Conversations about Genes

navigating personalized DNA testing and sequencing

The following guide is designed to assist community-health professionals working with vulnerable populations navigate the complex arena of personal genetic testing and DNA sequencing, otherwise known as personal genomics. It is recommended that staff using this guide, complete the online course or ½ day workshop entitled "Personal Genomics Today: Basics, Issues and Controversies."

What is Personal Genomics?

Personal Genomics is the collection and testing of biological material for the purpose of DNA sequencing by an individual outside of regular medical systems. The individual secures the testing usually from a private company and / or research institution directly, foregoing the intervention of medical personnel. The field is rapidly expanding and an increasing number of individuals and families are finding themselves able to afford these services. As costs decrease and testing simplifies, it is estimated that the number those seeking personal genomic services will increase.

When helping a client decide whether or not personal genomic testing is right for them, a number of key factors should be considered. As a community-health professional, it is recommended that you facilitate conversations with your client not only about the medical and scientific context of gene testing, but the personal and social contexts as well. Below are some ideas to get you started. You should not only facilitate the client's exploration of their decision, but also be prepared to offer critical input and / or "devil's advocate" positions to get them to unravel their decision-making processes and influences.

What are the motivations and expectations?

Individuals have many reasons why they want to have their DNA sequenced. It is important to understand what motivates their interest and to assess whether or not they have realistic expectations. Some questions you should consider asking are the following:

"How did you hear about personal genetic testing?"
Assess whether or not the client has responded to an advertisement from a testing company, have a friend or family member who has had their DNA sequenced, or found the option on their own. Each of these sources will have a slightly different influence on the person's decision and it is important to understand the originating source of the idea to engage in genetic testing.

"Why do you want to get this testing done?"
Here it is important to listen for outside pressures or influences that may be acting upon the client. As with any medical procedure, it is critical that the individual fully own the decision to proceed devoid of outside coercion or inducements. Also, it important to understand the key goal (i.e. medical risk, ancestry identification) the individual seeks.

"What do you think the possible outcomes might be?"
Genetic testing is often presented as a technology that is infallible and whose determinations are rock solid. As you understand, this is a bit of a myth and genetic results are complex interpretation of

complex biological systems that are also influenced by environmental context. When questioning your client about outcomes, you should try to understand to what extent the client understands the grey, rather than black and white, nature of this testing.

What is known about the testing facility?

There are a number of private and academic research facilities that provide personal genetic sequencing. No two facilities are the same and their primary interests influence the policies, procedures and use of genetic material collected by that facility. It is important that your client have an understanding of factors that impact testing facilities. Some questions you should consider asking are the following:

"Who is the key individual or institution involved and how easy is it to find information about them?"
Sometimes it is not always easy to know who is behind a personal genetic testing service. There are several projects originating out of major research institutions and research communities (i.e. AfricanDNA, The Personal Genome Project). Other services are connected to large pharmaceutical concerns. It will benefit your client to understand who "owns" or "controls" the testing service since this may influence the policy and procedures in place.

"What is the company's main objective in offering DNA sequencing?"
It is also important to understand what the driving force behind the service is. Is it to :

- further scientific or academic research agendas?
- provide a valuable service to the public?
- build up genetic information for future drug products?
- find new connections between social and racial groups?
- reconnect individuals to their past?

Being able to answer the above will help the client understand their role in providing their DNA. In many instances, your client is rarely just a "consumer." They should understand what other roles they are playing in the process.

"Where do they secure their funding from?"
Like understanding the individuals involved it may also help your client to understand what business partnerships or funders are behind the service. These financial backers exert influence as well and your client should understand to what extent the

What is involved in the collection and testing of the DNA?

Each testing facility has a different method for collecting and handling DNA samples and not every test is the same. Often, clients may feel that they will get a "definitive" answer to some health or personal question. You should work with them to understand to what degree they understand the limitations and reliability of DNA testing.

"What are the procedures that will be used to collect the DNA and how do you feel about them?"
Collection of DNA material can be as simple as mouth swabs to blood tests to skin scrapes. To what

extent does your client understand how collection of the biological material will be handled and by whom? Is it self collection or by trained professionals? What are the pros and cons of various collection methods? You should also be on the look out for apprehension, fear, or discomfort in this conversation which may indicate that certain methods are not appropriate for this client.
"What is sort of DNA sequencing test will be preformed and what do you know about this test?"
There are a range of DNA tests that are conducted and personal gene testing services tend to offer a limited selection or narrowed scope of sequencing. Does your client understand the pros and cons of the tests they are considering? Do they feel comfortable in their grasp of the scientific knowledge required to understand the tests? What strategies do they have for filling in any gaps they may have?

"What do you know about the reliability and limitations of the DNA test you will have done?"
Often the public perception is that DNA tests are infallible. However, we are complex biological beings and genes are one element that influence our health and ancestral paths. You should guide your client through a conversation about some of the issues related to DNA testing. They should get a sense of the state of DNA testing and scientific knowledge. They should be sensitized to how environment and social contexts come into play.

How will privacy and security be handled?

Most personal genomic services have some statement about privacy related to the biological sample, personal information, and data related to you DNA sequencing. Given the sensitive nature of this information it is important that your clients consider how much privacy means to them and how much openness they are willing accept.

"Who will be handling your sample and have access to your DNA information?"^
Depending on the service, your DNA sample may be handle by a central organization or company that then forwards the sample to a laboratory or other facility. How does the facility ensure that they keep track of samples and coordinate the sample with your identifying information? What is the loss and error rate of the facility and what protections are available to participants should an error be made?

"How secure will your sample and information be stored?"
Given that personal information like DNA can have affects for employment, insurance, and even family relationships security may play an important role for your client. You should assess who they feel comfortable sharing their DNA information with. Close family members? employers? researchers? insurers? lost or unknown blood relations? strangers with the same genetic heritage? for-profit commercial entities? These assessments of personal security requirements should be gauged against the assertions for protection offered by the testing service. It is also important to have the client understand that in some instances privacy may be impossible and that their information may be shared with unintended third parties. They should understand what their modes of redress are should their privacy expectations are breeched.

"Does the facility have in place any partnerships or agreements that would mean your sample and / or information will be shared with third parties?"
Many testing facilities share their aggregated DNA information with third parties such as research colleagues, pharmaceutical companies or even a third party individual who may share similar genetic heritage. The client should be aware of whether or not the facility they have chosen has any exchange relationship of this type in place. Your role is to understand how well they understand these

arrangements to assess how they might react should information get shared in ways they are unfamiliar with.

What are the personal risks?

DNA testing is not without risks. Often those who approach genetic sequencing from a personal motivation outside of medical necessity are often exploring personal curiosity, seeking ancestry information or may be trying to assess their likelihood of future disease. Results may come back in a manner that implies certitude with information that may be devastating to the client. Here are some questions to help clients assess risk.

"What do you think are the risks to you have as a result of conducting this test?"
Has the client considered what might happen if a negative or disruptive result comes back form the test (i.e. high likelihood of a terminal disease, family background that challenges identity)? Helping the client understand how much ambiguity is still possible with these tests may prove to be helpful. Have they considered that unintended legal or financial relationships may result form the test (i.e. higher insurance premiums, identification of paternity)?

"What are your strategies for handling confusing or disappointing results?"
One way to help clients prepare for unforeseen risk is to help them think through coping strategies. Providing resources and scenario role-playing are two methods that may be of help.

How might this information be used?

Like the eugenics movement of earlier days it is useful for your clients to understand how their personal information might be used to support a range of ideological positions. How might racial or gender biases is linked to specific genetic markers? How might this information be used to discriminate or oppresses certain subsets of the population? Does your client have the ability to decode and respond to potentially damaging social responses to their genetic information?

Next Steps

It is likely that through your conversations with your client several gaps in information or vague areas arose. It is also possible that some new ideas or information surfaced that created uncertainty or agitation. It is recommended that you schedule a second session with the client to follow-up. You may also want to share some additional resources for the client to consider. A sample list has been provided with this guide, but you should collect additional materials that are culturally and linguistically sensitive to your constituent base.

Resources

Bandelt, Hans-Jurgen, Yao, Yong-Gang, Richards, Martin, and Antonio Salas (2008) “The brave new era of human genetic testing” BioEssays 30, 1246-1251

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