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The motivations and methods behind sharing a pediatric Prader–Willi syndrome diagnosis

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Abstract

Prader–Willi syndrome (PWS) is a genetic condition caused by a lack of paternally-expressed imprinted genes at chromosome 15q11.2-q13 and characterized by hyperphagia, behavioral challenges, and variable intellectual disability. Once a PWS diagnosis is established, sharing diagnosis information with an affected child can be challenging due to its early age of onset and diverse phenotype. This mixed-methods study aimed to evaluate how parents and guardians have shared a PWS diagnosis with their child and examine the motivating and influencing factors behind their disclosure. Parents and guardians of children with PWS aged at least 5 years completed a survey, and a select group completed an interview. A total of 51 surveys and 15 interviews were completed, with the majority of participants ($n = 46$; 90%) having shared at least some diagnosis information with their child. Parents and guardians were more likely to disclose if they self-reported a higher level of knowledge about PWS ($p = 0.004$) and if their child is currently older ($p = 0.02$) and/or has at least one sibling ($p = 0.046$). Interview analysis revealed 15 themes and 10 subthemes that illustrated parents' motivations, methods, and experiences with disclosure. This research provides information for others considering disclosure of PWS or another rare diagnosis with their child.

KEYWORDS
community, disclosure, family, parents, Prader–Willi syndrome, psychosocial

1 | INTRODUCTION

Prader–Willi syndrome (PWS) is a complex and rare genetic condition with an estimated prevalence of 1 in 15,000 to 1 in 30,000 individuals and no increased prevalence based on race or sex assigned at birth (Alves & Franco Rocha, 2020; Butler et al., 2019). PWS occurs due to lack of paternally-expressed imprinted genes at chromosome 15q11.2-q13 through either deletion of this region, maternal uniparental disomy, or an imprinting defect (Alves & Franco Rocha, 2020; Cassidy et al., 2012). PWS may be suspected at birth due to neonatal hypotonia and feeding difficulties (Alves & Franco Rocha, 2020; Wheeler et al., 2023) and diagnostic testing, consisting of DNA methylation analysis, is more than 99% accurate in diagnosing PWS (Driscoll et al., 2023). In a recent United States-based study, it was found that the diagnosis of PWS is typically made within the first 6 months of an individual's life (Wheeler et al., 2023).

Whereas infants with PWS struggle with failure to thrive and feeding difficulties, hyperphagia, or an unsatiable feeling of hunger, begins to develop in childhood, (Alves & Franco Rocha, 2020; Cassidy et al., 2012; Schwartz et al., 2021) leading to an increased risk for

childhood-onset obesity (Alves & Franco Rocha, 2020; Butler et al., 2019; Cassidy et al., 2012; Schwartz et al., 2021). Furthermore, the cognitive, behavioral, and emotional phenotype of PWS can develop during childhood and includes varying degree of intellectual disability, anxiousness (Alves & Franco Rocha, 2020; Cassidy et al., 2012; Schwartz et al., 2021), temper outbursts (Dykens et al., 2021; Einfeld et al., 1999; Schwartz et al., 2021), compulsive behavior (Dimitropoulos et al., 2001), psychiatric illness (Sinnema et al., 2011), and skin picking (Butler et al., 2019).

Once a diagnosis of PWS is established, sharing information about the diagnosis with the affected child can be challenging due to its early age of symptom onset, variability of symptoms, and the nature of the PWS phenotype, which generally includes some degree of intellectual disability, behavioral challenges, or both. It is already known that sharing genetic information with children is a complex task and a process that may include many stages over time (Dennis et al., 2015; Gaff et al., 2007; Hamilton et al., 2005), (Dennis et al., 2015; Forrest et al., 2003), but studies have emphasized that children affected with a genetic condition may benefit from learning of their diagnosis at a younger age despite disclosure complexities (Goodwin et al., 2014; Metcalfe et al., 2008; Plumridge et al., 2010; Takahashi, 2005). Reported motivations for sharing a pediatric diagnosis are diverse, (Goodwin et al., 2014; Metcalfe et al., 2008; Plumridge et al., 2010), (Dennis et al., 2015; Faux et al., 2012; Goodwin et al., 2014) (Gallo et al., 2005; Kaneko et al., 2022), (Faux et al., 2012; Goodwin et al., 2014; Holt, 2006; Metcalfe et al., 2008; Plumridge et al., 2010) and many studies have described the dilemma parents face regarding what, when, and how to share diagnosis information with their child. (Cavanagh et al., 2010; Dennis et al., 2015; Franchi et al., 2023; Gallo et al., 2005; Gallo et al., 2009; Kaneko et al., 2022; Kiely et al., 2020; Metcalfe et al., 2011; Plumridge et al., 2010). Furthermore, parents in previous studies have expressed a lack of resources to guide them through these disclosure conversations (Abi Daoud et al., 2004; Fitzgerald et al., 2021; Franchi et al., 2023; Hallberg et al., 2010).

Broad pediatric disclosure guidelines have been developed that recommend considering a child's maturity, capability to understand complex medical information and cognitive development when making decisions regarding sharing diagnosis information (Bibace & Walsh, 1980; Wilfert et al., 1999; Woodard & Pamies, 1992). However, these recommendations are nonspecific, and do not address the nuances of disclosing a genetic diagnosis. Given the variability of the cognitive abilities, psychological implications, and health features between and within genetic diagnoses, disclosure conversations will inevitably look different across each family and condition. For that reason, further research and assessment of family communication and disclosure practices for specific genetic conditions is vital to ensure that families do not need to settle for a "one-size-fits-all" approach in an area where there is such great variability and nuance.

Therefore, the goal of this study was to understand the strategies that parents and guardians have used to share information about a PWS diagnosis with their child and examine motivations and influencing factors behind sharing or not sharing diagnosis information. We hypothesize that parents or guardians have different strategies and techniques for explaining information depending on their child's age, cognitive capabilities, and family birth order or family structure. Additionally, we predict that parents or guardians have diverse motivations for sharing or withholding certain diagnosis or health information, potentially guided by their child's age, specific phenotype, healthcare visits, and the parent and child's understanding of the diagnosis. Through the investigation of the parents' experiences sharing a pediatric PWS diagnosis, we aim to evaluate current practices and identify support areas for parents and guardians interested in discussing a PWS diagnosis with their child.

2 | MATERIALS AND METHODS

2.1 | Ethical considerations

Approval to conduct this human subjects research was obtained by the University of Alabama at Birmingham Institutional Review Board. All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the Helsinki Declaration of 1975, as revised in 2000 and no nonhuman animal studies were carried out by the authors for this article. Informed consent was obtained from all participants who voluntarily completed the online survey questions and participated in interviews.

2.2 | Recruitment

Participants of this study were parents or guardians of children with PWS, primarily recruited through PWS-specific Facebook groups, including those associated with the Foundation for Prader-Willi Research (FPWR), a nonprofit patient advocacy group that aims to advance PWS research and therapeutic development. A social media post that included the survey link was uploaded to the FPWR Facebook group page a total of three times, with follow-up 1 week after the initial post and then another follow-up repost after 3 weeks per the Dillman method (Hoddinott & Bass, 1986). The FPWR Facebook group page has over 2500 group participants, however, it is unknown how many Facebook group members viewed or interacted with the recruitment post out of the total membership. At the end of the survey, participants could volunteer to participate in a qualitative interview. Recruitment occurred from July 18, 2023, until September 1, 2023. The study inclusion criteria were parents or guardians of children with PWS aged at least 5 years. Participants had to live in the United States and be able to speak and read English.

2.3 | Survey

The survey was designed using REDCap electronic data capture tools hosted at UAB. REDCap is a secure, web-based software platform

designed to support data capture for research studies (Harris et al., 2009). The survey was developed by the principal investigator (VM) and reviewed by the other research team members (JD, JEB, and TVS), which consisted of a certified genetic counselor and two content experts on PWS from the FPWR. The survey questions were reviewed prior to dissemination by two individuals who have a child with PWS and served as representatives of the parent population ("parent advisors") to confirm the clarity of questions and survey logic. The REDCap survey included multiple-choice questions related to demographic information, sibling count and birth order, and multiple-choice and open-ended questions related to disclosure practices, disclosure resource utilization, and desire for resources (Figure S1) Survey responses remained anonymous unless participants voluntarily provided their contact information for interview selection consideration.

2.4 | Interview

Interviews were conducted via secure Zoom meetings to further understand participants' motivations for and methods behind disclosing a PWS diagnosis to their affected child. After evaluating the extent of information participants reported sharing with their child in the survey, participants were placed into one of three groups by the primary investigator: full disclosure, half disclosure, or no disclosure (Table 1). Maximum variation purposive sampling was used to choose survey participants to interview in an effort to obtain a well-balanced cohort of disclosure levels. These semi-structured qualitative interviews were conducted by the principal investigator (VM), a graduate student in genetic counseling, using one of three interview guides (Appendix A) based on the extent of a participant's disclosure. Interview guides were reviewed by the parent advisors to confirm the appropriateness of questions and clarity of wording. Interview questions explored motivating factors for disclosure conversations, parent and perceived child reactions to these conversations, use of resources in these discussions, and suggestions for future resource development. Interviewees were identified only by their REDCap Record ID after their interview was complete to keep interview transcripts anonymous. Zoom was used to record the interviews and transcribe the interviews verbatim; the principal investigator (VM) reviewed the transcripts for accuracy before coding was initiated. Participant interviews were concluded when theoretical sufficiency was reached.

TABLE 1 Disclosure status.

Disclosure status	Survey participants *N* (%)	Interview participants *N* (%)
Total	51	15
Have you discussed health differences/symptoms?		
Yes	46 (90%)	14 (93%)
No	5 (10%)	1 (7%)
Is your child aware that their health differences are related specifically to a PWS diagnosis?		
Yes, for all health differences	31 (67%)	6 (43%)
Yes, for some health differences	4 (9%)	3 (21%)
No	11 (24%)	5 (36%)
Disclosure group		
Full disclosure[a]	35 (69%)	9 (60%)
Half disclosure[b]	11 (21%)	5 (33%)
No disclosure[c]	5 (10%)	1 (7%)

Abbreviation: PWS, Prader–Willi syndrome.
[a]Shared health differences and child knows that health differences are associated specifically with PWS diagnosis.
[b]Shared health differences, but child does not know that health differences are associated specifically with PWS diagnosis.
[c]No diagnosis or health difference information has been shared with the child; participant has been omitted from qualitative analysis to maintain interview confidentiality.

2.5 | Data analysis

Quantitative survey data were exported into Excel version 16.78 and incomplete survey responses were removed from the dataset. Descriptive statistics for demographic information were completed using Excel version 16.78. Additional data were analyzed using the SAS software version 9.4 (Cary, NC, USA). An exact Wilcoxon test was used to compare the child's number of siblings, current age, and diagnosis age to disclosure status of either not sharing anything at all with the child (i.e., no disclosure participants), or sharing the diagnosis to some extent (i.e., full and half disclosure participants combined). Pearson correlation was calculated for full disclosure, half disclosure, and no disclosure across continuous variables such as the child's number of siblings, child's current age, and child's age at disclosure. An exact Cochran–Mantel–Haenszel test was used to assess association between disclosure status (i.e., full, half, or no disclosure), sibling birth order in the family, parent-reported level of knowledge about PWS and parent general education level. An exact Mantel–Haenszel test was also used to evaluate how symptoms shared with the child influenced disclosure status. The significance level for all tests was defined as <0.05.

Qualitative interview data transcripts were coded using the NVivo Lumivero version 12 computer-assisted qualitative data analysis software (CAQDAS). Transcripts were organized into full disclosure, half disclosure, and no disclosure groups. Inductive coding and reflexive thematic analysis were completed by two research members (VM and JD) following a constructivist paradigm, designating that analytic codes were developed for the first time from the transcripts as they were read through, with no prior expectations or pre-set themes created before coding. However, although no anticipated themes were developed prior to coding, there is subjectivity within the analysis given that the researchers (VM and JD) have backgrounds in genetic counseling, and this subjectivity is acknowledged within a reflexive thematic approach (Braun & Clarke, 2012, 2020; Grant & Giddings, 2002; Merriam, 2009). VM and JD discussed their codes

and consensus were reached before grouping codes into subthemes and themes. The transcripts were then recoded using the established themes by both VM and JD. Any discrepancies in coding were discussed and resolved. These final themes were then evaluated by TVS and JEB to ensure they accurately captured the lived experience of the target population.

3 | RESULTS

3.1 | Survey results

Parents and guardians of children with PWS were invited to complete a survey that asked questions regarding demographics, disclosure practices, and resource utilization to better understand PWS diagnosis disclosure. A total of 65 participants completed the survey from a pool of more than 2500 individuals who had the opportunity to view and interact with the social media recruitment posts. These 65 participants' family members' current ages ranged from 5 to 38 years old. Fourteen responses were incomplete and omitted from the final analysis, resulting in a total analyzed sample size of 51 responses. Demographic information for survey and interview participants is displayed in Table 2. Information about participants' children with PWS is shown in Table 3.

As shown in Table 1, a total of 46 (90%) survey participants reported they have discussed their child's health differences or symptoms with them to some extent, and 5 (10%) participants responded that they have not discussed their child's health differences or symptoms with their child at all. These five participants comprised the no disclosure group. In the further delineation of disclosure status, 31 (67%) participants reported that their child is aware that their health differences are related specifically to a PWS diagnosis for all of their relevant symptoms, 4 (9%) participants reported that their child is aware of the relation to PWS for some of their health differences or symptoms, and 11 (24%) participants reported that their child is not aware that their health differences are related to a PWS diagnosis. This latter group of 11 participants represented the half disclosure group, as their child is aware of health differences, but not aware that these symptoms are related directly to a PWS diagnosis.

To evaluate additional parent or child characteristics that correlated with disclosure status, Pearson correlation, exact Cochran–Mantel–Haenszel test, and exact Wilcoxon test were used. Parents and guardians were more likely to have disclosed to some extent to their child (i.e., full or half) if their child's current age was older ($p = 0.02$) and if the participant self-reported an increased level of knowledge about PWS (i.e., "Very Educated" versus "Uneducated") ($p = 0.004$). Parent and guardian participants were more likely to have fully disclosed to their child if their child's current age was older ($p = 0.0003$) and if their affected child had at least one sibling ($p = 0.046$). Not having disclosed was more likely if the child was an only child ($p = 0.046$). General parent or guardian education level (i.e., "High School degree or GED" versus "Graduate degree"), race, and child's sex assigned at birth were not significantly associated with disclosure status.

TABLE 2 Survey and interview participant demographics.

Demographics	Survey participants *N* (%)	Interview participants *N* (%)
Total participants	51	15
Parent age (years)		
18–30	1 (2%)	0
31–40	13 (25%)	3 (20%)
41–50	18 (35%)	6 (40%)
51–60	12 (24%)	5 (33%)
61–70	7 (14%)	1 (7%)
Relationship to Child		
Biological mother	44 (86%)	14 (93%)
Biological father	3 (6%)	1 (7%)
Adoptive mother	3 (6%)	0
Adoptive father	0	0
Legal guardian	1 (2%)	0
Sex Assigned at Birth		
Female	48 (94%)	14 (93%)
Male	3 (6%)	1 (7%)
Race		
White only	38 (75%)	14 (93%)
Other or multi-race[a]	13 (25%)	1 (7%)
Education Level		
High School degree or GED	2 (4%)	2 (13%)
Some College	4 (8%)	0
Associate's degree/2-year degree	7 (14%)	1 (7%)
Undergraduate degree/4-year degree	18 (35%)	6 (40%)
Graduate degree	20 (39%)	6 (40%)
Self-Perceived Education Level about PWS		
Uneducated	1 (2%)	1 (7%)
Somewhat educated	4 (8%)	0
Educated	18 (35%)	3 (20%)
Very educated	28 (55%)	11 (73%)

Abbreviations: GED, general education diploma, PWS, Prader–Willi syndrome.

[a]Three multi-race, three Asian persons, three Black or African American persons, and five Hispanic or Latino persons.

Overall, the symptoms that were most commonly discussed by the full and half disclosure groups with their children were special diet requirements (84%), physical challenges (72%), weight and obesity concerns (61%), hyperphagia (59%), and gastrointestinal problems (52%). For the full disclosure group, parents most frequently report their children know that their special diet requirements (78%), anxiousness and social differences (64%), and gastrointestinal problems (53%) are related to their PWS diagnosis. An exact Mantel–Haenszel test was used to evaluate how the types of symptoms shared with the

TABLE 3 Child demographic information and characteristics.

Information	Survey participants *N* (%)	Interview participants *N* (%)
Total	51	15
Child current age (years)		
Mean (range)	13.8 (5–38)	11.9 (5–24)
Child age at diagnosis		
Birth–2 weeks	10 (20%)	2 (13%)
>2 Weeks–1 month	19 (37%)	8 (53%)
>1 Month–6 months	9 (17%)	3 (20%)
>6 Months–1 year	4 (8%)	0
>1 Year–5 years	6 (12%)	1 (7%)
>5 Years–10 years	3 (6%)	1 (7%)
Sex assigned at birth		
Female	27 (53%)	5 (33%)
Male	24 (47%)	10 (67%)
Siblings		
No other siblings	9 (17%)	2 (13%)
1 other sibling	27 (53%)	9 (60%)
2 other siblings	9 (18%)	2 (13%)
3 or more other siblings	6 (12%)	2 (13%)
Birth order		
Child with PWS is the youngest child	20 (47%)	4 (31%)
Child with PWS is a middle child	4 (9%)	0
Child with PWS is the oldest child	19 (44%)	9 (69%)

Abbreviation: PWS, Prader–Willi syndrome.

child were associated with overall disclosure status. Parents or guardians who shared symptoms such as learning differences ($p < 0.0001$), gastrointestinal differences ($p = 0.005$), special dietary needs ($p = 0.0004$), weight and obesity concerns ($p = 0.002$), hyperphagia ($p < 0.0001$), physical challenges ($p = 0.0002$), and behavioral differences ($p = 0.002$) were significantly more likely to be in the full disclosure group compared to the half or no disclosure groups. The only symptom discussed with the affected child that did not statistically correlate with full disclosure status was social challenges ($p = 0.12$).

Twenty-nine (83%) participants in the full disclosure group responded that they did not use educational resources when initially discussing their child's PWS diagnosis with them. However, 20 (57%) participants in the full disclosure group responded that they had suggestions for resources that could have been used during the diagnosis disclosure. Some of these resource suggestions include videos, age-appropriate books, case examples of other children with PWS, information for their child about puberty, and infographics to define and explain PWS-related medical terminology with simple and age-appropriate language.

3.2 | Interview results

Interviews were conducted to better understand participants' unique motivations for and methods behind disclosing a PWS diagnosis to their affected child. Twenty-three parents and guardians of children with PWS were invited to participate in an interview and 15 interviews were completed. Interviews were conducted between July 31, 2023, and September 8, 2023, with an average interview duration of 43 min. Of the interview cohort, 9 (60%) participants were in the full disclosure group, 5 (33%) participants were in the half disclosure group, and 1 participant (7%) was in the no disclosure group (Table 1). This singular no disclosure participant was excluded from qualitative analysis due to concerns for confidentiality based on the small sample size. Inductive coding and reflexive thematic analysis were performed using three categories to organize themes and codes: motivations and influencing factors for disclosure (Table 4), methods for disclosure (Table 5), and disclosure experiences (Table 6). Representative quotes for each theme are included in Tables 4–6.

3.3 | Motivations and influencing factors for disclosure

Six themes and four subthemes were identified regarding factors that influenced or motivated parent or guardian diagnosis disclosure (Table 4). These themes include truth and honesty, child's cognition and understanding, disclose to explain symptoms (with subthemes: hyperphagia makes disclosure inevitable, and talk about symptoms as they come up), advocacy and empowerment, child guides, and motivates (with subthemes: child did not have questions, so did not share information, and child did have questions, so did share information), and PWS community motivates disclosure.

3.4 | Methods of disclosure

Six themes and four subthemes were identified regarding the methods that parents or guardians used to disclose a PWS diagnosis to their child (Table 5). These themes include use of multiple conversations over time (with subthemes: provide alternative dreams and options, and build a foundation of knowledge), be a unified team, use matter-of-fact language, normalize to others, talk on the way to appointments, and helpful phrases (with subthemes: diagnosis does not define you, and describing the diagnosis).

3.5 | Experiences with disclosure

Thematic coding and analysis were performed to evaluate parent and guardian experiences with PWS diagnosis disclosure (Table 6). Three themes and two subthemes were identified regarding parent and guardian disclosure experiences. These themes include just a part of

TABLE 4 Themes and subthemes: Motivations and influencing factors for disclosure.

Theme	Subthemes	Number of participants	Number of codes[a]	Representative quote[b]
Child's cognition and understanding	-	11 (73%)	18	"Him having good mental capability has been more reassuring for me and that's why my husband and I felt like he was ready. …We started talking to him about his diagnosis when he was 5 or 6, maybe 7 … I think a big reason why we were motivated to do that is because he was always doing well with the cognitive stuff." —Mother, Full Disclosure, Child age 11
Truth and honesty	-	10 (66%)	23	"My number one recommendation would be to be forthright and honest. Because if you hold it in, it gains a negative power. You need to explain where their differences come from and then show that this is not a bad thing." —Mother, Full Disclosure, Child age 20
Disclose to explain symptoms	Hyperphagia makes disclosure inevitable	9 (60%)	12	"…You know, everyone eats, we eat all the time. So it's just something that has come up a lot and of course very early on, and so I think that's why we have always been so open about the food stuff and him being hungry and feeling hungry." —Mother, Full Disclosure, Child age 17
	Talk about symptoms as they come up	8 (53%)	18	"We've often explained, well, you have 'X' symptom or 'X' impacts you because you have this overall thing called PWS, and he knows he has it… I guess he refers to it mostly as PWS, but he knows it stands for Prader–Willi syndrome." —Mother, Full Disclosure, Child age 11
Advocacy and Empowerment	-	9 (60%)	17	"My goal is to empower her with knowledge, and the vocabulary and the words so she can advocate for herself. That has been my number one goal…for her to feel comfortable in her own skin." —Mother, Full Disclosure, Child age 12
Child guides and motivates	Child did not have questions, so did not share information	5 (33%)	9	"And you know there will come a day where I expect him to have all of these questions. And we will explain 'Yeah this is what you have,' and "This is what it means"…but because he's so young and doesn't have those questions we know we can keep it light for now." —Mother, Half Disclosure, Child age 5
	Child did have questions, so did share information	5 (33%)	6	"In our experience it made sense to always share it from a point where he was aware and understanding of it, which is probably a couple of years old, if not a little bit older. Maybe by the time or the time they first start asking questions like 'Why do I go to this doctor' or 'Why do I go to this therapist?'" —Mother, Full Disclosure, Child age 11
PWS community motivates disclosure	-	5 (33%)	7	"It was on the way to a Prader-Willi fundraiser. He was like 7, maybe, and just kind of asking, 'Where are we going and why?' And so it just happened. I feel like driving to the fundraisers or PWS events are where we talk about it the most." —Mother, Full Disclosure, Child age 17

[a]This column refers to the number of separate times the theme or subtheme was identified and coded for in the complete set of interview transcripts.
[b]Child current age in years.

life, parental self-doubt and anxiety, and child's response to disclosure (with subthemes: positive response and negative response).

4 | DISCUSSION

Although some information is known about how parents and guardians share genetic information with children (Dennis et al., 2015; Franchi et al., 2023; Metcalfe et al., 2011; Plumridge et al., 2010), there is a gap in the literature regarding sharing diagnosis information-related specifically to PWS. Through the analysis of 51 survey responses and 15 parent interviews, this study evaluated how parents and guardians have shared PWS diagnosis information with their child and examined motivations for and experiences with disclosure.

Parents and guardians are more likely to disclose to their child if their child has siblings, if they feel knowledgeable about PWS, and if their child is currently of older age. These findings suggest that parents may be more inclined to share diagnosis information with their affected child if their child is able to recognize their differences in direct comparison to a sibling and absorb and understand the information due to their age-related maturity level. This is similar to previous literature, which has found that disclosure may be prompted by siblings and affected children wanting to understand their differences (Kaneko et al., 2022). Notably, parental general education level (i.e., "High School" versus "Graduate degree") was not statistically correlated with disclosure, but parental self-reported education level about PWS (i.e., "Very Educated" versus "Uneducated") was statistically correlated with disclosure. This result demonstrates that if

TABLE 5 Themes and subthemes: Methods for disclosure.

Theme	Subthemes	Number of participants	Number of codes[a]	Representative quote[b]
Be a unified team	-	10 (66%)	18	"You know, keeping him so involved in our conversations and telling him about his diagnosis really young I think really helped us feel like we were on a team and on his side. And now that he's older and we help him with a lot of things, I think that he feels better about us helping him with things because we've always just been on his team." —Mother, Full Disclosure, Child age 24
Use multiple conversations over time	Provide alternative dreams and options	9 (60%)	13	"As she got old enough to handle the discussion I would tell her, 'There aren't very many women with Prader–Willi syndrome that have children. So it is most likely that you're not going to have children.' And then I talked about other people that we know that haven't been able to have children that have adopted or that haven't and don't have children in their family…And she is an aunt to a lot of nieces and nephews and so talking about how 'You could be a really good aunt.' So just providing alternatives helps the conversation go smoother." —Mother, Full Disclosure, Child age 17
	Build a foundation of knowledge	9 (60%)	18	"It's not ever just one conversation, it might be a series of conversations that happen over time.…We try to find age-appropriate ways of describing it and continue to build on that foundation over time" —Mother, Half Disclosure, Child age 5
Use matter-of-fact language	-	9 (60%	16	"I would say, we've kept it pretty matter-of-fact. We haven't tried to minimize the condition, but we like to be matter-of-fact, not emotional. And treating things as if they're not something for her to worry about. We try to take that tone rather than 'Oh, my gosh, I can't believe this this! You're getting worse!'" —Mother, Full Disclosure, Child age 13
Normalize to others	-	9 (60%)	14	"It makes it better that we talk to him about stuff because he knows that it's normalized. Like we don't try to pass him off like he's normal, because he's not. But we just talk about how everyone has differences and it's okay to have differences. Like he's getting glasses, like 'Daddy has glasses, and you will have glasses,' and we try to emphasize that we are all the same in a lot of ways." —Mother, Half Disclosure, Child age 5
Talk on the way to appointments	-	9 (60%)	14	"We spend a lot of time in the car after school driving to different appointments and that's basically a good time and place where a lot of these conversations take place."—Mother, Full Disclosure, Child age 18
Helpful phrases	Diagnosis does not define you	9 (60%)	14	"…We've always tried to tell him, 'Your syndrome is not who you are and it doesn't define you. It's just a challenge that you live with and a part of your life.'" —Mother, Full Disclosure, Child age 24
	Describing the diagnosis	4 (26%)	7	"We didn't say disease or condition or syndrome. We just said, 'That's called Prader-Willi, and we're learning about it. And it's just going to mean special things we do with your body as far as how you eat and what you can do physically, and learning will help us understand how to support you.'" —Mother, Full Disclosure, Child age 13

[a]This column refers to the number of separate times the theme or subtheme was identified and coded for in the complete set of interview transcripts.
[b]Child current age in years.

parents are able to become more knowledgeable about PWS, they may feel comfortable disclosing regardless of their attainment of higher education, and supports previous studies where parents found it helpful to research their child's diagnosis and become knowledgeable about the condition (Dennis et al., 2015; Kaneko et al., 2022).

4.1 | Motivations and influencing factors for disclosure

Results from this study revealed that parents and guardians have a variety of unifying factors that motivate their disclosure conversations or influence the way they tailor their disclosure approach. First, parents and guardians are motivated to disclose due to a desire to be honest and truthful with their child. This draws on what has been found previously, as caregivers in other studies have reported disclosing because they do not want to create secrets (Goodwin et al., 2014; Metcalfe et al., 2008; Metcalfe et al., 2011) and do not feel a need to conceal the diagnosis from the child (Takahashi, 2005). Similar to other studies which identified that parents may make disclosure decisions based on their child's cognition and understanding (Dennis et al., 2015; Franchi et al., 2023; Gallo et al., 2005; Kaneko et al., 2022; Kiely et al., 2020), participants in this study felt encouraged to disclose if they felt confident that their child would

TABLE 6 Themes and subthemes: Disclosure and diagnosis experiences.

Theme	Subthemes	Number of participants	Number of codes[a]	Representative quote[b]
Just a part of life	-	10 (66%)	22	"We knew of her diagnosis when she was 3 weeks old. So, we've known from the beginning, basically. And so we just incorporated it as part of our everyday life. It's always been around, so it was just naturally a part of our life." —Mother, Full Disclosure, Child age 20
Parental self-doubt and anxiety	-	10 (66%)	18	"It's also hard too without all of the information that we need. If we don't have full information to be able to confront her concerns, and my daughter wants the information… that's when it starts getting irrational. And those are moments where, yeah, there can be anxiety-provoking moments and doubt. It's not always easy and it can make you wish that you never said anything in the first place. I mean… definitely being a PWS parent is tough. I don't want to paint this like it is all rosy to share a diagnosis." —Mother, Full Disclosure, Child age 13
Child's response to disclosure	Positive response	7 (46%)	8	"It made me happy to tell him about his diagnosis because I could see that having those answers did relieve stress for him kind of. Like as long as you tell them small doses at a time and are appropriate about it I actually think them knowing can be a stress reliever."—Mother, Full Disclosure, Child age 17
	Negative response	6 (40%)	10	"I know people that try to tell him like, 'oh that's okay, you don't have to do that because you can't or you're not normal, it's okay.' And I hate that. He has to do the stuff, and I don't want him to use his symptoms as an excuse." —Mother, Half Disclosure, Child age 6

[a]This column refers to the number of separate times the theme or subtheme was identified and coded for in the complete set of interview transcripts.
[b]Child current age in years.

understand the disclosure conversations. Published guidelines and recommendations for pediatric diagnosis disclosure similarly recommend considering a child's cognitive development and understanding when planning disclosure conversations (Bibace & Walsh, 1980; Wilfert et al., 1999; Woodard & Pamies, 1992). This recommendation to consider a child's cognitive abilities is increasingly pertinent for the PWS population, as the majority of individuals with PWS (90%–100%) have some degree of intellectual disability, with mean IQ scores falling in the range of 60–70. However, the range of cognitive ability is broad, with some individuals having moderate intellectual disability and others having cognition within normal limits (Driscoll et al., 2023). However, two previous studies differ, in that participants in these studies did not feel that their child's degree of learning difficulty influenced their decision to give them diagnosis information (Metcalfe et al., 2011; Plumridge et al., 2010), but rather the condition's morbidity was a more decisive factor prompting disclosure.

Parents and guardians in our study were also motivated to disclose their child's diagnosis to explain their child's symptoms to them. Many parents reported talking to their child about their symptoms as they came up and used symptoms to motivate disclosure conversations. While the concept of disclosing to help a child adapt to their condition and understand their diagnosis has also been described in previous studies (Gallo et al., 2005; Kaneko et al., 2022), a unique aspect of the PWS diagnosis discussion is the prevalence of hyperphagia as a part of the PWS phenotype, and many parents in this study described that since eating is an everyday occurrence, they disclosed not only to help their child understand their symptoms, but because hyperphagia made disclosure inevitable. Parents and guardians in this study also shared their child's PWS diagnosis with them to support advocacy and empowerment for their child. Building on what has been previously found (Dennis et al., 2015; Faux et al., 2012; Goodwin et al., 2014), their sentiments included a desire to have their child feel comfortable in their own skin, be able to stand up for themselves, and build a positive self-image. This result may encourage parents who are contemplating disclosure to consider the positive outcomes associated with sharing diagnosis information.

Additionally, for some parents in this study, their child's questions, or lack thereof, guided their disclosure decisions. Previous studies have similarly shown that it may be a successful strategy to allow the child to guide disclosure conversations through asking questions or demonstrated curiosity (Dennis et al., 2015; Kaneko et al., 2022; McConkie-Rosell et al., 2009; Metcalfe et al., 2011; Plumridge et al., 2010), and some parents in this study expressed that older children might be more inclined to ask questions about their symptoms or doctor's appointments than younger children, displaying their interest and readiness to learn to their parent. Finally, parents and guardians in the study also reflected that involvement with the PWS community (e.g. support group events, social media group interactions) motivated them to disclose, reinforcing the previously described idea that support groups can promote a child's exposure to individuals like them (Dennis et al., 2015), and motivate disclosure.

4.2 | Methods for disclosure

In addition to describing motivating factors, this study also characterizes parents' methods and strategies for disclosure. Parents and guardians describe using multiple conversations over time and

reinforced that "disclosure" was not one conversation that took place. Across these multiple conversations, parents describe that they first built a foundation or "planted a seed" of knowledge, and then later built on that foundation. This "seed-planting" strategy has been described in previous literature, and echoes the idea that disclosure is "a full process, and not just an act" (Dennis et al., 2015; Faux et al., 2012; Forrest et al., 2003; McConkie-Rosell et al., 2009). A seemingly novel finding of this study is that parents and guardians approached disclosure conversations with the attitude of being a part of a unified team and expressed that their child reacted better to subsequent conversations when they believed that their parents had their best interest in mind. This study finding is not unexpected, but it may be useful for parents and guardians to consider when they are deciding how to first approach an initial disclosure conversation. Parents and guardians also reported that using matter-of-fact language and normalizing the child's differences was a good strategy for disclosure. These findings build upon previous research, which found benefit in ensuring that disclosing does not make the child feel any different than before (Faux et al., 2012), and studies which encourage explaining that everyone has challenges (Dennis et al., 2015; McConkie-Rosell et al., 2009). Study participants also emphasized that talking with their child while on the way to their appointments is another way for them to make the conversations about their child's diagnosis more matter-of-fact and a part of everyday life. Finally, participants also described helpful phrases that they have used when talking about their child's diagnosis with them and emphasize reinforcing to their child that their diagnosis does not define them, and that the diagnosis is just a part of their life, but not their entire identity. This finding is consistent with what has been found in previous studies, which describe that a child's diagnosis is something that makes them special, and does not define them or make them "weird" or "abnormal" (Dennis et al., 2015; Faux et al., 2012).

4.3 | Experiences with disclosure

This study also identified themes related to parent and guardian experiences with disclosure. First, many parents described that their child's PWS diagnosis was "just a part of life" due to the early age of diagnosis, and that their family has never known differently. This experience may be similar for other genetic conditions where the diagnosis is made early on in a child's life and suggests that some parents may experience disclosure as an inevitable and organic process. Additionally, parents and guardians in this study expressed significant self-doubt and anxiety regarding sharing diagnosis information with their child, which is similar to previous literature (Abi Daoud et al., 2004; Dennis et al., 2015; Plumridge et al., 2010). Most of participants' anxiety stemmed from uncertainty regarding their disclosure strategy and doubts about being able to answer all of their child's questions. Significantly, this parental self-doubt and anxiety seems to be related to, but psychosocially distinct from, parents' worries about their child's potential negative response to disclosure. Regarding these responses, study participants reflected both positive and negative reactions from their child. For some parents, disclosure seemed to help their child better understand their diagnosis, and resulted in stress-relief and lessened psychosocial difficulties. For others, disclosure increased sadness for their child, led to a decreased self-image, or allowed their child to make excuses for themselves. These contrasting outcomes have been described before, as some studies have found disclosure to be stress-reducing and psychosocially beneficial (Faux et al., 2012; Goodwin et al., 2014; Metcalfe et al., 2008; Plumridge et al., 2010) and others have illustrated concerns with negative responses to disclosure (Dennis et al., 2015; Franchi et al., 2023; Gallo et al., 2005; Holt, 2006; Kiely et al., 2020; Metcalfe et al., 2011). Although complex and multifaceted, this study captured the nuances of considering and responding to disclosure conversations.

4.4 | Study limitations

There are several limitations to this study that need to be considered. First, the majority of the study participants were White, biological mothers, and assigned female at birth, lessening the universality of the results. Second, there is a possibility of ascertainment bias, as participants were recruited primarily through the FPWR, and may inherently be more actively involved and supported within the PWS community. This increased involvement might result in parents being more knowledgeable about PWS and therefore more comfortable with disclosure conversations. Additionally, response rate information cannot be determined given that the number of people who interacted with or viewed the Facebook posting is unknown; an inherent limitation to social media recruitment. Because of these limiting factors regarding social media recruitment, the high amount of parent to child disclosure and study findings may not be representative of the PWS parent community as a whole, and the findings were likely influenced by the selective population from which the study recruited from. Third, recall bias may have impacted the interview responses from participants, as many of the parents and guardians who were interviewed were recalling conversations and events, which may have happened many years prior. Another limitation is that for respondents who were placed into the "No Disclosure" group; it is uncertain whether or not their affected family member truly is unaware of their diagnosis or was simply not informed of their diagnosis from the responding parent. These affected individuals may have learned of their diagnosis through an alternative route, such as by a healthcare provider or physician. This nuance was not captured in this study. Furthermore, while other disclosure-influencing factors were directly evaluated in the study survey, affected individuals' cognitive level or IQ status was not directly recorded in the survey and this may limit the quantitative analysis of this specific disclosure-influencing factor. Finally, although the utilized survey and interview guides were reviewed by individuals familiar with the study population and PWS, these tools were not validated.

4.5 | Future research

As this study was the first to assess how parents and guardians have shared information about a PWS diagnosis with their child, there are additional studies that could complement the findings. First, more exploration about the PWS disclosure process from the perspectives of the affected child themselves or of siblings of the affected child could enhance disclosure understanding and evaluation of how these disclosure conversations were received. Given that siblings seem to influence disclosure practices, it would be interesting to evaluate siblings' experiences with these conversations. Finally, although the interviewed parents felt that resource development for PWS disclosure would be challenging due to the diversity of each family experience, future research might consider a more targeted approach to evaluate the feasibility and benefit of the development of PWS-specific disclosure resources.

4.6 | Practice implications

This study emphasizes the unifying motivations, methods, and experiences regarding PWS disclosure, and medical professionals, such as genetic counselors, are well-suited to address many of the disclosure needs expressed by parents. First, parents in this study expressed feelings of self-doubt and anxiety with disclosure, and providers can help parents feel supported and empowered through the disclosure process by using empathy and active-listening skills. Additionally, since increased parental knowledge about PWS is correlated with disclosure, providing parents with PWS resources and factually correct information to increase their knowledge about PWS may give them confidence to address their child's questions and disclose diagnosis information, if that is what they desire.

4.7 | Conclusions

Using a mixed-methods approach, this study provides information about the motivations and influencing factors behind sharing PWS diagnosis information, and describes strategies that parents and guardians use when approaching these disclosure conversations. These research findings support that while PWS disclosure is multifaceted and complex, there are similarities among the experiences and motivations for disclosure across numerous parents and guardians. These unified findings may be validating and informative for parents considering disclosure of PWS or another rare genetic diagnosis, as parents can recognize that they are not alone in the diagnosis disclosure journey, and that their concerns with disclosure may be echoed in other parents faced with similar challenges.

AUTHOR CONTRIBUTIONS

Victoria F. Moy: Conceptualization; development of methodology; data collection; data analysis; manuscript writing. **Jessica J. Denton:** Conceptualization; development of methodology; data analysis; manuscript writing; manuscript review and editing; supervision. **Jessica E. Bohonowych:** Conceptualization; development of methodology; content expert; manuscript review and editing; supervision. **Theresa V. Strong:** Conceptualization; development of methodology; content expert; manuscript review and editing; supervision.

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CONFLICT OF INTEREST STATEMENT

Victoria Moy, Jessica J. Denton, Jessica E. Bohonowych, and Theresa V. Strong declare that they have no conflict of interest. Jessica E. Bohonowych and Theresa V. Strong are employed by the Foundation for Prader-Willi Research.

DATA AVAILABILITY STATEMENT

The data that support the findings of this study are available upon reasonable request from the corresponding author, but the data are not publicly available due to privacy or ethical restrictions.

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SUPPORTING INFORMATION

Additional supporting information can be found online in the Supporting Information section at the end of this article.

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