NICHD IRB review of genomic and exomic sequencing projects

Purpose

To ensure that sequencing projects utilizing new and emerging technology specifically address ethical issues of incidental medical information, data base deposition, confidentiality, and consent/assent.

Policy

1. This policy applies to sequencing projects in which significant data will be obtained concerning genomic regions that are incidental to the disease process being studied. Specifically, this policy applies to exomic and genomic sequencing projects. Prior consent to sequence genes related to a specific disease process does not exempt a project from this review. Since incidental information could affect family members, death of the proband does not exempt a project from this review.

2. Proposed sequencing projects need to be submitted to the NICHD IRB for approval. These submissions could be either a new protocol or an amendment to an existing protocol. If an investigator has a waiver from the Office of Human Research, then a memorandum briefly describing the project and a copy of the waiver will suffice.

3. Investigators need, specifically, to address the following:

   a. **Incidental medical information.** Incidental medical information is sequence data unrelated to the disease of interest. For example, the identification of a deleterious *BRCA1* mutation in a subject being investigated for cognitive impairment, or identification of a polymorphisms that may influence risk for diseases other than that specifically being studied. The protocol should specify how incidental medical information that has potential health impact on either the subject or the subject’s family will be handled.

   b. **Database deposition.** Investigators should address the issue of deposition into databases of sequence data that contain sufficient information to identify a participant. Applicable NIH policies regarding data deposition should be addressed, as should patient/family confidentiality.

   c. **Consent.** Obtaining specific consent for this degree of sequencing is preferable. If an investigator is unable or elects not to obtain specific consent, then he/she needs to provide a justification, a plan to mitigate risks, and establish that this project is consistent with prior consent.

4. This policy will be superseded by any policy developed and implemented by NIH Office of Human Research.