Development of an Interactive Patient Education Tool for Genetic Testing in Autism Spectrum Disorder

Maurita Hung[1], Michael Corrin[1], Shelley Wall[1] and Ny Hoang[2,3,4]

[1]Department of Biomedical Communications and [2]Department of Molecular Genetics, University of Toronto, Toronto, Canada
[3]Department of Genetic Counselling, The Hospital for Sick Children, Toronto, Canada
[4]Genetics and Genome Biology, Research Institute, The Hospital for Sick Children, Toronto, Canada

Abstract

In recent years, technological advances have enabled **genetic testing** to become integrated into clinical practices in the field of **autism spectrum disorder (ASD)**. However, there is a lack of awareness on the genetic basis of ASD, the benefits of genetic testing and the outcomes that can be expected. This research project aims to bridge these communication gaps by creating an interactive web-based resource to teach parents and society about genetic testing for ASD.

Introduction

Autism spectrum disorder (ASD) is a neurodevelopmental disorder, characterized by impairments in communication and social interaction, as well as repetitive behaviours[1]. The cause of ASD is not fully understood, but there is general consensus among the scientific community that it is a **multifactorial disorder** with both genetic and environmental contributing factors[1-3].

The purpose of **genetic testing** in ASD are twofold, with **clinical- and research-based benefits**[2]. First, it may be used to identify other medical issues and genetic disorders that are associated with ASD, leading to medical management changes[2]. This information can inform recurrence risk estimates for parents and other family members[2].

Second, genetic data obtained from families with ASD may be analyzed by researchers to identify additional genetic variants that contribute to ASD risk, leading to a better understanding of the genetic etiology behind ASD which may inform earlier diagnosis and individualized intervention for future generations[3].

The objectives of this project are to educate families about the role of genetics in contributing to ASD, the benefits of genetic testing and the possible outcomes of genetic testing.

Materials & Methods

The format of the patient education tool is an **interactive web-based resource,** with the target audience being parents and family members of patients affected by ASD.

1. **Content document**

   A **literature review** was first performed to identify existing patient education resources and methods used to communicate genetic information about ASD.

   An **informal focus group** consisting of genetic counsellors working with an ASD population from The Hospital of Sick Children (n=3) was held to determine the scope of the project.

2. **Storyboards**

   Attended genetic counselling appointments in order to collect more information and revise the content document.

3. **Visual assets**

4. **HTML5 / CSS3 / Javascript coding**

5. **Website launch**

Usability testing will be conducted in the future to improve UI/UX design and to evaluate the effective of the resource in educating the target audience about genetic testing for ASD.

Results

The resource is separated into **four modules** that covers the roles of genes and proteins in ASD, the genetic basis of ASD, the purpose of genetic testing, and the results we can expect from genetic testing.

It will implement the use of **visual analogies and metaphors** to teach complex scientific concepts[2]. In Module 1, a car factory analogy is used to explain the roles of genes and proteins in the brain. In Module 2, a cup and ball analogy is used to communicate the concept of genetic and environmental factors and the risk of developing ASD (**Figures 1 and 2**)[2].

Figure 1. The cup model[2]. In this visual analogy, the cup represents an individual's chance of developing ASD. Blue balls represent genetic factors, while yellow balls represent environmental factors. The size of the ball is representative of how strong a role the factor plays in contributing to ASD. Only individuals with enough genetic and environmental factors (i.e. a full cup) will develop the disorder.

Results

The resource will also employ the use of visual images to accompany the text and to tell a story to engage the target audience (**Figure 3**).

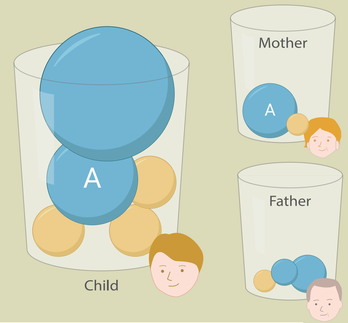

Figure 2. Inherited genetic factors and de novo genetic factors. The cup model is also used to explain the difference between genetic variants that are inherited from the parents (blue ball labelled "A") and genetic variants that spontaneously appear (unlabelled blue ball).

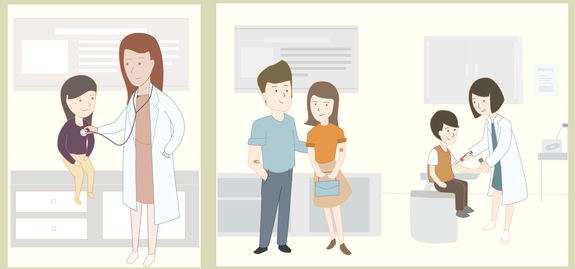

Figure 3. Process of genetic testing. A genetic counselling appointment involves a review of the medical and family histories and a physical exam (left panel). A blood sample may also be taken for genetic testing (right panel).

Conclusions

This patient education tool will aid in knowledge translation in the field of ASD genetics with the use of visual analogies and storytelling techniques. It will improve society's understanding of the genetic basis of ASD, the benefits and limitations of genetic testing, and the possible outcomes from genetic testing.

References

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Note: The design and content of this poster has been adapted for JBC.