetics and cancer genetics services in NSW, where relevant.

4.7.3 The NSWHSB will forward the dossier of relevant information to the nominated contact/clinician. The nominated contact/clinician will put the participant in contact with a treating clinician and potentially a genetic counsellor.

4.8 Reporting

4.8.1 Notification of outcome of the Incidental Finding will be recorded and reported annually to the Office of Health and Medical Research, NSW Health as required under the performance agreement. Data to be reported will include:

- who had been notified;
- what the finding was;
- how it was communicated; and
- any feedback from clinicians, participants, researchers regarding the process.

5. Roles and Responsibilities

<table>
<thead>
<tr>
<th>Position</th>
<th>Responsibilities</th>
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<tr>
<td>Researchers</td>
<td>Development of EDP; confirmation of validity of Incidental Findings; notification to NSWHSB of Incidental Findings.</td>
</tr>
<tr>
<td>Clinical Research Director</td>
<td>Development of EDP; assessment of returned Incidental Findings; organisation of confirmatory testing; reporting of findings to NCs and other relevant agencies</td>
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</tbody>
</table>
6. Legal and Policy Framework


6.2 Recommendations for reporting of secondary findings in clinical exome and genome sequencing, 2016 update (ACMG SF v2.0) - a policy statement of the American College of Medical Genetics and Genomics.

6.3 CSER toolkit: https://www.ashg.org/education/csertoolkit/medicallyactionable.html

6.4 Medically actionable findings in clinical genomics: a clarification; American College of Medical Genetics and Genomics: https://www.nature.com/articles/gim2016190/tables/1


6.6 NSWHSB_F_0001 Return of Incidental Findings:

7. Review

The List of Incidental Findings (Appendix B) will be updated formally annually, but can be informed by novel reports from literature in real-time. Processes will be reviewed annually at time of reporting. This procedure will be reviewed by 19/11/2019.

8. Risk

<table>
<thead>
<tr>
<th>Risk Statement</th>
<th>If misdiagnosis occurs due to a failure to report an incidental finding the consequences could result in reputational damage and/or further adverse effects to the patient’s wellbeing and that of their family.</th>
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<tr>
<td>Risk Category</td>
<td>Clinical Care and Patient Safety</td>
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9. Further Information

For further information, please contact:

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<thead>
<tr>
<th>Policy Contact Officer</th>
<th>Position: Clinical Research Director</th>
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<tbody>
<tr>
<td></td>
<td>Name: Craig Gedye</td>
</tr>
<tr>
<td></td>
<td>Telephone: 0432 286 616</td>
</tr>
<tr>
<td></td>
<td>Email: <a href="mailto:craig.gedye@health.nsw.gov.au">craig.gedye@health.nsw.gov.au</a></td>
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10. Version History

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

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<th>Approved By</th>
<th>Approval Date</th>
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<td>19/11/2018</td>
<td>Craig Gedye</td>
<td>19/11/2018</td>
<td>SC</td>
<td>High</td>
<td>Initial document</td>
</tr>
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Appendix A: Process flow for Incidental Findings

1. **Researcher** develops EDP (in consultation with NSWHSB)
2. **Researcher** applies to biobank, project is approved and tissue/data is distributed to researcher
3. If a **Researcher** discovers potential Incidental Finding using the medically actionable list
4. **Researcher** notifies NSWHSB of the Incidental Finding using the incidental findings form
5. NSWHSB receives notification from the researcher
6. If required, NSWHSB arrange for residual biospecimen to be analysed to confirm analysis
7. NSWHSB confirms that the finding is an Incidental Finding
8. NSWHSB re-identifies participant by re-joining the researcher information with the biospecimen and forwards dossier of relevant information to NC
9. **NC** notifies participant of the Incidental Finding
   - **Response**
     - NC follows up with other records
   - **No response**
     - Current participant contact details are not found
     - Current participant contact details are found
     - NC puts participant in contact with treating clinician +/- genetic counsellor
     - NC discusses Incidental Finding with participant

The re-identification of MBS/PBS data is not required for returning incidental findings.

**EDP** = Ethically Defensible Plan
**NC** = Nominated contact/clinician
**NSWHSB** = NSW Health Statewide Biobank
**CHeReL** = Centre for Health Record Linkage

**Author:** Simon Cooper  
**Risk Rating:** High

**Approved by:** Craig Gedye  
**Version:** 1.0

**Modified:** 19/11/2018  
**First Published:** 19/11/2018
Appendix B: American College of Medical Genetics and Genomics v2.0 genes and associated phenotypes recommended for return of secondary findings in clinical sequencing

In the absence of an Australian endorsed list of Incidental Findings, we will ask researchers to report findings on this US list.

<table>
<thead>
<tr>
<th>Phenotype</th>
<th>MiM disorder</th>
<th>PMID Gene Reviews entry</th>
<th>Typical age of onset</th>
<th>Gene</th>
<th>MiM gene</th>
<th>Inheritance</th>
<th>Variants to report</th>
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<td>Hereditary breast and ovarian cancer</td>
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<td>Adult</td>
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<td>113705</td>
<td>AD</td>
<td>KP and EP</td>
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<td>BRCA2</td>
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<td>Lynch syndrome</td>
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<td>MLH1</td>
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<td>20301519</td>
<td>Child/adult</td>
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<td>MYH-associated polyposis; adenomas, multiple colorectal, FAP type 2; colorectal adenomatous polyposis, autosomal recessive, with pilomatrixomas</td>
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<td></td>
<td>SMAD4</td>
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<td>Von Hippel–Lindau syndrome</td>
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<td>Multiple endocrine neoplasia Type 1</td>
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<td>613733</td>
<td>AD</td>
<td>KP and EP</td>
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</table>
### Procedure

#### Return of Incidental Findings

**Phenotype** | **MiM disorder** | **PMiD Gene Reviews entry** | **Typical age of onset** | **Gene** | **MiM gene** | **Inheritance** | **Variants to report** |
--- | --- | --- | --- | --- | --- | --- | --- |
Multiple endocrine neoplasia type 2 | 171400 162300 | 20301434 | Child/adult | RET | 164761 | AD | KP |
Familial medullary thyroid cancer | 155240 | 20301434 | Child/adult | RET | 164761 | AD | KP |
*PTEN* hamartoma tumour syndrome | 153480 | 20301661 | Child/adult | PTEN | 601728 | AD | KP and EP |
Retinoblastoma | 180200 | 20301625 | Child | RB1 | 614041 | AD | KP and EP |
Hereditary paraganglioma- phaeochromocytoma syndrome | 168000 (PGL1), 601650 (PGL2), 605373 (PGL3), 115310 (PGL4) | 20301715 | Child/adult | SDHD | 602690 | AD | KP and EP |
Tuberous sclerosis complex | 191100 613254 | 20301399 | Child | TSC1 | 605284 | AD | KP and EP |
WT1-related Wilms tumor | 194070 | 20301471 | Child | WT1 | 607102 | AD | KP and EP |
Neurofibromatosis type 2 | 101100 | 20301380 | Child/adult | NF2 | 607379 | AD | KP and EP |
Ehlers-Danlos syndrome, vascular type | 130050 | 20301667 | Child/adult | COL3A1 | 120180 | AD | KP and EP |
## Procedure

### Return of Incidental Findings

**NSWHSB_P_0001**

**Author:** Simon Cooper  **Risk Rating:** High

**Approved by:** Craig Gedye  **Version:** 1.0

**Modified:** 19/11/2018  **First Published:** 19/11/2018

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<th>Phenotype</th>
<th>MiM disorder</th>
<th>PMiD Gene Reviews entry</th>
<th>Typical age of onset</th>
<th>Gene</th>
<th>MiM gene</th>
<th>Inheritance</th>
<th>Variants to report</th>
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## Procedure

### Return of Incidental Findings

**NSWHSB_P_0001**

- **Author:** Simon Cooper
- **Risk Rating:** High
- **Approved by:** Craig Gedye
- **Version:** 1.0
- **Modified:** 19/11/2018
- **First Published:** 19/11/2018

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### Table of Phenotypes and Variants

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<th>Phenotype</th>
<th>MiM disorder</th>
<th>PMiD Gene Reviews entry</th>
<th>Typical age of onset</th>
<th>Gene</th>
<th>MiM gene</th>
<th>Inheritance</th>
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<td>KP and EP</td>
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<td>Wilson disease</td>
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<td>20301685</td>
<td>Child</td>
<td>ATP7B</td>
<td>606882</td>
<td>ARc</td>
<td>KP and EP</td>
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<td>Ornithine transcarbamylase deficiency</td>
<td>311250</td>
<td>24006547</td>
<td>Newborn (male), child (female)</td>
<td>OTC</td>
<td>300461</td>
<td>XL</td>
<td>KP, EP (hemi/het/hom)</td>
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<td>20301325</td>
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<td>RYR1</td>
<td>180901</td>
<td>AD</td>
<td>KP</td>
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**Phenotype:**
- MiM disorder
- PMiD Gene Reviews entry
- Typical age of onset
- Gene
- MiM gene
- Inheritance
- Variants to report