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Incidental Findings in IWK Clinical Genomics Next Generation Sequencing Policy

Incidental findings (IFs) are defined as unexpected clinically relevant findings identified during genetic testing analysis (i.e. not actively searched for). These are gene variants that are unrelated to the primary indication(s) for which the test is being performed. Analyses should minimize the identification of IFs when possible. The absence of incidental findings in a report does not suggest that these variants are not present within the genetic testing data. Additionally, the possibility of detecting IFs differs across patients, sample types and analysis approach.

For most Next Generation Sequencing (NGS) analyses, IFs are not actively searched for and are therefore not reported. The following guidelines are applied when IFs are identified (applicable **only** for Whole Exome Sequencing tests):

1. Only IFs classified as likely pathogenic or pathogenic are reported;
2. Reportable IFs are largely confined to a pre-defined list of genes recommended by The American College of Medical Genetics and Genomics (ACMG). Please refer to the latest version of the ACMG recommendations for complete details of the genes and associated disorders.
    a. These should be reported regardless of the age of the patient.
3. Actionable childhood-onset IFs are reported, including those identified as part of a primary finding (i.e. a cancer predisposition gene within a large pathogenic deletion);
4. The following IFs are not generally reported:
    a. Low-penetrance genetic predispositions;
    b. Pharmacogenetic findings;
    c. Carrier status
        i. note: carrier status may be reported when there is a significant risk to the family for conceiving an affected child
    d. Biological relationships that differ from those reported by the family, unless necessary to avoid diagnostic interpretation errors and/or recurrence risk estimates

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It is the responsibility of the ordering clinician(s) to provide comprehensive pre and post-test counselling to the patient and their family regarding IFs. Clinicians should discuss the possibility that testing may generate IFs and that they can opt-in or opt-out to have these reported. The patient and/or caregiver must be consented appropriately and their choice should be recorded in the patient's requisition and their medical record.

If the patient or the caregiver opts-in for analysis and reporting of IFs, these are reported in a separate document. The status of any IFs that are reported for the affected individual will be provided for all relatives included as a part of the proband's test. Reported variants will be confirmed using an alternate test method when appropriate (i.e. Sanger sequencing).