**Panel Summary**

**Ethical Challenges Associated with Enrolling Families in Genetic/Genomic Research**
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The February 2019 Ethics for Lunch considered the ethical challenges that can arise when enrolling families in genetic/genomic research. The panel discussion created a forum for attendees to learn about the importance of enrolling families in genetic research and to understand the issues that researchers face when enrolling individuals from the same family into a research study. The panelists provided examples of these issues from ongoing studies. A case study was presented to highlight the challenge of returning results to multiple family members enrolled in a genomic sequencing study.

Highlights of the panel discussion included:

1. A brief overview of the importance of enrolling families in genetic research studies. From the earliest pedigree research studies to the large-scale genomic sequencing studies common today, the inclusion of multiple members of the same family allows researchers to study the inheritance pattern, age of disease onset, symptom variability, and genomic factors responsible for an inherited disease/condition.
2. A description of the ethical challenges that can arise in genomic research as a result of the inherent tension between individual autonomy and the familial nature of genomic information. The panel outlined the tensions across the research life-cycle:
	* Recruitment: individual decision to participate or not in a research study vs. the need to enroll family members in order to conduct the study
	* Informed consent: individual informed consent vs. the family unit that may bear the risks and benefits of study participation; voluntary informed consent vs. family pressure to enroll
	* Return of research results: individual research result vs. familial information; individual right to learn genetic information vs. family members’ right not to know
	* Privacy/confidentiality: individual privacy vs. duty to inform/warn other family members of genetic risk
3. The panel outlined the factors that are fueling this decades-old issue, including the declining cost of genomic sequencing, the challenge of incidental findings, and the explosion of the commercial genetic testing industry. Large-scale genomic sequencing is relatively inexpensive, allowing for its widespread use in research, clinical, and recreational contexts. Genomic sequencing can now provide information on not just one gene, but on thousands of genes, and has led to the challenge of managing “incidental” or “secondary” findings (i.e. genomic findings of potential clinical significance outside the reason for research/testing). In response to patient/research participant interest in genomic information, research studies are increasingly returning genetic results to participants, results that can have implication for family members.
4. The panel presented data from ongoing research studies showing that individuals are motivated to participate in genomic research studies to benefit their family members and that individual decisions to participate (or not) in genetic research study can have long-term impacts (both positive and negative) on family relationships decades after testing.
5. A discussion of a case study in which a family of five (mother, father, daughter, affected son, and paternal aunt) enroll in a genomic research study in an effort to identify the cause of the affected son’s condition. All five family members (mother, father, daughter, affected son, and paternal aunt) individually consented to participate in the research and make choices regarding receiving study results. All five family members opted to receive findings related to the condition for which their family has enrolled (i.e., the boy’s condition). However, only the aunt elected also to receive results for other serious conditions as well.
	1. In the first scenario, the audience was informed that the researchers identified the cause of the boy’s condition – a recessive genetic disease that he inherited from both parents. The audience was then asked who in the family should receive the result. The scenario generated a lively discussion as to whether or not the aunt was entitled to learn the boy’s result. Opinions were divided. Some attendees believed that the result belonged to the parents of the young boy and it was the responsibility of the parents to share the information with the aunt. Others believed that since the aunt consented to be enrolled in the study and provided a blood sample, she was entitled to learn the boy’s results as they pertained to the condition for which she chose to enroll.
	2. The panel then discussed a different version of the same scenario, in which the researcher did not identify a cause for the boy’s condition, but instead found a mutation of a gene that can cause unexpected, sudden cardiac death in otherwise healthy individuals. The condition, called long QT syndrome, can affect both adults and children and can be treated. In this scenario, the father, daughter, and aunt were found to carry the mutation. Again the audience was asked who should receive this research result. This example sparked a lively debate and a range of opinions on what researchers should. Some believed that the researchers should respect the family’s choices for results and only return the information to the aunt. Others believed that the researchers should override the individual consent choices and return the information to each of the family members at risk (father, daughter, and aunt). The factors that appeared to influence opinions in favor of returning results to all of the family members at risk included the severity of the condition, the availability of treatment, the age of the daughter (a minor), and the researchers’ duty to warn the family of their potential risk.

Today, existing research and ethical guidance centers on the individual research participant; however, genomic research frequently enrolls family members. The question of how investigators should balance their duties to research participants from the same family, particularly when family members make different choices for results, remains unanswered.

**References:**

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