The Impact of a Diagnosis among Parents of Children with Severe, Progressive, Life-Limiting Diseases

[a]**Veronica Romines;** [a]**Clara Van Karnebeek,** MD, PhD, FCCMG; [a]**Hal Siden,** MD, MHSc; [a]**Shelin Adam,** MSc; [a]**Patricia Birch,** MSc; [a]**Gail Andrews,** MEd

[a] University of British Columbia, Vancouver, British Columbia / BC Children's Hospital Research Institute, Vancouver, British Columbia

BACKGROUND

Children affected by severe, progressive life-limiting diseases experience a variety of symptoms and disease trajectories that are not well understood or described. Many of these children do not have a specific disease or diagnosis, but are assigned a broad diagnosis that simply describes a cluster of their symptoms (e.g. "Severe Epilepsy").

Given the significant impact the child's life-limiting condition has on the parents, qualitative research was done to examine the parent's experience of living without a diagnosis for their child.

AIM

The study objective was to investigate the impact of a diagnosis on the parents of children with severe, progressive, undiagnosed conditions, who were eligible for genetic testing.

METHODS

Eligible children had neurological impairment, limited communication, full-time caregivers, typically mobilized in wheelchairs, and were between 0-20 years old.

Six families (6 mothers and 1 father) were given a semi-structured interview with a genetic counselor in order to understand how finding a diagnosis would impact their life. The interview data was transcribed and coded into themes using a grounded theory approach. The parents also completed 3 brief questionnaires to provide more insight into their experience.

In addition, an option was provided to the families to enroll in TIDEX to perform genome-wide sequencing that might identify a diagnosis for the affected child of these families.

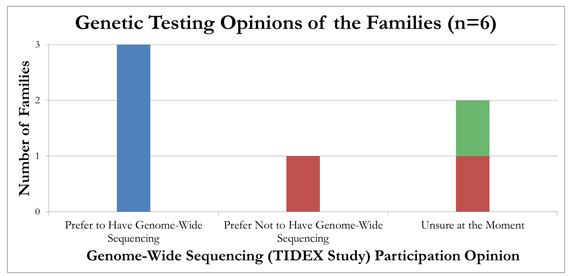

Figure 1

RESULTS

Children ranged in age from 5-20 years, with an average age of 11.5. The average number of years since their symptoms first appeared was 11.2 years. There were 5 boys and 1 girl.

Importance of Diagnosis

A majority (83%) of parents reported that **a diagnosis was of little or no importance to them**.

Two-thirds (66%) of parents indicated that **there was a time when a diagnosis was more important to them, and referred to the time of their child's infancy.** Three of these four parents also stated that the time they reached acceptance for not having a diagnosis for their child was in the **first two years.**

It was also found that **50% of parents did not perceive there to be a challenge with not having a diagnosis**.

> **"I feel like I'm not looking for an answer. My questions are answered. And I don't know…would it change anything?" (1008-2-7)**

Impact on Resources

Five out of six parents **felt they had enough community support already** and they did not believe it would be impacted by a diagnosis (Figure 2).

All six parents believed that there would be **little or no impact on school resources caused by receiving a diagnosis** (Figure 2).

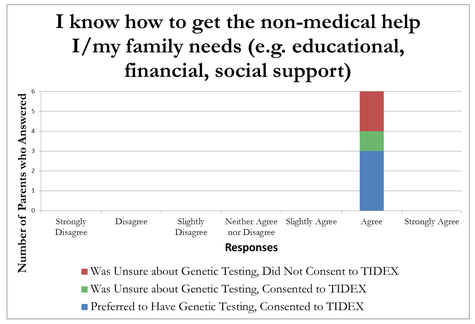

Figure 2

Reasons for Wanting a Diagnosis

I. 50% of the parents felt that **curiosity played a role in them wanting a diagnosis**, but all of these parents also admitted that curiosity was of low importance for diagnosis.

II. 50% of the parents thought **a diagnosis might be of some value when considering other family members' reproductive planning** (Figure 3).

III. A majority of parents (66%) also thought that **a diagnosis could potentially allow them to prepare for the future** more or envision a more accurate future trajectory of their child. (Figure 4).

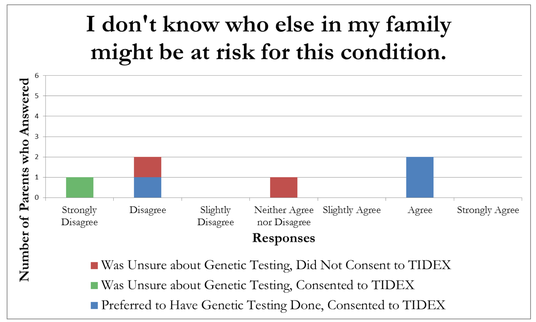

Figure 3

> **"Being able to help [son] to be able to understand himself, to better help him and prepare for what sort of things he needs and doesn't need." (1050-2-1)**

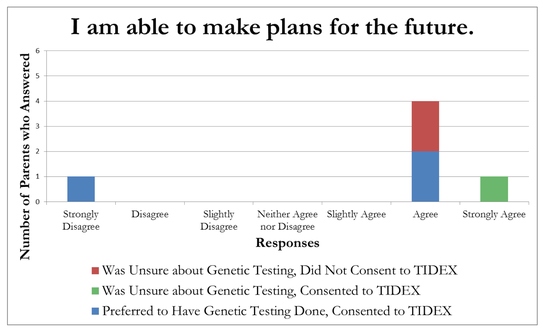

Figure 4

> **"If there was a diagnosis, and if there was something that the health practitioners could compare, it's kind of nice to maybe have an idea what to expect." (1047-2-3)**

INTERPRETATION

An explanation as to why many parents felt a diagnosis was not very important to them likely lies in the perceived low impact a diagnosis would have on school resources or community support for these parents. Curiosity and wanting to help other family members in their reproductive planning were brought up as reasons for wanting a diagnosis. However, the most common reason for wanting a diagnosis was in order to be more prepared for the progression of the child's condition in the future.

FURTHER DIRECTIONS

All six families had been coping with their child's condition for several years by the time of their interview, so the level of acceptance by the parents might be affected by the older age of the children. The one parent who classified a diagnosis as important to them is the only parent whose child's symptoms appeared after his first birthday. Further research into the importance of a diagnosis and age of the child when symptoms first appeared would help investigate whether this is associated.

The one parent who disagreed that they felt positive about the future and disagreed that they are able to cope with having this condition in their family was found to have another child with a genetic abnormality other than the affected child. This information should be used to more closely monitor and assist families with more than one child with a genetic condition.

CONCLUSION

The primary motivating factors in perceived importance of diagnosis might be impact on school resources and community support. The primary reason for electing to have genome-wide sequencing done was the chance to more accurately predict what their child's future holds.

ACKNOWLEDGEMENTS

This work is supported by the Canadian Institutes of Health Research (CIHR) MOP-89984 and BC Children's Hospital Research Institute.