GroopIt: An Innovative Platform to Speed Up Rare Genetic Disorder Research

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INTRODUCTION

- The CHD8 mutation is strongly associated with Autism Spectrum Disorder (ASD) (Bernier, 2014). Due to the rarity of this mutation, families are geographically widespread. Social media tools allow parents to share experiences and symptoms.
- Despite noted sleep issues within CHD8, very little is known about specific sleep problems associated with this genetic mutation.
- A new mobile platform, GroopIt (letsgroopit.com), may be a useful data collection tool to research rare genetic disorders at a faster pace than traditional research methods by reporting and tracking behaviors in real time.

METHOD

- Parents from the CHD8 Facebook group were invited to join and get familiar with the CHD8 GroopIt platform. We then conducted a month-long pilot to learn about sleep habits.
- Parents were asked to post observations every day for 30 days regarding characteristics of their child's sleep from the previous night (Figure 1).
- As observations were posted, they appeared in the newsfeed. This allowed parents to collaborate and comment about similar behaviors in their own children, while at the same time allowing researchers to collect structured data.
- We report the number of instances behaviors occured as well as how many children experienced the behaviors at all. These results were qualitatively and quantitatively described back to the parents via the GroopIt platform.

This mobile platform successfully captured daily sleep behaviors about children with a rare genetic mutation.

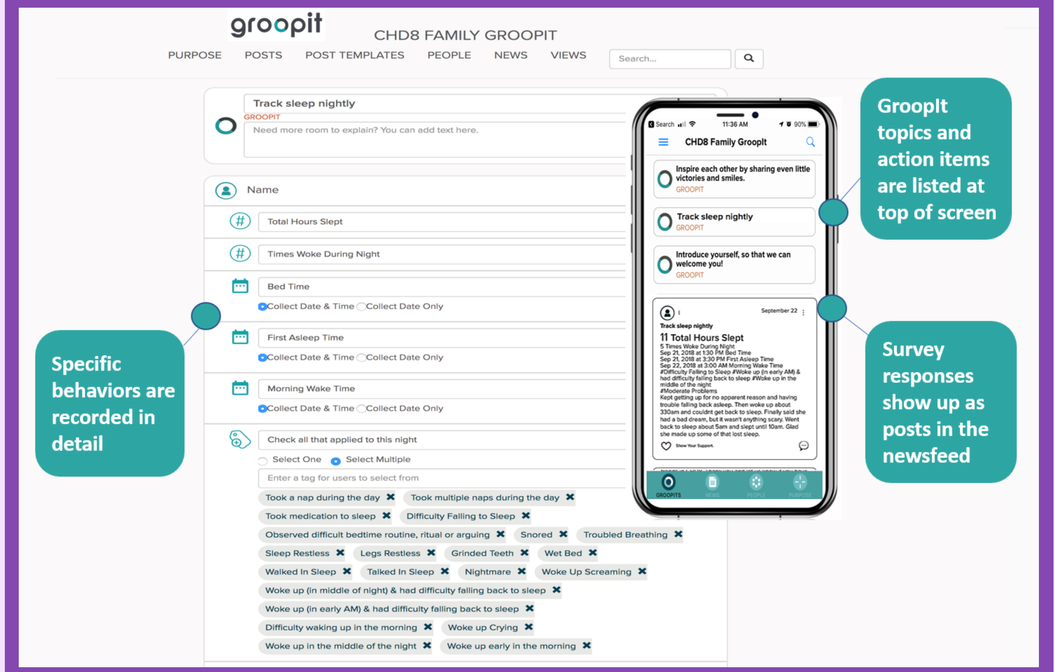

Figure 1: Groopit platform interface on desktop and mobile.

Sleep Domain	Question Item	# of Instances Reported (n=159)	# of Children (n=8)
Woke up crying or screaming	**Total**	**30**	**5 (63%)**
	Woke up Crying	27	3
	Woke Up Screaming	4	3
Mid-night/ early morning awakenings	**Total**	**62**	**6 (75%)**
	Woke up (in middle of night) & had difficulty falling back to sleep	23	4
	Woke up (in early AM) & had difficulty falling back to sleep	18	5
	Woke up in the middle of the night	18	3
	Woke up early in the morning	6	5
Difficulty falling to or back to sleep	**Total**	**102**	**7 (88%)**
	Woke up (in middle of night) & had difficulty falling back to sleep	23	4
	Woke up (in early AM) & had difficulty falling back to sleep	18	5
	Difficulty falling to sleep	54	7
	Observed difficult bedtime routine, ritual or arguing	7	3
Sleep quality disturbances	**Total**	**39**	**5 (63%)**
	Wet Bed	8	2
	Talked In Sleep	3	1
	Nightmare	3	2
	Snored	10	2
	Troubled Breathing	4	1
	Sleep Restless	7	3
	Legs Restless	4	2

DISCUSSION

- **Was this platform successful?**

Although only 8 parents participated during the pilot phase, the frequency of reports (159 reports over 30 days) was successful. This study gives us a month-long snapshot of what sleep looks like in the broader CHD8 phenotype.

- **Challenges we faced and what we learned?**

Choosing appropriate language for the survey was more difficult than we expected. We discovered the importance of gauging behavioral perceptions before constructing a survey. We also learned that reporting daily (to build a habit) and reporting both negative and positive events (to build encouragement) promotes higher data collection.

- **Ideal for all situations?**

In-person study visits continue to be the standard for research studies. However, this study shows collecting real-time observation data about rare genetic mutations can yield unexpected insights and be a strong compliment to standard in-person research studies.

- **Future application of findings?**

Mobile platform data collection connects families to researchers, ultimately helping to shape the direction of research and intervention as a partnership, and may help improve the quality and speed by which we discover phenotypic similarities associated with subgroups of ASD.

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