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Would I have Wanted to Know? A Qualitative Exploration of Women's Attitudes, Beliefs and Concerns about Non-Invasive Prenatal Testing for de novo Genetic Conditions after having a Child with a de novo Genetic Disorder

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Abstract

Non-invasive prenatal testing (NIPT) for a panel of 25 single gene disorders became available in Western Australia in 2020 and potentially may be able to test for panels of hundreds of disorders as is the case with reproductive carrier screening. How this information would be used by parents in a population screening model is unknown. We used a phenomenological approach to explore retrospectively whether mothers of children with single gene or chromosomal disorders would have wanted to know about their child's genetic diagnosis

prior to delivery. Themes were identified such as *having a child with a de novo disorder and effect on pregnancy outcomes in hypothetical situations, impact on family function, the diagnostic journey* and *personal growth.* These themes related to both the concept of expanded NIPT (ENIPT) and the situation of having a child with a de novo genetic disorder that could now hypothetically be detected through ENIPT. Opinions were divided about whether participants would have wanted to know about their affected child's condition, indicating any expanded NIPT testing panels would need to be offered in the context of an appropriate comprehensive counselling program. How this would be provided on a population screening level and the role of genetic counselling needs further exploration.

Keywords
NIPT; de novo disorder; prenatal diagnosis; decision making; lived experience

1. Introduction

De novo conditions are those which are not inherited from either parent, but result secondary to a mutation in the sperm or egg prior to conception or during early embryogenesis in the embryo itself. De novo mutations have varied age of onset, severity and prognosis. De novo mutations have been shown to be a major cause of severe early-onset genetic disorders such as intellectual disability, autism spectrum disorder, and other developmental diseases. Many de novo mutations are of paternal origin and increase with paternal age. The detection of the genetic defect underlying the phenotype establishes a genetic diagnosis that can be used to provide a prognosis and recurrence risk [1]. Carrier testing before pregnancy exists for many recessive and X-linked conditions and is now commercially available in several developed countries [2]. However, the prenatal detection of de novo conditions has been more challenging, until recently usually only recognised following abnormal fetal ultrasound features suggestive of a specific genetic disorder, such as a cardiac defect associated with 22q11 microdeletion syndrome [3]. The advent of non-invasive prenatal testing (NIPT) has led to a progressive increase in the number of fetal conditions able to be diagnosed. The initial NIPT screening panel consisted of testing for trisomy 21, 18, 13 and sex chromosome anomalies. Since 2017, NIPT has evolved further with expanded panels of deletion and duplication disorders available [4], although this is not without some controversy in relation to low positive predictive values and other ethical issues [5]. The ethical questions related to NIPT include how to balance access to testing, informed choice, potential for stigmatization or eugenic selection and managing counselling around the variable relationship between phenotype and genotype [6].

NIPT provides higher sensitivity and specificity for the common trisomies than either first trimester combined screening test using maternal blood and ultrasound and second trimester maternal serum measurements [7]. As a result, NIPT is acknowledged as having a low risk profile for women because it involves a single blood test and reduces the number of invasive diagnostic tests required to detect genetically-affected fetuses. However, there are limitations to NIPT. Despite its relative accessibility, the cost of NIPT is not universally covered in many countries and NIPT can be undertaken as a routine test without appropriate counselling for detecting serious

genetic disorders. Further expansion of NIPT could provide a screening risk assessment with lower predictive value for an increasing number of exceedingly rare genetic disorders. Few experts and so far, no national or international organisation, support the clinical implementation of expanded NIPT screening for conditions other than the common trisomies. At the International Society of Prenatal Diagnosis meeting in October 2020, delegates discussed the evidence in favour of extended NIPT to screen for conditions other than the common trisomies. Initially, 65% of delegates favoured expansion of the scope of NIPT, but after robust debate, the majority of delegates (59%) changed their view to requiring more evidence before clinical implementation of extended NIPT [8]. Nevertheless, commercial NIPT for a panel of 25 single gene disorders has become available [9] and has extended the prenatal screening capacity to some de novo genetic disorders (eg. achondroplasia, Costello syndrome).

In the context of the increasing availability of NIPT for a variety of different genetic disorders, most of which are rare and not generally known by pregnant women, an informed pre-test consent process is recommended to promote personal choice based on evidence [10]. A 2015 Australian study of NIPT showed a high level of NIPT awareness (62%) among pregnant women, but also demonstrated that women's understanding of screening processes and of the role of NIPT was limited [11]. Furthermore, in an American study of women who had received a low probability/negative NIPT result the level of informed choice was still only 44% [12]. A 2019 study from Canada reported that the relative ease of performing NIPT as compared with diagnostic testing (eg. amniocentesis or chorionic villus sampling), may reduce women's ability to make a personal and informed reproductive choice [13]. There are also ongoing concerns that the non-invasive manner of this testing process will lead to it becoming routinely offered by providers and accepted by parents without receiving adequate information of possible outcomes [12], which in the setting of population screening for an expanded panel of de novo conditions raises concern. In the Netherlands, where NIPT is offered within a national prenatal screening program, women value high-quality counselling that emphasises informed decision-making based on personal values and freedom to choose [14]. Women who participated in the Dutch TRIDENT 2 study rejected the view that NIPT is a morally responsible screening test for disorders that can only be prevented by termination of pregnancy [15].

More importantly, there is a paucity of information in the current literature about what women felt they would want to know about being pregnant with a child affected with a disability. It is a delicate question and difficult to answer, as one can only hypothesise from their current situation in that moment what their response will be. This response may change during different stages of life and be influenced by their parenting experience and indirect experience of other parents who have children with disabilities [16].

Given the expansion of NIPT beyond the common aneuploidies and microdeletion/microduplication disorders, we conducted this qualitative study to explore women's attitudes to the prenatal recognition of de novo genetic conditions, where they had previously delivered a child with such a disorder that was not identified prenatally. We aimed to address the complexity of prenatal test information needs and how to provide appropriate pre-test information to parents in the context of non-invasive population screening for multiple disorders in pregnancy.

2. Methods

This was a qualitative study to explore women's attitudes, beliefs and concerns about non-invasive prenatal testing for de novo genetic conditions conducted in Western Australia from 2015-2016. Participants were women who were at least 18 years old with English as their first language, who had previously delivered children with de novo genetic conditions. The women were recruited via social media, snowball sampling and websites (such as the Tuberous Sclerosis Association; https://tsa.org.au/wa-research-study/). We chose internet based platforms as 93% of social media users use Facebook, representing around 45% of the Australian population based on data from the Sensis Social Media Report May 2015 (Sensis, 2015).

Through purposeful sampling we identified healthy women (defined as women who perceive themselves without any physical or mental disability that impacted on their day to day living) who had a child or children with a de novo genetic disorder[1], and who did not know the child's diagnosis prenatally. We excluded women who had a child with trisomy 21, trisomy 18 or trisomy 13, as these conditions are currently screened for in pregnancy. Additionally, further analysis of the study data aimed to examine knowledge regarding de novo genetic conditions that may be detected with NIPT in the future, but that cannot currently be detected or have only recently been able to be detected.

We used a phenomenological approach to explore the lived experience of women who had a child with a de novo genetic disorder. This methodology involved in depth, semi-structured, face-to-face interviews, thematic analysis of transcripts to identify possible relationships between themes and arrangement of those themes into a systematic summary of common themes [17]. These interviews occurred before the commercial availability of NIPT for single gene disorders in our geographical area. All enrolled participants gave informed written consent and participation in interviews was voluntary. No incentives were provided.

During the interview participants were asked open-ended questions about aspects of their child's diagnosis, including whether they would have wanted to know if their child had a genetic disorder during their pregnancy. This question allowed a retrospective exploration of their experiences in raising a child with a de novo condition, the benefits and drawbacks they perceived and their expected choices in a future hypothetical pregnancy given the opportunity for early diagnosis.

The women were asked what they felt the positive and negative aspects of having a child with a *de novo* genetic condition were and about the diagnostic journey they had experienced. Furthermore, they were asked if they would have wanted to know about their child's diagnosis during pregnancy.

2.1 Data Collection

Semi-structured interviews were selected to enable participants to contribute as much or as little information as they wished and to elicit rich, in depth responses to questions. The interviews were conducted by an experienced genetic counsellor (SL), trained in non-directive client centred counselling. The use of non-directive and non-judgemental question style was implemented to

[1] De novo refers to a gene mutation present for the first time in an individual without either parent having the mutation.

reduce social desirability response bias and avoid participants perceiving there were 'correct' or socially desirable answers [18]. The interviewer's perspective, from working as a genetic counsellor, was that every parent has the right to make informed decisions about having a child with a disability. The interview schedule comprised primarily open-ended questions with some closed ended questions to confirm factual information. Interview duration ranged from 20 minutes to 75 minutes and took place in the participants' homes.

Interviews from the cohort of mothers with children with de novo conditions were conducted until data saturation was reached with no new themes emerging. Interviews were transcribed verbatim and de-identified for analysis.

2.2 Data Analysis

Transcripts were uploaded to NVivo10 (QSR International Pty; NVivo 10.0 edition) and coded following the Braun and Clarke (2006) methodology for thematic analysis (Braun & Clarke, 2006). Coding involved reading and re-reading a transcript, line by line, to identify key themes in the data. This process was repeated for each transcript and themes were identified and refined as coding progressed. An experienced qualitative researcher (RL) independently reviewed a sample of the transcripts to evaluate dependability and confirmability of the data (Lincoln & Guba, 1985).

2.3 Ethics

All procedures were in accordance with the ethical standards of the University of Western Australia for studies involving human participants and approved by the institutional Human Research Ethics Committee (RA/4/20/5077) and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards.

3. Results

Ten participants were recruited to the study and all transcripts were eligible for analysis, being clear and complete. The demographic details are shown in Table 1.

Table 1 Participant demographics.

Demographic Data		C1 (N = 10)
Age (years)	31-35	1
	36-40	1
	40-45	1
	>45	7
Martial status	Married	9
	Divorced	1
Annual combined income (AUD)	Less than $60,000	1
	$60,001 to $100,000	1
	$100,001 to $140,000	3

	Greater than $140,000	4
	left blank	1
Ethnicity	Australian Caucasian	9
	North European	1
Religion	Christian	5
	Other	1
	None	4
Highest level of education	Year 10 or equivalent	2
	Year 12 or equivalent	2
	TAFE*/ Vocational training/ apprenticeship	2
	University; Undergraduate degree	1
	University; Post graduate degree	3
Employed	Yes	8
	Home duties	2
Work Mode	Full time	1
	Part time	6
	Business Owner/ self employed	2

*TAFE; Technical and Further Education Institution

The participants ranged in age from 36 to 60 years with children aged from 15 months to 41 years of age.

The conditions affecting the children, their age and gender, and the age and gender of unaffected siblings are shown in Table 2.

Table 2 Ages and conditions of children (Age, sex, condition).

Identifier	Affected children (Age, sex, condition)	Unaffected children (Age, sex)
C1.01	36, F, Angelman syndrome	44, M/ 38, M/32, F
C1.02	26, M, Angelman syndrome	23, F
C1.03	14, F, Angelman syndrome	12, F
C1.04	41, F, Tuberous Sclerosis	43, F/ 35, M
C1.05	10, M, Tuberous Sclerosis	6, F/ 8, M
C1.06	7, F, Williams syndrome	6, F /4, F
C1.07	18, M, Duchenne Muscular Dystrophy	16, F
C1.08	36, F, Tuberous Sclerosis	33, F/ 31, F

C1.09	15 months, F, Angelman syndrome	3, F
C1.10	18, M, Duchenne Muscular Dystrophy	

Given the ages of the affected children ranged from 15 months to 41 years old, some women answered questions from a retrospective viewpoint whereas those women with younger children were speculating on their future. Despite these differences, four common themes emerged.

1. Having a child with a de novo disorder and effect on pregnancy outcomes in hypothetical situations
2. Impact on family function and relationships
3. The diagnostic journey
4. Personal growth.

3.1 Having a Child with a de novo Disorder and Effect on Pregnancy Outcomes in Hypothetical Situations

When asked if they would have wanted to know their child's condition, six women said they would have wanted to know about their child's condition and four said they would not have wanted to know. The six women who wanted to know were all over 40 years old and had children ranging from 10 years to 36 years. The women who did not want to know ranged in age from 31 years to 60 years and their children's ages ranged from 15 months to 41 years.

Of the four women who did not want to know, two explained they just wanted to enjoy the pregnancy and two felt they would have terminated the pregnancy and were glad they did not. There was no difference in the age range of the children in each of the 'yes' or 'no' groups with participants with both older and younger children being represented in each group. One woman explained she would not have wanted to know at this stage in her life but as her affected child grows older she doesn't know how she will feel in the future.

"I'm glad it happened the way it did because we are so in love with her, and although I'm very aware that we're in the very early stages of it, and it's very appropriate to have a one-year-old that's dribbling and who's in a nappy and all the rest of it, it might be a very different story when she is 18" C1.09

The participants who said they did want to know, gave responses such as they would have preferred to terminate the pregnancy or they were undecided about termination but would have liked to have known to be prepared.

"If I had known that I was carrying a child with a genetic disorder I don't know that I would have continued with the pregnancy." C1.06

Some women discussed what they felt they would do in a subsequent pregnancy, and for those that had subsequent pregnancies what they had experienced. Four women who had a child with a genetic condition said they would want to know if their unborn child had a genetic disorder. They felt they could not proceed with another pregnancy affected with a disability when they already had a child with special needs.

"Like if it was in my situation where I already had a disabled child and it was my second pregnancy, I would pay thousands to find out that that person was okay." C1.02

3.2 Impact on Family Function and Relationships

Various viewpoints were discussed relating to the condition affecting the child and the impact of the affected child on family function. As an example, one participant discussed the sleep disturbance seen in children with Angelman syndrome. The effect of sleep deprivation on the family and how it affects work, social functioning, behaviour of other children and relationships was unique to her child. However other women discussed losing friends due to their child's challenging behaviours, the impact of the affected child on siblings and relationship break downs between the parents of the affected child.

"The number of split families of special needs children is just phenomenal." C1.07

"Huge mental health impact and out of the marriages, two (of seven) survived and that was pretty much the percentage right across a lot of women that I met over that period of time." C1.08

Fear of the future was discussed by some participants concerned how their affected child would be looked after when they had passed away.

"I'm 60 next year and it's getting really hard to look after him because you've seen how big he is." C1.02

"Who's going to look after your child when you can't look after them anymore and is your child going to suffer by having that condition?" C1.08

Several participants discussed the impact an affected child had on the family finances. These impacts included cost for medical equipment and medications, and lost income due to a parent ceasing employment to become a caregiver. In all instances where a parent stopped work to become a caregiver, it was the mother. This also reduced the woman's potential superannuation contributions over the years of childcare (Superannuation in Australia is a compulsory system of allocating a minimum percentage of income into a fund to support financial needs in retirement. Contributions are invested to provide the best possible retirement outcome).

"A friend actually worked out that in the first sixteen years of a boy with Duchenne's life; it cost AUD$1.8 million to the family." C1.07

3.3 Personal Growth

All participants discussed the personal growth they experienced in relation to the positive aspects of having a child with a genetic condition. Tolerance and acceptance of differences in other people due to differences in their own child were emphasised.

"Of course there are positives because it does take your life on a different path and you do learn different skills, different levels of tolerance. You learn the reality of life. Yeah, you do learn a lot. Yeah, I guess that's probably it, in a nutshell." C1.04

"The loss that I now know that we would have suffered is quite unthinkable; and on a very selfish note the opportunity for personal growth, again, for both of us, for our other children who we are much better parents to." C1.06

There was acknowledgement that while having a child with a serious genetic condition was not inherently a positive thing, the experience of parenting that child and knowing that child as a person was considered an immensely positive affirmation.

"It can be the best thing that's ever happened to you in a [way]... Not the best thing that's ever happened to you but it brings out some of the best experiences that you're ever going to have." C1.10

While acknowledging that siblings of the affected child may have missed out on parental attention some of the time, several participants also discussed how mature the siblings were, how protective of their affected brother or sister they were and how they had developed tolerance to challenging behaviour or situations.

"I think in the most part, siblings also come out of this journey being more tolerant, more accepting, and all the rest." C1.01

3.4 Diagnostic Journey

Each woman experienced a different diagnostic journey, however common issues that emerged were not being listened to, having concerns dismissed, wrong diagnoses and the length of time between noticing 'something was wrong' and receiving the final diagnosis.

"Difficult baby, horribly horribly difficult baby, and I kept going back to the paediatrician and just saying something's up…I was really sick of having everybody pat me on the back and saying, 'Is this your first baby dear?' Or, 'Just put her in a room and close the door, she'll stop. Eventually they all do'." C1.06

When a diagnosis was presented it was usually in negative language for many parents, without presenting much hope for the future. The prognosis sometimes predicted a poorer outcome than was experienced by the child and parents.

"When my son was born we were told that he would never walk, he would never talk and now he's in mainstream schooling." C1.05

Misdiagnoses also occurred, extending the diagnostic journey and potentially increasing the distress of the parents and their underlying trust in their health care providers.

"However we were referred to genetics at (Hospital) and we had some tests for Fragile X, various things, which came back there was a false positive for Fragile X when she was two." C1.03

However there were positive aspects of being provided with a specific diagnosis such as finding support groups for parents of children with the same condition and adjusting expectations about what their child may be able to achieve in the future.

"I think we were fortunate because we'd gone seven years looking for an answer. And my metaphor was that we've been stumbling in the tunnel in the dark for seven years and someone just opened a window. We didn't get out the end of the tunnel but we could see our way through, so that was better." C1.03

4. Discussion

The past decade has seen a significant alteration in prenatal testing with the expansion of non-invasive testing for rare conditions and fetal testing in the absence of medical indications such as a prior family history or sonographic fetal abnormalities [5] both in Australia and overseas. The diagnostic capabilities provided by expanded NIPT regimens (such as potentially those for de novo genetic disorders) will likely place parents in the previously unchartered area of facing pregnancy management decisions for genetic conditions which they are largely unaware of and yet have the potential for significant long term disability. While de novo conditions as a group are highly variable, there are several relatively common conditions that can have the potential for severe outcomes. Conditions such as Angelman syndrome, as outlined in this study, or others not represented here such as Neurofibromatosis type 1, Duchenne Muscular dystrophy (which occurs

de novo or from X-linked inheritance) and Rhett syndrome can lead to significantly shortened lifespans, intellectual delay and physical disability. Parental decisions when faced with such diagnoses during pregnancies, as opposed to the previously more typical scenario of postnatal and childhood diagnoses, are influenced by multiple factors including the medical implications of the diagnosis, their personal expectations of caring for a child with disability, their views on pregnancy termination and societal influences [16]. The increasing trend for normalisation of the NIPT screening process [13] and "more is better" philosophy, has the potential for creating a reproductive conundrum between parental desire for knowledge about their fetus and the decision to terminate an affected pregnancy. Specific prenatal counselling prior to expanded NIPT programs needs to be evolved and matured to achieve parental reproductive autonomy.

This study was designed to assess the embodied experiential knowledge of women who have delivered children with prenatally unrecognised disabilities and to ascertain their retrospective views on whether they would have wanted to know about the condition prior to delivery. The phenomenological approach aimed to explore the lived experience of a small cohort of women who have children with de novo genetic disorders. It also aimed to explore themes relating to knowledge of prenatal testing and how it may be used, as a baseline for expanding subsequent research to parents who are provided with a diagnosis in pregnancy, where reproductive options are available.

4.1 Would I have Wanted to Know (Both with the Affected Child and Subsequent Pregnancies)

The participants in this study were divided between 'yes', they would have wanted to know, mainly as they would have terminated the pregnancies, and 'no', they would not have wanted to know as they did not want to change the outcome of their pregnancies.

Experience of disability is one facet of information people draw on when making a decision about continuing or ending a pregnancy with a fetal diagnosis of disability [19], but there are multiple other complex factors involved. A recent study [16] assessing parental decision making for a range of disabilities reported issues such as religious affiliation, cultural backgrounds, the severity of the specific condition, the economic impact of raising the child, expectations of achieving a socially desirable good life and societal stigmata as influencing their choices. Most women have not had prior experience of genetic or congenital malformation syndromes, and therefore the participants in our cohort represent a unique opportunity to assess attitudes about de novo genetic conditions potentially diagnosable by NIPT using their experiential embodied knowledge.

In the context of a hypothetical non-invasive test for hundreds of different genetic disorders, this divide over whether participants would or would not have wanted to know certain information should be addressed during an informed consent process, with the aim of promoting personal choice based on evidence [10]. Parental decision making around the use of NIPT is not a static process and involves not only their own experiential knowledge, but also the availability and use of knowledge from their perceptions of the societal use of NIPT, the expected emotional impact of genetic information and perceptions of the utility of genetic information [20]. These decision making processes tend to be separate from information provided by clinicians and are typically related to societal influences and views, the latter often not well defined for rare genetic conditions.

4.2 Family Function and Relationships

Fathers and partners were not interviewed for this study and it would be useful to explore their thoughts in the context of not being the primary caregiver in most families [21] but having a larger share of the burden of earning in the families interviewed. The mother is the main carer in 85% of cases in Australian families having children with a disability [21].

Participants in this study reported a higher rate of divorce was one outcome of having a child with a physical or intellectual disability. However there have been many conflicting studies about whether this is reflected in the population and recent studies have suggested the effect is quite small and is dependent on other factors such as age of having children, loss of a child, income and the effect on other siblings [21, 22]. All participants in this cohort were married except for one, who was divorced.

The negative financial impact on families has been acknowledged in previous studies on families of children with autism [23] and on households with a person with a disability [24]. There is a direct link between poverty and disability in Australia and other countries such as the United States [21]. Participants in our study discussed their experience of having to become the primary caretaker and that continuing with existing career paths was not possible, resulting in a loss of superannuation for the mother. In the event of a marriage breakdown this could significantly affect the mother's retirement prospects as well as potentially lead to emotional outcomes such as regret or blame.

4.3 The Diagnostic Journey

The diagnostic journey for parents of children with rare disease involves multiple interactions with health care professionals, difficulty being heard or acknowledged and false diagnoses during this time [25, 26]. Previous studies have observed parental attitudes as more positive in terms of raising a child with disabilities than health care providers [16] and this theme was evident in some of the participants' comments in our study. Challenges during the diagnostic journey are being recognised and attempts to ease the burden on parents of children with rare disease during this period of their life are currently being addressed [27].

In cases presented here, parents without a diagnosis for their affected child were still offered prenatal testing for other conditions in pregnancy, and in most cases, accepted. Since none of the participants had a diagnosis for their affected child before having other children, they justified prenatal testing to lessen the chance of having another child with a different, unrelated disability to their affected child. Some of the older parents reported not having a genetic test for their affected child, or one being developed through their diagnostic journey where it previously had not existed.

4.4 Personal Growth

The theme of personal growth, as an individual and as a family unit, was raised by all mothers in our sample. Many of the mothers also raised the positive effect of having a child with a disability on the siblings. They recognised that while having an affected sibling had been difficult for their children, they also reported that their unaffected children have high tolerance levels and empathy that they may not have otherwise had.

Post diagnostic growth (PDG) in the context of having a child with a disability has been previously explored [28]. While having a child with a diagnosis of a disability can still be distressing, overcoming parenting challenges and building resilience is related to PDG [28]. The importance of acknowledging PDG in the parents of children with disabilities is not to downplay the negative aspects of their child's diagnosis but to enable a positive framework for their experience. Parents who could identify the benefits or positive aspects of having an affected child are associated with having higher levels of mental well-being [29].

Our study dispels a 'one approach fits all' strategy in the context of offering a non-invasive test for complex disorders. For population screening, it is likely that general practitioners (GPs) would provide the first line of contact for NIPT for single gene disorders. However the burden on GPs to communicate adequate information to make an informed choice and consent to a complex prenatal test may not be possible [12]. With the scope of NIPT growing rapidly, there is an urgent requirement to address the provision of clear guidelines and information for women contemplating expanded NIPT and to explore the role of genetic counsellors in the process.

4.5 Limitations

As with other qualitative research this study does not aim to represent a whole population but identifies themes that can be refined for use in larger quantitative studies. The sample of women is small and consists of well-educated women from a higher income bracket than average women in the community. This may reflect the mothers who have means or interest in participating in a time-consuming interview. We did not interview fathers or partners in this study, and this is an area for future research to evaluate their opinions and beliefs. The women in this study all have experiential embodied knowledge, obtained from personal lived experience, and we recognise this is not the usual clinical scenario as more rare genetic disorders occur for women with no prior experience. However, as NIPT for rare conditions expands, prenatal diagnosis will become more common and the use of the experiential knowledge may assist health care providers in developing information counselling for NIPT provision. We plan to extend this research to a larger cohort of women with no prior experiential knowledge to further develop prenatal counselling information for expanded NIPT.

5. Conclusions

This study highlights the importance of integrating women's experiential knowledge into strategic proposals for expanded NIPT for de novo genetic disorders. Guidelines and policies should integrate support for patient autonomy while acknowledging the previous lived experiences of women with children who have de novo genetic disorders.

Author Contributions

Authors Sarah Long and Peter O'Leary confirm that they had full access to all the data in the study and take responsibility for the integrity of the data and the accuracy of the data analysis. All of the authors gave final approval of this version to be published and agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

Competing Interests

The authors have declared that no competing interests exist.

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