A “cure” for Down syndrome: What do parents want?

Angela Inglis^{1,3,*}, Zoe Lohn^{2,*}, Jehannine C. Austin^{1,3}, and Catriona Hippman^{1,2}

¹University of British Columbia Department of Psychiatry, Vancouver, Canada

²Women’s Health Research Institute, Vancouver, Canada

³University of British Columbia Department of Medical Genetics, Vancouver, Canada

Abstract

Recent advancements in molecular genetics raise the possibility that therapeutics or a “cure” for Down syndrome (DS) may become available. However, there are no data regarding how parents of children with DS perceive the possibility of mitigating specific manifestations such as the intellectual disability (ID) associated with DS, or curing the condition entirely. To explore these issues, we distributed a questionnaire to members of the Lower Mainland Down Syndrome Society in British Columbia, Canada. Questionnaires were completed by 101 parents (response rate=41%). A majority (61%) viewed the possibility of reversing ID in DS positively, but only 41% said that they would “cure” their child of DS if it were possible. Twenty-seven percent of respondents said they would not “cure” their child, and 32% were unsure if they would “cure” their child. The most commonly cited motivation for opting for a “cure” was to increase their child’s independence. However, parental attitudes’ towards a “cure” for DS were complex, affected by ethical issues, perceived societal values, and pragmatic factors such as the age of the individual and long-term care-giving burden. These findings could be used by healthcare professionals supporting families who include a member with DS and to direct future research.

Keywords

Down syndrome; intellectual disability; therapeutics; cure; attitudes; parents

INTRODUCTION

The intellectual disability (ID) associated with Down syndrome (DS) affects learning, language, and memory, with broad phenotypic variability¹. Clinical trials investigating the effects of donepezil pharmacotherapy on learning and memory have shown mixed results^{2–6}. More recently, mouse models have provided valuable insights^{7–10} into the pathogenesis underlying DS, which have led to novel targets for pharmacologic interventions^{11–13}. In addition, Jiang and colleagues¹⁴ recently reported inserting an *XIST* transgene into DS

Joint Corresponding Authors: Catriona Hippman, Women’s Health Research Institute, Rm B324A – 4500 Oak Street (Box 42), Vancouver, BC., V6H 3N1, Tel. 604 875 2424 x. 4909, Fax. 604 875 3895, catriona.hippman@ubc.ca. Jehannine C Austin, Departments of Psychiatry and Medical Genetics, Rm A3-112, CFRI Translational Lab Building, 938 W28th Ave, Vancouver, BC., V5Z 4H4, Tel. 604 875 2000 x. 5943, Fax. 604 875 3871, jehannine.austin@ubc.ca.

*Joint First Authors

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pluripotent stem cells *in vitro*, which, when induced, epigenetically inactivated the extra chromosome 21. Altogether, these developments suggest that therapeutics for specific manifestations of DS or even a holistic “cure” may eventually become available.

Though little is known about how families perceive the option of therapeutics for DS, studies have shown that a child with DS can have an overall positive impact on a family. Compared to other conditions such as autism and Fragile X syndrome, parents of children with DS exhibit better psychological well-being^{15,16}, report more positive experiences¹⁷, and report less stress associated with care-giving¹⁸. A study of parents of children with DS found that 79% felt their outlook on life was more positive as a result of their child, and 4% regretted having their child¹⁹. Together, these findings oppose the notion of burden that is associated with the medical model of disability²⁰ and suggest that there may be little support for therapeutics or a “cure” for DS. To date, only one study has investigated parental decision-making related to therapies for DS. Specifically, parents who pursued alternative medicine (e.g. nutritional supplements, stem cell therapy, plastic surgery) stated that, while they strove to accept their child, they also wanted to help their child reach their fullest health-related and developmental potential²¹. No study has explored whether, or to what extent, parents of children who have DS would support new therapies to mitigate ID or that could potentially cure their child of DS. We explored the perceptions of parents of children with DS towards these issues within the context of a larger study that aimed to better understand the experiences and perspectives of parents of children with DS.

MATERIALS AND METHODS

Study Design

A self-report questionnaire addressing three distinct research questions was developed in collaboration with board members from the Lower Mainland Down Syndrome Society (LMDSS), based in British Columbia, Canada. The LMDSS is the largest Canadian provincial non-profit organization that provides support and resources for individuals with DS and their families. The LMDSS distributed the anonymous questionnaires in study packages directly to their entire membership ($N=260$) in November 2008 and January 2009. A reminder letter was sent approximately three weeks after each mail-out. Study packages included: the questionnaire (described below), a cover letter from the LMDSS, a letter of introduction from the investigators, a BBC news article, which described the potential for pharmacotherapy to mitigate ID in those who have DS (Published: 2003/10/31 <http://news.bbc.co.uk/go/pr/fr/-/2/hi/health/3231285.stm>), and a stamped return-addressed envelope. Consent was implied with the return of the questionnaire. The University of British Columbia and Children's and Women's Hospital Ethics Boards approved this study (IRB #H08-01118; CW08-0178).

Participants

LMDSS members were eligible to participate if they were the parent or step-parent of an individual with DS and were fluent in English.

Questionnaire

The questionnaire comprised of sociodemographic items, questions relevant to the other two research questions^{22,23}, and four questions asking participants about their perspectives towards a cure for DS: 1) “The BBC article reports that researchers hope their work will eventually be able to reverse intellectual disability. If this were ever possible, do you think that would be a good thing or a bad thing?”, response options: a. Good thing, b. Bad thing, c. Not sure; 2) “If it were possible to “cure” your child of Down syndrome, would you want to?”, response options: a. Yes, definitely b. Yes, probably c. Maybe, I’m not sure, d. No, probably not, e. No, definitely not; 3) “Do you think researchers should be trying to find a “cure” for Down syndrome?”, response options: a. Yes, b. No, c. Not sure; 4) “What sorts of issues do you think researchers should be focusing on to try to help improve the quality of life of individuals and families affected by Down syndrome?”. For these four questions, in addition to the response options listed, respondents were provided with a blank space to provide comments. We did not define “cure” in the questionnaire or in the packages sent to participants.

Data Analysis

Quantitative data were entered into a database and data were described using frequencies and percentages. Three authors (AI, CH, JA) independently rated the degree of severity of the phenotype of each child with Down syndrome using a 3-point scale (1=mildly affected, 2=moderately affected, 3=severely affected), taking into account the age of the child, the number and nature of medical issues reported, and the child’s level of independence in relation to their age. Discrepancies in severity ratings were discussed and consensus reached, as described elsewhere²². In regards to the question “If it were possible to “cure” your child of Down syndrome, would you want to?”, responses were collapsed into three groups: yes (‘Yes, definitely’ and ‘Yes, probably’), no (‘No, probably not’ and ‘No, definitely not’), and maybe (‘Maybe, I’m not sure’). Chi-square analyses compared parents’ answers to this question, as well as responses to questions 1 and 3, with the severity ratings for the children with DS.

For the qualitative data, text responses from the open-ended questions were annotated in short sections (1–2 sentences) by two researchers independently (CH, ZL) to identify emergent themes using a descriptive analysis approach²⁴. Discrepancies were discussed and consensus reached. Participant verbatim quotes were selected to illustrate key points. All themes were reported by at least 5% of participants.

RESULTS

Response Rate

In total, 101 of 246 potentially eligible individuals responded, yielding a conservative response rate of 41%.

Participant Characteristics

Characteristics of the study participants and their children with DS, including consensus severity ratings, are summarized in Table 1. The majority of participants were female ($n=87$,

88%) and European ($n=79$, 79%). The participants were diverse in terms of age (range=29–97) and religious affiliation. The individuals with DS were predominantly diagnosed at birth ($n=78$, 80%) and had a variety of medical issues.

Attitudes Towards Treating ID and Curing DS

Parents' responses to questions about reversing ID, whether they would cure their child, and whether researchers should be trying to find a cure are shown in panels a, b, and c respectively (Figure 1). There was no relationship between the frequency with which parents indicated that reversing ID would be a good thing, would cure their child, or that researchers should be trying to find a 'cure' for DS and the severity of their child's condition (Chi-square=4.17, $df=4$, p -value=0.38; Chi-square=4.21, $df=4$, p -value=0.38; Chi-square=2.58, $df=4$, p -value=0.63, respectively). Parents provided diverse and rich responses in the open comment spaces provided for questions 1, 2, and 3. Careful analysis revealed commonality and overlap between responses to the three questions that rendered them inseparable. Thus, they were analyzed together, and from them six themes emerged, as follows. Further illustrative quotes are presented in Table 2.

1. Improving Quality of Life—Two thirds of the participants indicated that the major benefit to a cure would be increasing their child's independence and ability to perform tasks associated with daily living (e.g. “*ability to brush own teeth*”), which would ultimately make both the parents' and child's “*life easier*”.

2. Stigma, Societal Attitudes and Celebration of Diversity—The responses of approximately half the parents reflected views that society should be more accepting of diversity, including individuals who have DS, and questioned the motivation for “curing”. Many parents described the value of particular attributes of their children who have DS.

For some, the term “cure” was emotive; for example one parent said that it is “*highly judgmental to assume she [her daughter] should be ‘fixed’*” and another wondered “*would those who refuse it [the cure] be stigmatized?*”. But, consideration of specific interventions to improve quality of life was more acceptable.

3. Personality and Appearance—Some parents expressed concern that a cure would change their child's personality, and three parents stated that if a cure improved intellectual capacity, then they would feel the need to alter their child's outer appearance (e.g., through surgery).

4. Impact of a Cure on Parents—Some parents described the impact that a cure for their child would have on themselves. Many parents expressed concern about the care of their child after they are unable to provide care, which could be ameliorated if a cure increased the individual's independence. One parent said, for example, that she didn't want her daughter to be “*just another number...in a group home*”.

5. Ethical Lens—Some parents' responses related to the ethical principles of beneficence, nonmaleficence, autonomy, and justice, in considering whether or not to support a cure. To some parents, the principle of autonomy was most important, while other parents' supported

a cure based on prioritizing beneficence. Many parents expressed support for the cure, but only if the cure was “risk free”, with no “side effects” and did not result in harm to the individual (nonmaleficence). Some parents used the principle of justice to support a cure for DS, while others used it to argue against a cure for DS.

6. It Depends on What a ‘Cure’ Means—Approximately a quarter of respondents explicitly questioned what a cure would mean in terms of risks, psychological impacts, and medical implications. Interestingly, some parents ($n=12$, 12%) distinguished between curing infants or children compared to curing adults. Several parents expressed that they would have considered the option of a cure at the time of the birth of their child, but that curing an adult would be more difficult. Another of a 23-year-old stated, *“It may bring on a whole different set of issues at her age. If she was just born, I would consider it.”* Several parents expressed that a cure initiated “in utero” or “at or before birth” would be optimal because it could “change their abilities to function and learn and interact with others” early in life.

Some parents made the distinction between a cure and therapies for selective aspects of the syndrome. Other respondents defined what a cure would mean to them. Some participants ($n=16$, 16%) were skeptical, stating that they “doubt” that a cure could be developed, comparing it to “waving a magic wand to make it ‘all go away’”. Some parents believed that a cure would not be possible due to the genetic nature of Down syndrome. For instance, one parent stated: *“You cannot ‘cure’ DS as it is genetic. You can’t take the extra chromosome out.”* One respondent stated the possibility of a cure could cause false hope if a cure was not substantiated.

Areas of Research

When parents were asked what issues researchers should be focusing on to improve the quality of life of individuals and families affected by DS (question 4), several themes were described (Table 3).

Also in response to this question, participants spontaneously described a need to improve political support, public education, fundraising and support for life after high school. One parent stated:

“There needs to be more support from the community and extended family. It is not socially-accepted to have a child with Down syndrome...No one wants to help or even know what it is involved in raising a child with Down syndrome. It is incredibly challenging and beyond what a person can handle without proper support.”

DISCUSSION

This is the first study to report the perspectives of parents of children with DS towards therapeutics targeted at ID, and a “cure” for DS. In this study, broadly comparable proportions of parents indicated that they would, and would not, “cure” their child if it were possible. This diversity of opinion is unsurprising given the complexity of the question,

different potential interpretations of the term “cure”, and the tension between social²⁵ and medical models of disability²⁶.

It was interesting to note that the proportion of parents who felt that reversing ID would be a good thing was greater than the proportion that would want to cure their own child. This discrepancy could arise from parents believing that a cure should be an option for others, even if they wouldn't want to use it themselves, or from parents understanding reversal of ID and curing DS as two different issues, as explicitly described by some parents in this current study.

There were two rationales for supporting the pursuit of a cure that came up repeatedly for parents in this study. First, increasing their child's independence and quality of life would allow the parents to feel more secure about their child's future. This supports the findings of recent research showing that life-long dependence was a major concern for parents of children with DS^{19,27}. Second, their child deserved to reach their full potential. This too is consistent with previous work, showing that parents seeking alternative medicine for their child with DS did so to promote maximum well-being²¹.

Themes that emerged for parents not supporting the pursuit of a cure for DS often related to concern that a cure would change their child's personality or identity. A recent study of parents' attitudes towards medication treatment for attention-deficit hyperactivity disorder revealed that similar concerns constituted an important barrier to pursuing this treatment²⁸. Though some authors have suggested that parents' willingness to engage in interventions may increase as their child ages and the permanence of functional impairments become clearer²⁹, in this study, some parents were more comfortable with the idea of curing an infant as compared to an adult. This observation may be partially explained by reported concerns about altered personality. Specifically, whereas the personality of an adult has been established, an infant's personality is not yet fully known.

Relatedly, the positive impact that children with DS have on their families' lives was clearly articulated by participants in this study, as in previous work^{19,27,30–32}. Together with data from several studies showing that individuals with DS are happy with their lives and have high self-esteem^{33,34}, this undermines the assumption of individual suffering that is foundational to the medical model of disability from which the pursuit of a cure for DS has evolved²⁰. Though families that include a child with DS face unique challenges, including increased emotional, physical, and financial demands that can have a profound impact on the functioning and well-being of the family^{32,35,36}, parents in this study asserted that there should be a focus on changing negative societal attitudes and increasing social infrastructure to properly support individuals with DS and their families, rather than trying to change individuals with DS. This position is solidly in line with the social model of disability, which, in contrast to the medical model, suggests that barriers in society contribute significantly to difficulties experienced by those with DS²⁷. Future research could explore how these conflicting models of disability influence parents' desire to cure their child.

Strengths and Limitations

This study elicited both quantitative and qualitative data; the latter adds depth and richness to the former. In addition, since the LMDSS facilitated the distribution of the questionnaire and completed questionnaires were anonymous, bias towards socially desirable answers was reduced. The fact that we did not define “cure” in the questionnaire afforded benefits and limitations. While it allowed participants to explore the concept of cure as it was most meaningful to them, it also presented some difficulty in interpretation. While this study sheds light on an under-researched area, these data have a number of limitations. Although there were no obvious duplicate responses, the questionnaire was distributed twice to LMDSS members with no means of preventing someone from completing the questionnaire twice. In addition, the study participants were primarily Caucasian and female and all were English-speaking, which limits the generalizability of the results. Similarly, families who are not members of an advocacy organization may have different perspectives on research into new therapeutics for DS. Views expressed in this study may not be generalizable outside of the Canadian context or outside of the organization sampled. Other studies are needed to explore these issues in other contexts.

Future Study

Further exploration of the attitudes of others, including family members (fathers and siblings), healthcare providers, people with DS, and the general population, towards the possibility of ID therapeutics or a cure for DS would be valuable. As there have also been developments in potential therapeutics for Fragile X syndrome³⁷ and other genetic neurodevelopmental disorders³⁸, exploring the openness of stakeholders relevant to these conditions would be interesting, given both commonalities and differences between these conditions and DS. Future research should also examine the issue of how to operationalize patient-centred/patient-oriented research when the perspectives and priorities of the patient population are divided, especially given the ascendancy of patient-centred/patient-oriented research in North America.

Conclusions

Parents’ perspectives regarding a cure for DS are complex and influenced by several factors including the child’s age, potential side-effects, impact on personality, societal values, ethical issues, and long-term care-giving burden. This complexity is illustrated by the fact that similar proportions of parents indicated that they would, and would not, be interested in curing their own child. Parents in this study wrestled with the question of what a cure might mean, and raised concerns related to the role of the intellect in identity and personhood. These findings emphasize the importance of exploring philosophical and ethical questions in parallel with the laboratory science that is rapidly advancing.

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Figure 1.

Parents ($N=101$) responses (%) to three questions: whether reversing intellectual disability in individuals with Down syndrome (DS) would be a good thing or bad thing (panel A), whether they would cure their child (panel B), and whether researchers should be trying to find a cure for DS (panel C).

Table 1Sociodemographic and Clinical Characteristics ($N=101$)

	<i>n</i> (%)
Parents	
Sex	
Female	88 (87)
Male	13 (13)
Age	
19–29	1 (1)
30–39	16 (16)
40–49	40 (40)
50–59	31 (31)
60–69	10 (10)
70–79	1 (1)
80–89	1 (1)
90–99	1 (1)
Ethnicity	
European	79 (79)
Asian	6 (6)
Mixed	8 (8)
Other ^a	7 (7)
Religious	
Religious	44 (44)
Non-Religious	55 (54)
No Answer	2 (2)
Offspring with DS ($N=102$)^b	
Sex	
Female	53 (52)
Male	48 (48)
Age	
0–9	34 (34)
10–19	34 (34)
20–29	27 (27)
30–39	3 (3)
40–49	1 (1)
50–59	1 (1)
Medical Problems^c	
Surgery ^d	59 (60)

	<i>n</i> (%)
Vision Loss	48 (49)
Heart Defect	41 (42)
Hypotonia	41 (42)
Thyroid dysregulation	31 (32)
Dental problems	30 (31)
Hearing loss	29 (30)
Gastrointestinal problems	17 (17)
Timing of DS Diagnosis	
Prenatally	7 (7)
At Birth	78 (80)
>1 day post-delivery ^e	13 (13)
Level of Independence	
Under 5 years old	16 (16)
Between Age 5 – 19	53 (52)
Standard Curriculum at School	4 (4)
Modified Curriculum at School	47 (46)
Left School	2 (2)
Over Age 19	
Living with parent(s) – full time	19 (19)
Living with parent(s) – part time	4 (4)
Living with parent(s) – unspecified amount of time	4 (4)
Not living with parents	6 (5)
Independent Work/Volunteer	5 (15)
Supported Work/Volunteer	17 (17)
Not Working	11 (11)
Severity rating	
Mild	38 (37)
Moderate	41 (40)
Severe	23 (23)

^aThe only response given when participants selected “other” was Canadian. The options “African” and “Aboriginal” were also given, but no respondents selected these options.

^bThere was one family with twins who have Down syndrome (DS).

^cParticipants could select as many medical problems as were applicable.

^dSurgery for any indication

^eThe time of DS diagnosis ranged from 1 week after birth to 11 months.

Table 2

Themes from Responses to Questions 1, 2, and 3

Theme	n (%)	Illustrative Quotes
Improving Quality of Life	67 (66)	<p><i>"Now that my son is older the concern for his quality of life for him grows ... If I knew he would be able to take care of himself I would feel so much better."</i></p> <p><i>"if it would [...] expand his horizons then it is something to consider."</i></p>
Stigma, Societal Attitudes and Celebration of Diversity	49 (49)	<p><i>"We don't look to 'cure' blonde hair. This may seem like fallacious reasoning, but there is a line which we ought to respect in terms of accepting genetic diversity."</i></p> <p><i>"There is nothing 'wrong' with my daughter - She is perfect the way she is - it is how society views her that is wrong."</i></p> <p><i>"The term 'cure' is too problematic. Could I, without negative impact, restore or increase cognitive function, or reduce communication difficulties experienced by my daughter, I'd likely do so."</i></p> <p><i>"She has opened our world to so many positive experiences and teaches us every day so I would not want her to be changed in any way. I would want to be more like her not her to be more like me."</i></p>
Personality and Appearance	19 (19)	<p><i>"I wish he was smarter. But I would never trade his loving, outgoing, fun-loving personality."</i></p> <p><i>"If she still looked like someone who had Down syndrome then no one would hire her".</i></p>
Impact of a Cure on Parents	14 (14)	<p><i>"As a parent I worry about who will take care of my son when I'm gone."</i></p> <p><i>"I feel quite traitorous thinking that she'd be better if 'cured'."</i></p>
Ethical Lens	33 (33)	<p><i>"As with any 'cure' it will be up to an individual/family if they want to try the 'cure' and that option should be available for consideration."</i> (Autonomy)</p> <p><i>"I don't see taking a drug to cure or improve intellectual disability any different from wearing glasses or taking drugs for epilepsy."</i> (Beneficence)</p> <p><i>"Everyone wants what's best for their children and while there are many positives to Downs, no one can deny that my son will be at a disadvantage in life."</i> (Beneficence)</p> <p><i>"If it simply improved her mental or physical ability and there was no side effects or necessity to 'relearn her life' or 'relearn who she is' as an individual, then I would think it a very good thing."</i> (Nonmaleficence)</p> <p><i>"It would depend on how successfully this could be done. As with many things in medicine there are often side effects/complications, these would need to be known first."</i> (Nonmaleficence)</p> <p><i>"The funding would be better spent promoting the value of all people...Financial resources could also be spent on enhancing the quality of life for people with D[own] syndrome."</i> (Justice)</p> <p><i>"[Down syndrome] costs society the lives of many potentially productive individuals - who become dependent on public funding for their entire lives. In a society with socially funded health care, education etc this is a burden on the taxpayers"</i> (Justice)</p>
It Depends on What a 'Cure' Means	26 (26)	<p><i>"my son has had 21 years of learned behavior as a person with developmental delays...and increased intelligence won't likely change these behaviors. How do you un-do 21 years of being treated 'specially'?"</i></p> <p><i>"I think I would not want a cure for the attributes and characteristics but cures for the health concerns and learning deficiencies."</i></p> <p><i>"Intellectual challenges are only one part of Downs. Physical issues play a big part too. Is a "cure" something that addresses those issues too? I think it would be better to find ways for people w/ Downs to learn more easily, help w/apraxia, improve memory."</i></p> <p><i>"Yes, It would mean that children with Down's syndrome could have a better quality of life ... A cure for Down's syndrome would mean that these people would be able to lead a normal life."</i></p> <p><i>"Why would you not want a "cure" for a condition that causes so many medical/developmental problems? I love my son very much just as he is, but to have him be a "typical" 8 year old boy would be a dream come true".</i></p> <p><i>"I think the promise of a cure may lead to giving unrealistic hope to parents/other family members and prevent them from accepting the situation and moving forward with appropriate therapies, future life planning"</i></p>

Table 3

Areas of Future Research Suggested by Participants (Question 4)

Area of Research	n (%)	Illustrative Quote
Learning styles & communication, including cognitive function	36 (36)	<i>"I would be interested in research to find more effective ways to teach and support people with Down syndrome... We know that they can and do learn. We just need to find the best ways to do that."</i>
Medical concerns	34 (34)	<i>"I would say that any major health issues such as heart defects, leukemia, deafness, are all critical to the person with DS's overall development, so they should certainly be a main focus of research."</i>
Cure, therapy, causes of Down syndrome	6 (6)	<i>"Early gene therapy/more physical and mental enhancements via new hormonal interventions. Better understanding of muscular/bone problems and possible early interventions. Nutritional therapies for brain development might be another avenue"</i>
Information collected regularly to provide accurate information to parents	5 (5)	<i>"Researchers could gather more practical tips from parents on what works and what does not work in handling the many situations and behavioral problems of DS children and could publish that information in books that are not expensive. Researchers could make that information available to teachers and parents. Also, researchers could collect & publish up-to-date medical and growth stages, physically and mentally of DS people"</i>
Behavioral, psychological issues	5 (5)	<i>"To control the obsessive/compulsive personality traits, perhaps the defiance and stubbornness"</i>