Abstract

Poster Title: Creating and Evaluating a Parent Co-Developed Resource for Genomic Testing Results

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Abstract

Genomic testing is a powerful tool, but the complexity of the testing process and results causes stress to many families. Families report feeling "abandoned and lost" after receiving genomic test results. Our team's goal was to co-create a simple, customizable tool to help families understand genomic test results, appreciate their implications, and navigate available resources.

In collaboration with parent-partners, we chose a customizable, printable e-booklet format with three sections: background, individualized genomic test results, and avenues for support. The language is family-user friendly, yet the content is rich and explanatory, as requested by the parent-partners. The tool facilitates result individualization by physicians and genetic counselors through a series of dropdown menus, check-boxes and editable text boxes. Furthermore, it serves as a practical take-home resource for families that are receiving genomic results, thus extending clinical support.

The booklet was edited in response to feedback from genetic physicians, genetic counsellors, and patient-partners, after which the final version was provided to families and evaluated in a genomic study of atypical cerebral palsy (the IMAGINE study). Ongoing survey and interview evaluations target acceptability and usability for both genetics professionals and patients.

Initial evaluations from family users within the IMAGINE study were universally positive about layout, content, ease of use, and potential to support families during and after sharing genomic results. Notably, 89% of respondents felt that the booklet reinforced information received during their post-testing appointment, while 66% of users anticipated they would refer back to the booklet again in the future. Moreover, the booklet was deemed to be appropriate and supportive given the emotional and stressful nature of receiving medical testing results. Evaluation of the booklet will continue throughout the IMAGINE study, with feedback anticipated from additional families.

Preliminary results demonstrate that the booklet is meeting the needs of families while enabling genetics professionals to share results in an efficient, personalized manner that supplements in-person genetic counselling. We intend to make this tool available for research and clinical use beyond the IMAGINE study.