Gene Screen BC

Incidental Findings

Incidental Genomic Findings

Unexpected genetic changes that have important medical or social implications unrelated to the reason for testing

While the development of new genomic technologies offers the promise of identifying mutations that underlie many diseases, this same technology is also likely to uncover many unexpected genetic changes that have important medical or social implications unrelated to the disease for which the testing was obtained. Such findings not only have implications for the person being tested, but also potentially for their family members.

• What should be done with these unexpected findings?

• What are the public, parental, ethical and medical perspectives?

Historical Example: a genetic test is performed (e.g. for cystic fibrosis) and unexpectedly a child's father is revealed not to be the biological father. Typically a genetic counsellor will not breach confidentiality in this instance as there is no medical benefit for the father in this circumstance. The potential harm that could be done to the family out-weighs the benefit of his knowing this information. The counsellor may rather reveal the information to the mother and allow her to decide if it should be revealed. However with an unexpected finding that might have medical implications we might feel differently.

Current technology has the potential to magnify these types of incidental findings thousands of times and the likelihood that they will have medical implications is high. Currently there are no clinical guidelines for disclosure or non-disclosure of information found in this way.

• Should the scope of duty to disclose should vary depending on "the type of study, clinical significance and reliability of the information?

• What kind of information should be returned?

  o Raw sequence (e.g. like providing a book full of incomprehensible letters)?

  o Clinically relevant information (what is relevant today versus tomorrow differs)?

  o Results with uncertain significance (e.g. we do not yet understand them)?

Some Recent Examples

Miller Syndrome: a rare genetic condition characterized by distinctive craniofacial malformations that occur in association with limb abnormalities. In a research study of 3

families with this condition, a gene responsible for ciliary dyskinesia was found in one family that explained their recurrent lung infections. This was a welcome incidental finding as it helped manage this issue

Array Technology: used to screen a 7 year old girl with an unknown diagnosis for her intellectual disability. Test revealed a deletion of 47 genes on chromosome 17 and several of these genes were responsible for her symptoms. However, another gene responsible for preventing cancer had also been deleted giving her an 80 – 90% lifetime risk of developing cancer.

Useful Link –
http://phsa.mediasite.com/mediasite/Viewer/?peid=b31fdca2bacf4723bcb79f4abd4b17e71d