Young cancer patient perspectives on undertaking whole genome sequencing: A qualitative study

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Background

Whole genome sequencing (WGS) is moving into clinical practice, with the goals of identifying gene variants that increase individuals’ risk of disease and guiding prevention strategies. However, little is yet known about the ethical, psychosocial and behavioural implications of WGS. The Genetic Cancer Risk in the Young Study (RisC) is recruiting 1,000 young cancer patients and blood relatives to undertake WGS to investigate heritable genetic disease drivers. A young age at diagnosis increases the likelihood of inherited predisposition.

Aims

The current psychosocial sub-study (PiGeOn) aims to explore participants’ motivations, understanding, experiences and views about WGS longitudinally. Baseline proband qualitative results are reported here.

Methods and sample

Purposive sampling was used to ensure diversity in cancer types and demographics. Eighteen participants with a cancer diagnosis under 40 years old or multiple cancers diagnosed before 60 years of age participated in a 30 minute semi-structured interview via telephone. Seventy-two percent (13/18) of the participants interviewed were female, and participants ages ranged from 32 to 78 years. Framework analysis was used to analyse the data.

Findings

Motivation

Participants were curious about their own genes, felt a sense of responsibility to family and wanted to contribute to science and help others in a similar situation. They viewed participating in the study as seizing a one-off opportunity to gain knowledge.

“because I already have been through the whole process twice, I’ve sort of learnt to do things for the here and now and so I can’t really do much about it, but if there is something I-I can, just by eating a bit more healthy or, I mean, I wouldn’t go out and get a full, sort of, hysterectomy or anything like that but, um, for the right here and now, I’m okay.”
Female, 37, brain

“because if it helped researchers understand how people that don’t have history or younger children or whatever, um, get different types of cancer, then you know why wouldn’t you participate in something like that.”
Female, 39, breast cancer

Emotional readiness

Participants felt emotionally prepared for WGS due to their previous experience with genetic testing and the belief they would be able to cope with results, whether they be positive or negative. They also felt that WGS results could give them information to change their lifestyle and reduce their uncertainty about future health risks.

“Not like I think about it every day... but I am aware of the consequences, should I decide to know, I’m aware of the consequences and then I don’t know how I feel about it, like there are some days where I think, well that would kind of cool to know, but then there are other days when I think... whatever happens, happens and then I’ll deal with it when it comes... I think I’m a bit in two minds about that.”
Female, 40, breast

Autonomy

Participants recognised their results could have implications for other family members, but family culture influenced the degree of engagement with family members prior to consenting as well as who they planned to pass their results onto.

“if my daughter were to be tested in the future just for her to have the knowledge, would help her perhaps to find, um, you know, to be in control a little bit more control of her own health and sort of maybe pick up on those earlier signs and perhaps not get a horrific cancer diagnosis.”
Female, 34, ovarian and pancreatic

Understanding

Participants often lacked full understanding and desired lay-appropriate explanations, but felt embarrassed to request this type of information.

“any information that can be learned about cancer is going to help hopefully, help people, um, downstream with treatments and detection so, um, however it can help is good information gathering”
Female, 37, colorectal

Discussion

Young cancer survivors see WGS as an opportunity for the individual, family and wider community to reduce future risk. Participants had high hopes of actionable results. A cancer diagnosis may motivate patients and family members to be more active in managing their risk, but may need interventions to promote better understanding and communication within the family about WGS results. Most participants considered WGS to be beneficial and found the experience of testing to be positive. Support may be needed with family communication about testing and results.