MARQUETTE UNIVERSITY
Office of Research Compliance

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Issues to Consider When Conducting Research Involving Genetic Testing

As a researcher who will be taking samples for genetic testing, make sure to address the following areas in your protocol form and consent document. Depending on the type of genetic testing you are conducting some of these issues may be more or less relevant.

Issue	Sample Consent language
Specificity of the testing- the researcher must balance between being too general describing the testing and being so specific that any change in testing procedures would necessitate a reconsent of the subjects. Ideally, the description of the testing should focus on a specific disease, a non-commercial or non-profit use of the specimen and/or the medical research use of the specimen rather than studies of origin	"DNA will be extracted from the specimen you provide and will be tested for markers of XX disease" "Your DNA sample will be used only for medical research and will not be used for commercial development or studies of origin"
Will results be returned to the subject? Typically, only done if • results are confirmed by a CLIA certified lab • results are clinically significant and clinically actionable • resources should be in place to support subject • Subjects should be given the option to decline receiving any results	"In generally we will not give you any individual results from the study of the samples you give us. If we find something of urgent medical importance to you we will inform you although we expect that this will be a very rare occurrence" "Please check here if you would prefer to not receive any results of the testing".
How will you handle incidental findings (findings you were not testing for but that were discovered during the process)	"It is possible we will discover that you have a gene variant that is unrelated to the purpose of this study. If we believe that the information is of urgent medical importance we will share this information with you. You should not assume that if you are not contacted that you do not have any gene variants that might be related to a disease."
Will you be keeping the samples/extracted DNA in a bank for future studies? • If so, subject needs to know the details- for how long, what they will be used for, how/when they will be destroyed • Consider making this an option they can opt-into	"Your samples, genomic data and health information will be stored and may be shared with other researchers for use in studies that have been approved by an Institutional Review Board. Samples that are shared [will/will not] contain identifying information." "May we collect your tissue samples, health information and genomic information to study [specific project]?" "May we share your tissue samples health information and genomic information with other researchers to study [specific disease or disorder]?" "May we share your tissue samples, genomic data and health information with other researchers for future research projects related to other topics?"
Should be clear if subjects may be recontacted for consent for future studies	"Researchers may want to ask you to participate in additional studies. May we contact you in the future to get your permission to use your samples, health information and genomic information for additional studies?"

Consider how testing for heritable disease, especially if family members will be tested, may have unintended results.	"You may learn something about your genome that relates to the health of your relatives. If so, your relatives may want to know so they can decide whether to get tested or follow-up in other ways. It is also possible that they might not want to know." "It is possible that we will learn that assumed family relationships are incorrect (such as learning that a child is adopted or has a different father). We will not give you these results/We will tell you these results only if they are relevant to your health."
Studies involving children • As in any other study, children who turn 18 during the course of a research study must give consent to continue participating. • How will you handle testing for adult-onset conditions?	"As part of the study, your child's samples, genomic data and health information will be stored and used for future research. When your child reaches age 18, we will try to contact him or her to ask whether he or she wants to continue to participate in the research. If we cannot find your child we will remove identifying information and continue to include his or her samples, genomic data and health information."

References and resources:

National Cancer Institute:
https://biospecimens.cancer.gov/global/pdfs/NCI_Return_Research_Results_Summary_Final-508.pdf

National Human Genome Research Institute:

https://www.genome.gov/27561533/human-subjects-research-in-genomics/