PSYCHOLOGICAL IMPACT OF GENETIC RISK

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DISCLOSURE OF INTEREST

None
QUESTION TO THE AUDIENCE

In which of the following scenarios related to somatic and germline testing might oncologists feel LESS confident?

1. Ability to interpret somatic (tumor) genomic results
2. Ability to support psychosocial support to cope with a somatic alteration with adverse prognostic implications
3. Identify a family history of a potentially inherited condition
4. Counsel an individual to decide whether or not to have presymptomatic genetic testing
5. Provide psychosocial support related to coping with a genetic test result
Oncologists’ somatic and germline genomic confidence (n=27)

- Ability to interpret somatic (tumor) genomic results
- Ability to explain somatic genomic concepts
- Identify a family history of a potentially inherited condition
- Ability to provide psychosocial support to cope with a somatic alteration
- Obtain informed consent to evaluate an inherited cancer risk syndrome
- Counsel an individual to decide whether or not to have presymptomatic genetic testing
- Provide psychosocial support related to coping with a genetic test result

Gray et al. Genetics in Medicine (2016)
WHY WOULD A PERSON UNDERGO GERMLINE GENETIC TESTING?

GERMLINE GENETIC TESTING IN CANCER

- Familial cancer risk
- Early detection and prevention
- Targeted therapy
- Psychological impact
KNOWN PREDICTORS OF PSYCHOLOGICAL IMPACT AFTER SINGLE GENE TESTING

Positive result (van Roosmalen, 2004)
Previous cancer history and baseline cancer worry (Bosch, 2012)
Having children (Arver, 2004)
Cancer risk perception (Lebel, 2003)
First degree relative deceased with cancer (Meiser, 2005)
FROM SINGLE GENE TESTING TO MULTI GENE PANELS

Image from http://www.health.am

From phenotype oriented testing… …to multi-gene cancer panels
From phenotype oriented testing…

…to multi-gene cancer panels

Change in genetic counselling model?

Different psychological reactions?
CHALLENGES OF MULTIGENE CANCER PANEL TESTING

UNCERTAINTY

Han, 2017
UNCERTAINTY

1. PROBABILITY

[Diagram showing probability scale from Impossible to Certain with corresponding chances]

[Diagram illustrating 1000 women with 10 having breast cancer, 990 not having breast cancer, and the distribution of those who have breast cancer and test positive or negative, or do not have breast cancer and test positive or negative]

[Mutations in BRCA1 and BRCA2 genes with percentages: 55-65% for MUTATED BRCA1, 45% for MUTATED BRCA2, and 12% for NORMAL BRCA]

[Link to cancer.gov/breast-fact-sheet]
UNCERTAINTY

2. AMBIGUITY

Taber, 2015; Newson, 2016
UNCERTAINTY

3. COMPLEXITY
WHICH CONTENT AND QUANTITY OF INFORMATION IS TO BE SHARED WITH THE PERSON?
Genetic counseling is the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease.
GENETIC COUNSELLING:
PATIENT’S HOPES AND EXPECTATIONS

Unrealistic expectations

Understanding genetic testing limitations

DID MY GENETIC TESTS COME BACK?
Yeah, sit down.
IS IT BAD NEWS?
NO, WHAT ARE MY RISK FACTORS?
WE CAN’T BE SURE ABOUT THIS, BUT WE’VE ANALYZED GENES ON SEVERAL OF YOUR CHROMOSOMES, AND IT’S HARD TO AVOID THE CONCLUSION:

AT SOME POINT, YOUR PARENTS HAD SEX.
Oh, God!
STAY CALM! IT’S POSSIBLE IT WAS JUST ONCE!

I... I NEED TO BE ALONE.
Utility of family history: health beliefs, communication patterns, create rapport with the person, support family communication

Ormond, 2013; Bennett, 2004
BRCA2 MUTATION: THERAPEUTIC DECISION FOR THE PATIENT
BRCA2 MUTATION: PREDICTIVE TESTING IN THE FAMILY

Offspring’s risk, cancer risk assessment, early detection, prophylactic surgery

PBSO    PBSO
Occult primary Fallopian tumor
New paradigm in medicine

Teaching model: Directive
Disease-Based, Doctor-Centered Medicine

Counseling model: Non-directive
Patient-centered Medicine
Conversational model

Key themes (tier 1 elements) in the tiered-binned model

1. Testing can identify varying risks (high, moderate, and uncertain) for a wide spectrum of cancers.

2a. Implications of results vary depending on the gene and result.

2b. Finding a mutation may or may not change your current medical care.

3. The evidence and support for medical recommendations varies by gene and result (e.g., in some cases there are evidence-based recommendations, others consensus guidelines, others no clear consensus or guidelines).

4a. Some genes are associated with risks for cancer in childhood, while others only confer risks in adulthood. Some genes are associated with disorders other than cancer.

4b. For some genes, there is currently insufficient information to recommend testing for relatives.

5. There is the potential for various uncertainties (e.g., uncertain results, uncertain risks, and uncertainty regarding medical management options).

6. Given a range of testing options and variable risks, benefits, and utilities, patients should be aware that testing is a choice.

7. Risk estimates and cancer spectrum may be revised over time. Ongoing communication with the cancer genetics team will be beneficial, particularly in the setting of uncertain results or recommendations.

Bradbury, 2016
This subscale evaluates adverse psychological reactions about one's test result such as sadness, anxiety, nervousness.

MICRA Distress (range score 0-30) after multigene panel testing

Irene Esteban et al, Psychooncology 2018
TAKE HOME MESSAGES

- Challenge of multigene cancer panel is potential uncertainty
- Recognize the content and quantity of information to share with each individual is key
- Information alone is not enough to make decisions: autonomous decision making is socially contextualised, influenced by emotional and situational issues
- A conversational model with the individual might help to satisfy his/her needs, adjust expectations, and promote deeper understanding
THANK YOU!