Assessing Genomic Sequencing Information for Health Care Decision Making

Patient Care and Health Decisions

A Patient Perspective

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Background

- Cardiomyopathy, Ventricular Tachycardia

Whole exome sequencing

Perspective on RESULTS:

- The Good: likely candidates!
  - Reported with clear rationale and support for why likely
- The Bad: actionable?
- The Ugly(-*ish*): beginning, not end
  - This new beginning has great potential
  - Snapshot of today, tomorrow will be different

Other Concerns/ Opinions:

- Missed known variants
- 3rd hit not confirmed (?)
- Only “top” 3 reported
  - Unreported:
    - known deleterious mutation (by nature) in unknown gene
    - Unknown variant in known gene
    - Parameters for defining relevance not defined
      - Ex: Expression?, Disease?, etc?
    - Actual coverage not disclosed
- Waive the rights to receive raw data (but changed upon complaint)

Other Concerns/ Opinions:

- Difficult/impossible to interpret some technical aspects of how the test was conducted by report alone
  - Filter order doesn’t fit logic
  - Variant number doesn’t match filter description
  - Filter parameters are not well defined
  - Without listing variants, can’t use deductive reasoning to guess

Upon what evidence is the decision made to use large-scale sequencing over a more targeted approach?

- Targeted approaches did not yield answers
  - Single gene
  - Panel
- Answer would be useful for health of self and offspring

Does reimbursement play a role in ordering a whole genome test?

- Yes (unfortunately)
- Opinion: If test is being conducted for diagnostic purposes, insurance should cover as it would for other diagnostics

How do you see the role of patient preferences in what testing is done and what information is disclosed?

- Patient opinion is important!!

Patient preferences in what testing is done (and disclosed)

- Patient opinions can be based on other influencing factors:
  - Ex: Test result could also influence health of potential offspring, therefore time is limited for an answer to be useful for pre-conceptive consideration.

Patient preferences in what information is disclosed

- Patient data = patient choice

- Patient should not be denied data
  - Need for portability
    - Change in provider
    - Change in specialist/field
    - Lab goes out of business
  - Need for updates

How well do patients understand discussions about genomic testing?

You are welcome to ask to find out!

- Molecular techniques, cloning
  - Novel protein design and engineering (chimeric, tagged, catalytically inactive, etc)
  - cDNA

What is the patient response to reports of incidental findings?

Patient data = Patient decision

Full disclosure desirable for preventative care

- ACMG recommendations:
  - Disclose results
  - Opinions:
    - Good intensions
    - Frequent updates/expansions desirable
    - extension of preventative medicine

Incidental Findings

None confirmed

- Later onset
  - Not guaranteed, but useful for prevention
  - A potential hit could carry a “secondary finding”
- Carrier status
  - Variants missed
  - Following up means Sanger sequencing twice, when useful answer could be gotten with one (1x = less money, time)

Post-result state of mind

Positive!

- Likely candidates **are** really likely candidates
- Even if results don’t yield a definitive answer today, it might be definitive tomorrow.
- Having the potential to have an answer, if not today, but maybe tomorrow, is a much more promising position to be in. It offers hope of an answer at anytime to a scenario which previously had exhausted all potential.
- Possibility can equal optimism.

Thank you!