The Perspective of a Sub-Saharan African Cleft Cohort on the Disclosure of Secondary Findings from Genetic Research

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Background: A scarcity of knowledge exists on the perceived risks and benefits of Whole Genome Sequencing (WGS) in minority populations. WGS has the potential to help unravel the diagnostic odyssey associated with diseases presenting with a complex aetiology, including Oro-facial clefts (OFCs). Also generated with WGS data are secondary findings (SFs) of potential health importance, which may burden the patient and their families. Thus, this study aims to evaluate patients' perspectives in OFC studies regarding SFs and their choices in the return of results (RoR) from genetic testing.

Methods: An 87-question online survey was used to assess the perspectives of parents of children attending participating cleft clinics in Ghana and Nigeria via two domains; the willingness to receive information regarding SFs and available resources to obtain information and follow-up regarding any positive SFs**.**

Results: A total of 197 responses were recorded. 27.9% were familiar with genetic testing, and 24.9% were confident in understanding genetic information. While the majority (88.8%) lacked access to resources that could enable them to obtain the needed genetic information, those with access cited physicians as their primary source (64%). Overall, 95.4% of the participants were willing to receive all the information gotten from genetic testing (including SFs).

Conclusion: Majority indicated a strong desire to receive SFs. However, access to genetic information was cited as a barrier to the return of results (RoR) in this cohort. Our results also indicated the need to support patients as they receive results and share them with their families. Overall, patients' preferences around the return of SFs could inform the development of educational materials and decision support tools to guide patients and providers through the process of RoR. Also, findings from studies like this could improve the understanding of the personal utility of genome sequencing & inform practice guidelines/policies related to RoR.

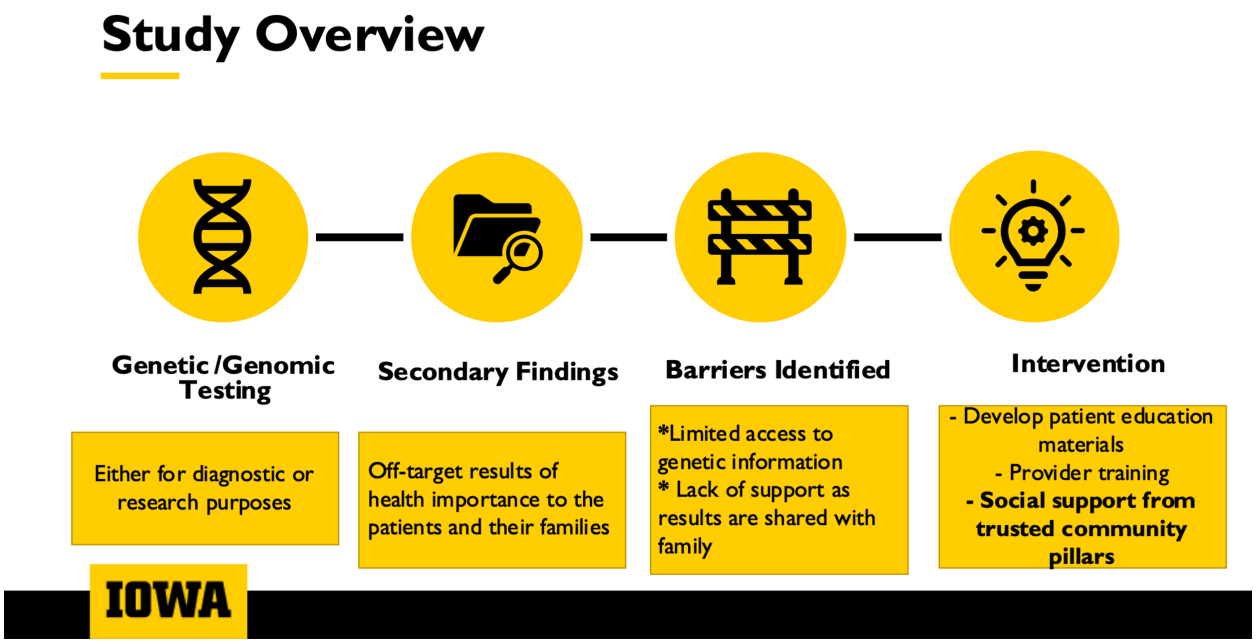

Figure 1: Study Flow Diagram

Familiarity with genetics	%
I am confident in my ability to understand information about genetics.	22.9
It would be easy for me to get information about genetics if I wanted to.	20.7
I would be able to understand information about how genes affect my or my child's health.	37.8
I would have a good idea about how genetics may influence risk for disease generally.	27.4
I would be able to explain to others how genes affect health.	14.1
I have a good idea about how my own genetic make-up might affect my risk for disease	12.6

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Limitations: The COVID-19 pandemic slowed down the response return rate due to the risk-mitigating measures implemented across the participating clinics. The civil unrest in Ethiopia had similar effect.

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