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| **eMERGE Network: Proposal for Analysis**  Project/Manuscript Concept Sheet | |
| **Reference Number** | NT277 |
| **Submission Date** | 3/28/2018 |
| **Project Title** | Operationalizing participant choices about genomic results: Beyond all or none ACMG recommended genes |
| **Tentative Lead Investigator** *(first author)* | C. Hoell & C. A. Prows (joint 1st authors) |
| **Tentative Senior Author**  *(last author)* | L. Rasmussen & K. Marsolo (joint last authors) |
| **All Other Authors** | M.F. Myers, M.E. Smith; site investigators involved in operationalizing participant choices – S.A. Aufox; investigators at central laboratories – H. Zouk, E. Venner, Y. Yang, D. R. Murdock, Y. Jiang |
| **Sites Involved** | CCHMC, Northwestern, LMM, Baylor, other sites where participants were offered choices about secondary results (all or none –v- some of the ACMG 56/59 genes) |
| **Background / Significance** | In clinical practice, patients are typically given the option to learn all or zero secondary sequencing results recommended by the ACMG. As the field becomes more comfortable with the return of sequencing results, and we move towards more patient-centered care, it is necessary to provide patients with more granular choices in deciding what to learn about their panel results. There is considerable variation between sites regarding study participants’ engagement in prospectively choosing the type of e3 sequencing panel results they want to learn. Investigators at several eMERGE 3 sites were challenged to create tools that enabled participants’ granular choices. |
| **Outline of Project** | This project will describe the tools created at sites and the EHR accommodations necessary to return some but not all results for genes analyzed and reported by the eMERGE 3 central laboratories.   1. Describe how laboratories manage individual choices for incidental / secondary results 2. Describe tools desired / envisioned and actually created and used for patient preferences.    1. Rationale for creating tools    2. Reasons for differences between desired and actual tools    3. Successes, challenges and limitations of using tools from both the clinical and the laboratory perspectives 3. Describe sites’ accommodations to EHRs and/or modifications made to central laboratory reports    1. Automatic / manual accommodations? 4. Estimated person hours required to operationalize participant choices with tools and EHR / report accommodations 5. Recommendations for future tools to automate / facilitate individualized genomic sequencing results |
| **Desired Variables**  *(essential for analysis*  *indicated by* ***\*****)* | 1. Type of choices offered at each site 2. Type of tools created at each site to accommodate choices 3. Estimated person hours to create and implement tools |
| **Desired Data** | Narrative and descriptive data of variables listed above |
| **Planned Statistical Analyses** | Content analysis and descriptive statistics |
| **Ethical Considerations** | None |
| **Target Journal** | GIM or JAMIA |
| **Milestones\*\*** | Approval – Spring 2018  First draft – Fall 2018  Submit for publication – Winter 2018 |

***\*\**** *This section should include the timeline for completion of project, including: approval, project duration, first and second draft of the paper and submission.*