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RESEARCHBRIEF

An update on HDI's research in the field of developmental disabilities

# Having a Son or Daughter with Jacobsen Syndrome/11q Deletion Syndrome: Perspectives of Parents

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## Background

Jacobsen syndrome (JS) is a rare genetic condition that often causes intellectual disabilities and health issues and has recently been added to various cell-free DNA prenatal screening tests. This condition often includes multiple physical features, a blood disorder, serious congenital heart defects in about 56% of patients, and a range of intellectual disabilities. Individuals may also experience mild to moderate impairment in expressive language; vision problems, digestive issues; and common infections of the upper respiratory system [Grossfeld, 2004]. However, because JS is relatively rare, psychosocial research has never been performed on this population. Correspondingly, a patient education resource about this condition has not yet been published that reflects "the medical and psychosocial implications of the diagnosis" identified as a priority in the ACMG guidelines when providing information to prospective parents learning about screening results.

Research on parents or prospective parents of children with Down syndrome, a similar genetic condition that also causes ID and some medical issues, indicates that parents want information about health issues, developmental delays, life expectancy, and other challenges, and they also want information about family and life outcomes, support services, psychosocial resilience, and photos depicting life with the condition [Levis, 2012; Sheets & Will, 2011]. Therefore, this research is critically important in demonstrating the psychosocial outcomes for families impacted by JS so that patient education materials at the moment of diagnosis can include the balance of information about common medical and developmental issues and also the psychosocial outcomes for families. This study seeks to answer some of the most commonly asked questions during prenatal and postnatal counseling sessions: What is life actually like for parents who have sons and daughters with JS? How many of them love their son or daughter with JS? How many of them regret having their child? In this largest study to date, parents from across the world respond. This information can provide a more complete picture of genetic conditions that are shaped not only by the karyotype and list of medical information but also the availability of services and supports, as well as family and community inclusion.

## Method and Research Questions

We created a survey instrument based on similar research conducted by Dr. Brian Skotko at Boston University Medical Center on "Having a Son or Daughter With Down Syndrome: Perspectives From Mothers and Fathers." Because there is no national registry of individuals who have JS in the United States, the final survey instruments were distributed to approximately 1,000 family members involved in the 11q Research and Resource Group, the only patient advocacy group for JS/11q Deletion in the US. The survey instrument was distributed through the 11q Research and Resource Group via its email distribution list and two Facebook groups administered by the organization.

### Survey Instrument

As no already existing survey instruments gathered the information that we were seeking, we created a five- page questionnaire for parents and guardians, informed by

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previous research with permission from Dr. Brian Skotko [Skotko, 2005a,b]. The surveys gathered both quantitative and qualitative information, using open-ended questions and a series of Likert statements on a scale of 1–7 (with "7" indicating strong agreement and "1" indicating strong disagreement). The questions measured parent attitudes and their perceptions of their child's health conditions and educational challenges. As optional measures, parents were asked to report sociodemographic information. In addition, we analyzed our results to determine if there was any difference in parent perceptions depending on whether or not the child had major health conditions.

## Results

Regarding parental perceptions of impact on family and community, respondents were only slightly more likely to agree that their child with JS was putting a strain on their marriage/partnership (21%) than they were to agree that their children without JS were putting a strain on their marriage/partnership (17%). Many (48%) agreed that their marriage was stronger because of their child with JS, and clear majorities felt that support groups were helpful (72%) and that they were comfortable answering questions from their children about JS (84%). See Figure 1.

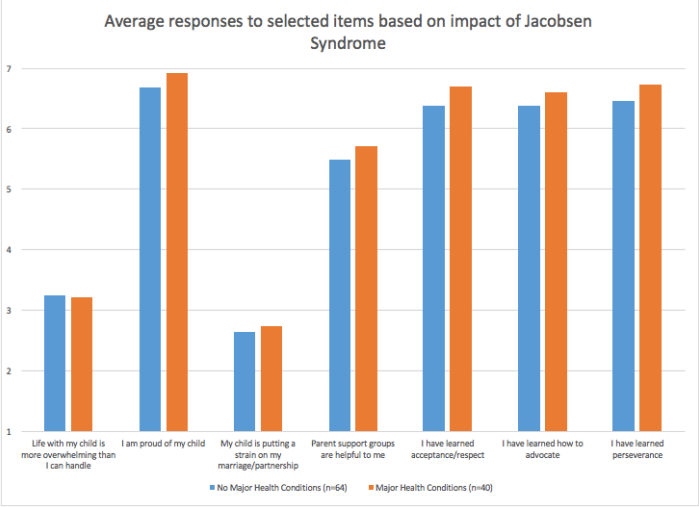


**Fig. 1: Average Responses to Selected Items Based on Impact of Jacobsen Syndrome**

Responses were most positive among Life Lessons learned from having a son or daughter with JS. Among the highest rated lessons were perseverance, patience and love. Only lessons regarding religion / faith (61%) and setting higher expectations for others (76%) were agreed with by less than 85% of respondents. When asked about advice for expectant parents of a child with JS, most agreed that there would be challenges but that they would learn how to advocate (98% each). Many stressed being positive (98%), patient (97%) and that parents would experience love (99%). Fewer, but still most respondents agreed that their child with JS has a good quality of life (79%) and that new parents should keep expectations high (82%). See Figure 2.

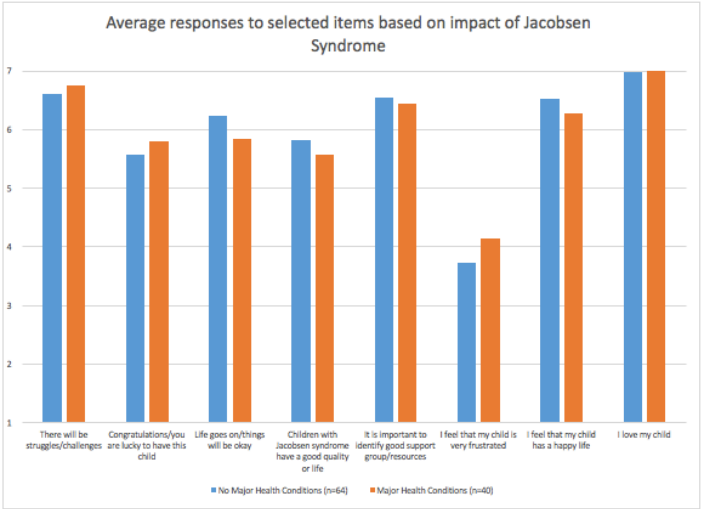


**Fig. 2: Average Responses to Selected Items Based on Impact of Jacobsen Syndrome**

## Discussion

Because medical guidelines for genetic screening encourage clinicians to provide up-to-date, accurate, and complete information about genetic conditions when counseling new or expectant parents [Gregg et al., 2016], this study of parents and guardians worldwide offers a uniquely important perspective regarding the psychosocial outcomes when parenting a son or daughter with a rare condition like JS that can cause ID and medical issues.

The overwhelming majority of parents responded that they love their child (100%), are proud of his or her accomplishments (97%), and enjoy spending time together (98%). A majority of parents (77%) also felt like their outlook on life was more positive because of their child with JS. A significant majority also felt their child had a happy life (94%). A smaller majority of parents (77%) also agreed that their child was very challenging. Therefore, even though the majority of parents perceived that their children experienced challenges, they also perceived that their children were happy. Put simply, parents did NOT perceive that a life of challenges precluded a life of happiness, pride, and love. One mother noted in the comments that her daughter is "the light of my life!"

A minority of parents also conveyed difficulties as illustrated by 28% of the parents who indicated that life with their child was more difficult than they could handle. We examined the data for possible underlying demographic trends for those parents who agreed most strongly that their child with JS was more overwhelming than they could handle (specifically those who marked "Agree" or "Strongly Agree" with the statement on the Likert scale, which totaled 10 respondents). The only significant patterns that emerged were that all 10 respondents indicated that their child had an ID and a learning disability, and 3 of the

10 respondents were from the Western Europe. The President of the 11q Research and Resource Group suggested that one demographic question that could be added for clarity in future survey instruments would be to ask parents if their child also has more severe behavior issues associated with autism, a common condition impacting about 47% of individuals with JS [Akshoomoff, Mattson, & Grossfeld, 2015].

Less than half of the parents (42%) indicated that their child with JS was very frustrated. Specific frustrations noted were that one parent said the child had difficulty finding friends his or her own age. Another parent also expressed difficulty finding a medical professional who understands JS. Interestingly, given the high scores from parents who perceived their child was happy, the results indicate that parents can identify their child with JS as being both very frustrated while also living a happy life, and the two psychological experiences are not viewed as mutually exclusive.

Most parents also indicated that their children taught them lessons in patience (97%), love (96%), joy (95%), perseverance (98%), advocacy skills (96%), and kindness/empathy (96%). Parents also felt it was important for new and expectant parents to know that there will be struggles and challenges (98%), and they will experience love (99%), personal growth/perspective (96%), and patience (97%). The high scores for these seemingly contradictory experiences (struggles and challenges versus love and joy) further demonstrate that life for the parents of children with significant disabilities can be challenging and rewarding at the same time. Parents also advised new and expectant parents to learn to be positive (98%) and identify good support group/resources (96%) as coping strategies. These outcomes further suggest that these skills (patience, advocacy skills, support groups, and a positive outlook on life) are important strategies for experiencing joy, love, and kindness/empathy while raising a child with a significant disability. One mother noted that the "11q family has been supportive and most helpful in this journey."

While US respondents were more likely than international respondents to indicate that their children with JS strengthened their marriage and faith, there was no statistically significant difference in the way the two groups perceived their children with JS. These results suggest that the life outcomes are similar for families worldwide in their individual experiences with their children [Noack, 2015].

Similar to a study of parents of children with Down syndrome conducted by Skotko et al. [2011], we found that the parents' perceptions of their child's health were not associated with their ratings of love or pride or happiness. Given that 100% of parents indicated that they loved their child with JS, it does not appear that variables such as race, educational level, age of child, etc. impact a parent's love for their child. Therefore, parent satisfaction was not dependent upon being more or less affected by the condition.

A majority of parents also believed that siblings had a good relationship with their brother or sister with JS and were more caring and sensitive because of the influence of the child with the condition. These results also echo the findings reported by Skotko et al. [2011] for sibling relationships when one child had Down syndrome. The relationship between the children was primarily described as positive, not associated with the degree of medical problems in the child with JS and not related to his or her level of functional activities. Moreover, nearly half of the respondents indicated that having a child with JS strengthened their marriage, and the percentage of those who did indicate that their child with JS caused a strain on their marriage (21%) was not much higher than the percentage of those who indicated their children without JS were putting a strain on their marriage (17%).

## Conclusion

Research indicates that families want written information about family and life outcomes, support services, psychosocial resilience, and photos depicting life with the condition [Levis, 2012; Sheets & Will, 2011]. The present study provides evidence-based psychosocial information that can be included in informational booklets during prenatal counseling with expectant parents, whether done in the offices of obstetricians, family practitioners, geneticists, and genetic counselors or in printed resources:

1. All of the parents reported that they love their son or daughter with JS, and the vast majority are proud of them regardless of their level of disability.
2. The overwhelming majority of parents who have children with JS report that their outlook on life is more positive because of their son or daughter with JS and that their child with JS has a happy life.
3. Parents who have children with JS report that their children have frustrations and challenges and yet their children with JS bring them both love and joy. They cite life lessons in patience, advocacy, and personal growth/perspective.
4. The overwhelming majority of parents who have children with JS say that their other children have good relationships with their brothers and sisters with JS and are more caring and empathetic because of their sibling with JS.
5. The majority of parents who have children with JS say that they have found parent support groups helpful and benefit from lessons in advocacy and information about positive outcomes.

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**HDI Research Briefs** were initiated to highlight the research activities at HDI. Projects at HDI focus on individuals with disabilities and include projects with emphases in early childhood, school age persons, adults, and issues across the lifespan. Many of these projects have significant research components and involve HDI staff, students in graduate programs, and other faculty at UK. With each issue of **HDI Research Briefs**, we will try to provide a cross-section of HDI's research activities. The brief reports are typically "mini" versions of more involved studies. The brief reports are intended to give an overview of the research project and emphasize the implications of the studies.

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