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Perspectives of Patients and Clinicians on Reproductive Health Care and ADPKD

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Introduction: Family planning and reproductive care are essential but complex aspects of lifecycle management for individuals with autosomal dominant polycystic kidney disease (ADPKD), given the potential genetic transmission and pregnancy-related complications. In this qualitative study, we studied the experiences and perspectives of patients with ADPKD and clinicians to identify areas for potential improvement in reproductive lifecycle care.

Methods: Focus group discussions (FGDs) were conducted in the Netherlands with patients with ADPKD, both men and women, who had children through varied reproductive choices; and clinicians, including (pediatric) nephrologists, obstetric gynecologists and geneticists. Thematic analysis, utilizing a grounded theory approach, was performed on verbatim transcriptions of recordings, followed by consensus discussions to finalize themes.

Results: Nine focus groups involving 31 participants (16 patients and 15 physicians) identified 6 key themes. These included the need for timely and comprehensive information dissemination from puberty on, understanding patient-specific decision-making factors, improving tailored psychosocial guidance and communication, the need for systematic efforts to take care of missed (minor) at-risk patients, addressing inequities in access to care, and improving multidisciplinary collaboration.

Conclusions: This study represents the first qualitative study of patient and physician perspectives on reproductive lifecycle care for ADPKD. We present valuable insights into factors influencing patients' reproductive decision-making, a comprehensive comparison between the perspectives of patients and clinicians on family planning and follow-up care of minors at risk for ADPKD, and recommendations for enhancing overall care quality. Incorporating these insights into clinical care could enhance patient-centered care and foster interdisciplinary collaborations to further improve the quality of reproductive health care services for individuals with ADPKD.

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ADPKD presents significant challenges for family planning due to genetic, ethical, and pregnancy-related concerns. In addition, the absence of consensus on early disease detection in children at risk for ADPKD further complicates the landscape. ADKPD is the most prevalent hereditary kidney disease, affecting approximately 12.5 million people worldwide.[1,2] The disease, mostly caused by pathogenic variants of the *PKD1* or *PKD2* genes, is characterized by the development of cysts in the kidney, leading to kidney parenchyma damage, hypertension, proteinuria, and progressive kidney failure. Next to this, liver cysts and/or intracranial aneurysms can occur.[2] Diagnosis of ADPKD is typically established by reviewing a combination of symptoms, imaging to detect cysts, and family history. Molecular genetic testing plays an integral role, not only to confirm the genetic type of ADPKD, but especially in cases with uncertain clinical diagnosis or atypical disease manifestation, and can be critical when evaluating treatment options or making reproductive decisions.[3–5]

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Reproductive Care for Patients With ADPKD

Reproductive decision-making for patients with ADPKD can be complex and options depend on local availability. Patients have a 50% risk of transmitting the disease to their children. Options include natural conception (and accepting the risk of an affected child), *in vitro* fertilization with preimplantation genetic testing (PGT) to prevent disease transmission, alternative family planning solutions (i.e., oocyte/sperm donation, adoption, abstaining from having children) and prenatal diagnostic testing after conception and termination of pregnancy in case of an affected fetus.[6] Of note, in the Dutch health care system, PGT is legal and reimbursed (up to 3 cycles).[7] Women with ADPKD face higher risks of developing pregnancy complications such as preeclampsia, fetal growth restriction, and preterm birth. These risks increase with advancing stages of kidney disease.[8–10] In the Dutch health care system, midwives typically manage low-risk pregnancies, whereas physicians handle high-risk cases, including those involving ADPKD.[11] In addition, for Dutch nephrogenetic patients, only clinical geneticists provide genetic counseling, not genetic counselors.

After delivery, there are different approaches to follow-up with children who may inherit ADPKD. The onset of disease symptoms primarily occurs in adults and is rare during childhood. Currently, there is no international consensus on presymptomatic diagnostic testing for minors. Recommendations vary from avoiding diagnostic testing and endorsing blood pressure monitoring,[5] to supporting ongoing surveillance or immediate diagnostic testing as equally valid clinical approaches.[12,13]

In the Netherlands, there is hesitation to perform presymptomatic diagnostic testing in children due to the rarity of symptoms in childhood and a current lack of treatment options for children. There is a preference to wait until adulthood when children can decide on diagnostic testing for themselves, thus preserving the child's autonomy.[14] However, a recent report revealed that up to 20% of children with ADPKD may develop hypertension and proteinuria at a young age, disease manifestations that may go unnoticed.[15] To prevent neglect of treatable disease manifestations, recently new monitoring recommendations were introduced, advising blood pressure and proteinuria surveillance from age 5 for children at-risk, without establishing a genetic diagnosis.[16,17]

Addressing the Knowledge Gap in Reproductive Care and Family Planning for Patients With ADPKD

There is no consensus guideline that specifically addresses the reproductive care or family planning needs of patients with ADPKD. The existing expert consensus primarily focuses on providing genetic counselling, including discussing PGT with prospective parents.[5]

Previous, mostly quantitative studies, have not thoroughly explored the experiences of patients and clinicians concerning family planning in ADPKD.[18–21] In addition, whereas various studies have documented differing views between patients and clinicians, there is a noticeable absence of comparative analysis within the same geographic context.[20] Existing qualitative studies have focused on presymptomatic testing for individuals at risk of ADPKD[22] and were conducted prior to milestones such as the US Food and Drug Administration approval of tolvaptan,[23] the 2015 Kidney Disease Improving Global Outcomes consensus report,[5] its expected 2024 update,[24] and the international consensus report on follow-up of children at-risk for ADPKD.[12]

To address this, our qualitative study using focus groups aimed to explore the attitudes of patients with ADPKD and clinicians toward family planning and reproductive care, including the approach to follow-up with minors at risk. The goal is to lay the groundwork for a patient-centered family planning guide that addresses the comprehensive needs of patients throughout the entire spectrum of reproductive care.

METHODS

This study examined the perspectives of patients and clinicians on family planning and reproductive care across 3 reproduction phases: prepregnancy, during pregnancy, and postpregnancy, which includes follow-up of children at risk for ADPKD. The study, conducted in the Netherlands, adhered to the Consolidated Criteria for Reporting Qualitative Research.[25] The University Medical Centre Utrecht medical ethics committee confirmed that ethical approval was not required (reference 22/849).

Study Design

A qualitative study design was adopted, following grounded theory methods.[26] FGDs were conducted according to qualitative research guidelines[27] among 2 study groups: patients and clinicians.

Participant Recruitment and Selection

Participants were recruited via multiple ways: (i) calls on the University Medical Centre Utrecht and the Dutch Association for Kidney Disease Patients social media channels; (ii) Dutch Kidney Foundation online patient forum (www.nieren.nl); and (iii) academic and general outpatient clinics for genetics, nephrology, and obstetrics in 6 different hospitals, both general and academic. For patients, the following inclusion criteria

were applied: (i) established clinical/genetic ADPKD diagnosis, (ii) aged ≥ 18 years, (iii) conceived in last decade, (iv) Dutch speaking, and (v) having the capacity to provide informed consent. For clinicians, the following inclusion criteria were applied: (i) practicing adult or pediatric nephrologist, clinical geneticist or obstetric gynecologist, recruited via the researchers' professional network; (ii) experience in family planning and reproductive counseling of patients with ADPKD; (iii) Dutch speaking; and (iv) having the capacity to provide informed consent. Eligible participants received an invitation, study information, and a consent form via e-mail, followed by telephone contact to confirm participation.

The composition of FGDs aimed to foster interaction through relatable experiences and to create a safe environment in which participants could freely express their views. Therefore, patient and clinician FGDs were organized separately. Purposive sampling was used to ensure a diverse representation of different perspectives regarding reproductive choices, experiences, and medical specialists. Patients were purposively sampled based on the following: (i) different reproductive choices (natural conception vs. *in vitro* fertilization/PGT); and (ii) gender, with different reproductive experiences (women that experienced the pregnancy themselves or men as partners). To secure psychological safety, patients with similar reproductive choices and genders were grouped together. This way, an open dialogue could follow allowing participants to freely share their personal experiences and perspectives. By organizing various FGDs based on different reproductive decisions and experiences, multiple perspectives and diverse data could be captured. The composition of clinicians' FGDs was sampled based on the following: (i) specialty (adult or pediatric nephrologists, clinical geneticists, obstetric gynecologists) and (ii) type of hospital: ensuring a diverse mix of clinicians working in academic versus general hospitals.

Data Collection

Prior to the FGDs, participants received an online survey (Castor software)[28] to gather sociodemographic and/or clinical information (Supplementary Material S1), facilitating the FGD's composition. From October to December 2022, 2-hour FGDs were held live (at the University Medical Centre Utrecht) or online (utilizing Microsoft Teams) based on participants' preferences. In all FGDs, an observing researcher was present. The FGDs consisted of 2 parts. First, the participants engaged in discussions about the 3 reproductive phases: prepregnancy, during pregnancy, postpregnancy, which includes follow-up of minors at risk. These discussions were guided by a detailed topic list, developed from a literature review and expert consultations and reviewed by the Dutch Association for Kidney Disease Patients (Supplementary Table S1).

During the second part, participants were asked to review topics identified as most important during the initial discussion using Lucid chart, an online diagramming tool.[29] This process allowed for refinement and further exploration of the discussed subjects. The FGDs were directed by a facilitator, while an observer made field notes, evaluating both the process and participants' nonverbal communication. All FGDs were audio recorded with consent and were transcribed verbatim.

Data Analysis

Sociodemographic data were summarized using IBM SPSS Statistics Version 29.[30] NVivo 12 software was used for thematic qualitative data analysis.[31] Based on the transcripts, data were coded in an inductive manner. Themes were derived from the data following grounded theory principles.[26] Transcripts were systematically and independently coded by 2 researchers to identify themes through: (i) independent open coding (breaking down data and labeling with codes), (ii) axial coding (categorizing codes), and (iii) grouping codes into themes. Coding was an iterative process, constantly reviewing and adapting the codes. Coding decisions and derived themes were discussed within the research team ensuring investigator triangulation. Recruitment was stopped at data saturation: when no new information was obtained from FGDs. Prefinal themes were circulated to participants via e-mail to enable feedback, which was used to finalize themes. Quotes were translated into English using DeepL software (DeepL SE, Cologne, Germany).

RESULTS

In total, 31 participants (16 patients and 15 clinicians) participated in 9 FGDs, with discussions lasting between 108 and 123 minutes. Informed consent was provided by all. The participants represented diverse genders, reproductive choices, experiences, preferences, and medical specialties (Table 1), reflecting the multidisciplinary nature of ADPKD care in both academic and general hospital settings.

Themes

Thematic analysis identified the following 6 main themes across all reproductive phases: (i) timely information, (ii) patient factors influencing decision-making, (iii) psychosocial guidance and communication, (iv) care for patients at risk, (v) inequity in access to care, and (vi) multidisciplinary collaboration. In Figure 1, we provide a schematic overview of the

Table 1. Baseline characteristics

Parameter	Frequency, *n* (%)
Patients	*N* = 16
Gender	
Male	6/16 (37%)
Female	10/16 (63%)
Age, yr, median (IQR)	38.5 (6.3)
Country of birth	
The Netherlands	16/16 (100%)
Religious	2/15 (13%)
Christian	2/2 (100%)
Highest level of education	
Vocational education	1/16 (6%)
Higher vocational education	7/16 (44%)
Pre higher professional education / pre university education	3/16 (19%)
University	5/16 (31%)
Employment	
Paid employment	16/16 (100%)
Experiencing signs/symptoms of kidney disease in daily life[a]	9/16 (56%)
Hypertension	2/9 (22%)
Abdominal pain / fullness	6/9 (67%)
Side effects Tolvaptan	2/9 (22%)
Side effects immune suppressive medication after transplantation	2/9 (22%)
Other	3/9 (33%)
Member of Dutch patient society for kidney disease (NVN)	4/16 (25%)
Knowledge of ADPKD occurring in family before first pregnancy	15/16 (94%)
Visited clinical geneticist[b]	13/16 (81%)
To discuss reproductive options	10/13 (77%)
To discuss ADPKD diagnosis & implications for family	2/13 (15%)
Other	1/13 (8%)
Received prepregnancy information on heredity and pregnancy complications, by	15/16 (94%)
Obstetric gynecologist	3/15 (20%)
Nephrologist	10/15 (67%)
Clinical geneticist	7/15 (47%)
Received information on PGT, by	15/16 (94%)
Obstetric gynecologist	0
Nephrologist	9/15 (60%)
Clinical geneticist	10/15 (67%)
Other	2/15 (13%)
Reproductive decisions[c]	
Natural conception	9/16 (56%)
Preimplantation genetic testing (PGT)	7/16 (44%)
Prenatal diagnostic testing for ADPKD	1/16 (6%)
Amniocentesis	1/16 (6%)
Chorionic villus sampling	0
Termination of pregnancy after PND for ADPKD	0
Age of children, median (range), yrs	3.0 (0–18)
Offspring	
Diagnosed with ADPKD	1/14 (7%)
Unknown	9/14 (64%)
Not affected (all after PGT-pregnancies)	4/14 (29%)
Offspring at risk for ADPKD visiting pediatric nephrologist	1/9 (11%)
Children above age of 5 yrs	1/4 (25%)
Clinicians	*N* = 15
Age, median (IQR)	45.0 (13)
Country of birth	
Netherlands	14/15 (93%)
Morocco	1/15 (7%)
Religion	
Not religious	10/15 (67%)
Christian	3/15 (20%)
Islam	1/15 (7%)
No statement	1/15 (7%)
Employed in	
Academic hospital	10/15 (67%)
General hospital	5/15 (33%)
Involved in ADPKD research	3/15 (20%)
Part of medical expert team	4/15 (27%)
Medical profession	
Adult nephrologist	5/15 (33%)
Pediatric nephrologist	3/15 (20%)
Obstetric gynecologist	4/15 (27%)
Clinical geneticist	3/15 (20%)
Yrs working as medical specialist, median (IQR)	10.8 (12)
Sees this number of patients with ADPKD per yr, median (IQR)	10.0 (13)

ADPKD, autosomal dominant polycystic kidney disease; IQR, interquartile range; NVN, Dutch Association for Kidney Disease Patients; PGT, preimplantation genetic testing; PND, prenatal diagnostic testing.
[a]Outside of pregnancy;
[b]Up to this time, not specifically before the first pregnancy;
[c]Up to this time, when asked if one of the following reproductive decisions was ever taken.

derived themes and their cohesion. As illustrated in Table 2, distinct important themes emerged for patients and clinicians during the different reproductive phases. In Table 3, illustrative quotations per theme are provided. In Figure 2, we present a reproductive care timeline for clinicians, aligning with both patients' and clinicians' needs and suggestions based on the findings of the study.

Theme 1: Timely Information

Prepregnancy. Both patients and clinicians emphasized the importance of receiving early information about heredity, reproductive options, prepregnancy counseling, and diagnostic or monitoring options (quote Q1 and 2). Reproductive options include natural conception, PGT, alternative family planning solutions (i.e., oocyte/sperm donation, adoption, abstaining from having children) and prenatal diagnostic testing with termination in case of an affected fetus. There was consensus on the need for these topics to be discussed proactively from puberty on, prior to patients having an "active" wish to conceive (Supplementary Quote [SQ] 1 and 2 and Supplementary Table S2). Not all patients were aware of the option of PGT before their first pregnancy. In subsequent pregnancies, almost all patients (15/16) had received information on PGT. Patients should be informed on reproductive options and care by their responsible health care provider at least globally and should be referred to a specialist (nephrologist/obstetrician/geneticist) for further

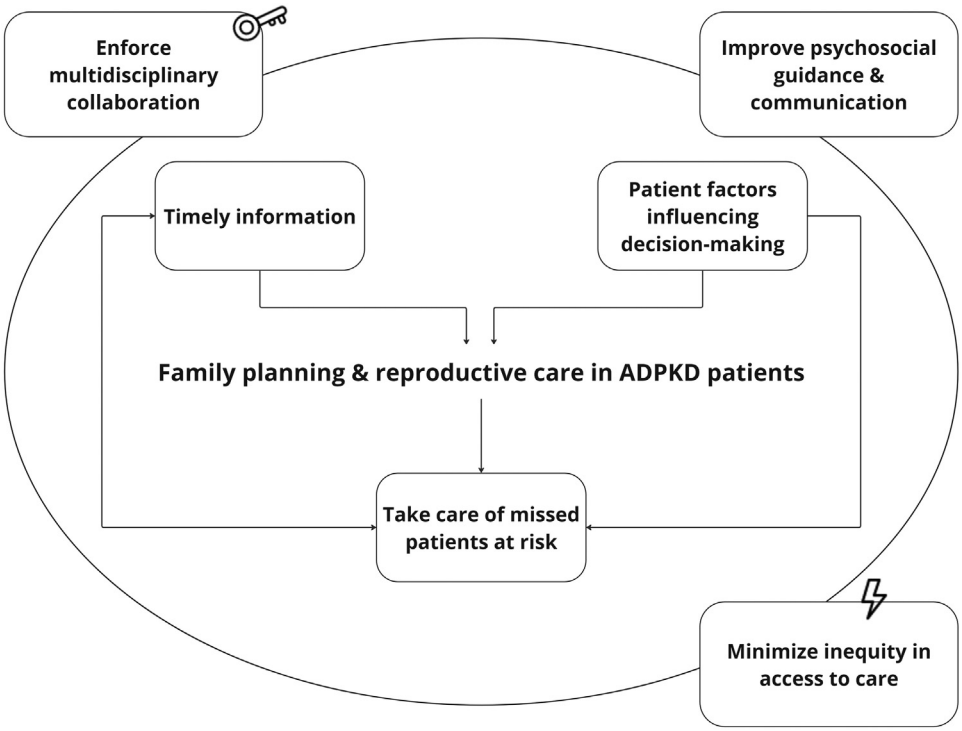

Figure 1. Overview of the main themes and their connection. In this qualitative study on family planning and reproductive care in ADPKD patients, 6 main themes were found. First, to improve reproductive care for patients with ADPKD, "timely information" from puberty onwards is key. Second, insight in "patient factors influencing decision-making" is necessary to provide tailored guidance to patients during this process. Third, this "psychosocial guidance and communication" can be improved throughout the entire reproductive period. Fourth, it is important to "take care of missed patients at risk": a patient group with 50% risk of inheriting ADPKD, resulting from natural conception in individuals diagnosed with ADPKD. In addition to monitoring, with the possibility of early treatment of disease manifestations and information, these patients should also receive timely information on family planning and reproductive care. Fifth, encompassing the entire lifecycle, there is an undesirable situation of "inequity in access to care" which should be minimized. Lastly, encompassing the entire lifecycle as well, (reproductive) care for ADPKD patients could be improved by enforcing "multidisciplinary collaboration" and consensus. ADPKD, autosomal dominant polycystic kidney disease.

specialized counseling when applicable. Earlier information allows patients time to reflect, plan, prepare and make well-informed decisions regarding family planning (Q2 and SQ3). The results also revealed a lack of reproductive counseling for patients at risk for ADPKD, pointing to a need for improved awareness of available reproductive options. Patients and clinicians also noted a shared feeling of responsibility regarding addressing family planning and informing children at risk (SQ15).

During Pregnancy. Patients indicated that timely information on the logistics of antenatal visits and delivery was important, as well as clarity about which health care providers were responsible for their reproductive care (SQ14). Several female patients with ADPKD noted they had received insufficient information about the potential impact of maternal medications on their babies during pregnancy and breastfeeding (SQ5).

After Pregnancy, Offspring at Risk. Postpregnancy discussions highlighted the need for timely information on monitoring children at risk of ADPKD, considerations for future pregnancies, and contraception.

During all Reproductive Phases. Patients indicated that during all reproductive phases, timely information on possibilities of psychosocial support should be provided (Q1 and SQ4).

Theme 2: Patient Factors Influencing Decision-Making

Prepregnancy. Patients shared their diverse experiences and factors influencing their reproductive and diagnostic decisions (Q3–9). Family history and perception of disease severity played an important role in reproductive, diagnostic, and monitoring choices. Some patients highlighted financial concerns, particularly fears about obtaining a mortgage or life insurance if diagnosed with ADPKD (Q3). Conversely, others found these fears unfounded after diagnosis (SQ6). Another argument against having a diagnosis was doubts about the added value for current treatment options (Q4). In contrast, others felt a genetic diagnosis offered them a greater sense of security and autonomy by having more insight into their reproductive choices and the disease's prognosis (SQ7). Furthermore, patients experienced diagnostic pressure during

Table 2. Overview of six main identified themes and subthemes during reproductive care

	Prepregnancy	During pregnancy	Post pregnancy
Timely information			
Patients & Clinicians	• Early proactive discussion and timely information from puberty onwards, regarding: o Reproductive options & prepregnancy counselling o Diagnostic & monitoring possibilities future child	• Timely information on: o Diagnostic options child during pregnancy & after delivery	• Timely information on: o Follow-up/monitoring child at risk o Future pregnancies & contraception
• Timely information on possibilities psychosocial support			
Patients	• Less dependent on doctor when properly informed	• Timely information on: o Antenatal visits & delivery, clarity on responsible health care providers o Consequences medication use during pregnancy for baby & breastfeeding	
Clinicians	• Ensure uniform and equitable information dissemination (including patients at risk for ADPKD)		
Patient factors influencing decision-making			
Patients	• Variable experiences & choices reproductive & diagnostic decision-making • Family planning causes time & diagnostic pressure • PGT-trajectory burdensome	• Prenatal diagnostics and termination of pregnancy rarely chosen option	• Parents wish to avoid burden for child, quality of life is the most important aspect • Variable opinions about follow-up minor at risk
Psychosocial guidance and communication			
Patients & Clinicians	• Social work support: o During decision-making & assisted reproductive treatment useful, but not necessary for all patients	• Intensified monitoring during pregnancy: mixed feeling safety & strain • Some patients desire to 'de-medicalize' pregnancy with positive & personal approach • Shared care options appreciated • Unclear who is responsible health care provider: insecure feeling	• Social work support: o Providing tools in explaining disease & implications minors at risk
• Psychosocial support primarily by clinicians: acknowledge tense moments during reproductive lifecycle			
Patients	• More updates desired during waiting time PGT • Family planning is not private anymore & many steps in process • Autonomy personal reproductive choice important		• Closer follow-up 6-wk post-ADPKD pregnancy desired
• Fellow peer support, partner & surroundings, open & clear communication is important			
Clinicians	• Patients' reproductive choice is personal • Despite uncomfortable feeling important to discuss family planning proactively	• Do not forget 'basic' pregnancy care	• Variable opinions on "follow-up & ADPKD diagnosis burden" minors at risk
• Guide patients following their experience of severity & burden of disease, quality of life for patients (at risk) most important			
Care for patients at risk			
Clinicians	• Young patients represent a missed group regarding reproductive & prepregnancy counselling	• Missed group during ADPKD-pregnancy, when no (clear) diagnosis is established	• Variable opinions & experiences follow-up advice minors at risk and most recent monitoring advice is not widely accepted • Follow-up can serve as safety net for the missed group of patients at risk for ADPKD
• Need to make systematic efforts to take care of missed patients at risk			
Inequity access to care			
Patients & Clinicians	• Patients at risk for ADPKD are a missed group • Interpatient differences exist in health literacy, finances, accessibility of dedicated care centers, support systems, family's provided information • Patient is dependent on clinician • Interclinician & interregional differences exist in experience, prepregnancy counseling referrals, adequate antenatal monitoring, advice follow-up minors at risk, multidisciplinary collaboration		
Multidisciplinary collaboration			
Clinicians	• Currently (inter)national differences exist in multidisciplinary reproductive lifecycle care • (Inter) national, well-implemented, multidisciplinary consensus (reproductive) ADPKD lifecycle care would help clinician & patient		

ADPKD, autosomal dominant polycystic kidney disease; PGT, preimplantation genetic testing.
Themes are stratified for (i) patients, clinicians, and both patients and clinicians, (ii) 3 reproductive phases: prepregnancy, during pregnancy, and postpregnancy.
Bold black lines represent subthemes that concern the entire reproductive period (prepregnancy, during pregnancy, and post pregnancy)

Table 3. Illustrative quotations per theme

Theme	Illustrative quotations
Timely information	
Early proactive discussion reproductive options, prepregnancy counselling, diagnostic options, monitoring minors at risk and psychosocial support options	Q1. "What kind of information are you missing? […] That there are choices, that it exists. […] There has been no information but also no question whether I wanted […] social work or emotional support". (FPGT, 1) Q2. "You can inform patients who are not thinking about having children yet, so they bear something in mind, knowing they have options. It's always nice to make well-informed choices." (N4)
Patient factors influencing decision-making	
Variable experiences & choices diagnostic decision-making	Q3. "With the knowledge I have now, I wouldn't do it that way again (diagnosis of ADPKD at age 18). Because we've had so many problems with mortgages. And I just can't get an insurance." (FNC, 5) Q4. "If there is a treatment that ensures that it really stops (disease progression) that would influence that choice (diagnosis in childhood) tremendously. If you can ensure that your child is not going to feel the effects of a disease in his or her life, that would be decisive for me." (MNC, 3)
Variable experiences & choices reproductive decision-making	Q5. "Beforehand I was pretty adamant, 'if I can get it out, I'm going to do that' (PGT)… then you hear that it's not that promising, it was actually more intense than I expected. It's also worth a lot if we can just get (pregnant) lovingly and without stress and medical treatments. Then the risk (of transferring ADPKD to progeny) is 50%, but by the time (the children) are grown up, there are so many new developments. Now retrospectively, I sometimes struggle with a sense of guilt, I could have stopped it after all." (FNC, 2) Q6. "Particularly the severity of hormone treatment, my wife would have had to start taking certain medication to undergo embryo selection, we didn't want that" (MNC, 2) Q7. "Also a certain difficulty with the (embryo) selection itself. Not from a religious perspective, but that it had to be that way. They (people with disease) are also just people. It felt very wrong for us to terminate (a pregnancy) and to decide in advance. You're really selecting life". (MNC, 3) Q8. "For me it was just clear, if there's a chance that we will not pass it on, and stop it (transferring ADPKD) with this, then it's worth trying". (MPGT, 2) Q9. "I couldn't see (the option of) abortion (in case of carrying a child with ADPKD). Then you're pregnant, then you can't do that anymore. It doesn't work out. Not that I'm against it… I can't handle that emotionally." (FPGT, 1) Q10. [on the option of termination of pregnancy after prenatal diagnostic testing] "Then you would have a baby taken away (who is affected) that has not been proven to actually have symptoms." (FNC, 5)
PGT-trajectory mentally & physically burdensome	Q11. "That disappointed me so much. That it took so long. […] harvesting was very intense. I luckily got some morphine, but it went on and on […] After that I had the hyperstimulation syndrome […] over three years I found it very tough (FPGT, 2)" Q12. "What I found most difficult was that you had a transfer and then you had to wait. You just don't know whether it is a hit, or not. That uncertainty." (FPGT, 4)
Variable opinions follow-up minor at risk	Q13. "I think there is also a task (with the nephrologist). If he knows, there are two daughters, that something can be said about it via that route. What is possible and where you can get information even if you don't know yet if you have it (ADPKD)." (FNC, 1) Q14. "I'm really not going to send my child to hospital. That might also be because of my family history. Symptoms start a bit later in life (in our family). I want to keep him/her completely out of hospital for as long as possible." (FNC, 1) Q15. "If it doesn't help, it won't hurt, it's about your child. If you don't have to diagnose yet, if you still have the choice and he/she can get used to the fact that there is this risk of carrying something, I wouldn't be negative about it". (MPGT, 2)
Psychosocial guidance and communication	
Open and clear communication is important	Q16. "Just straightforward information, I think that helps. If you evade the issue, I don't think that benefits anyone and then you won't make any progress in the process with your partner of what choice you want to make". (FNC, 4)
Social work support during decision-making & assisted reproductive treatment, not necessary for all patients	Q17. "The social worker […] was specialized in this disease, she really knew everything […] That really helped me a lot. I had a lot of questions, I just didn't know how to find out (the answers). (FNC, 1) Q18. "It was […] like the doctor obligated us (to see a social worker), that it's a step you have to take during this journey." (MPGT, 1) Q19. "I think I would have liked it if someone had just asked me "how are you" instead of "how is your kidney function or pregnancy" […] "How are you. Are you still managing a bit? How are things going at home?" I think that's just important." (FPGT, 3) Q20. "Yes, I think that's underexposed (psychosocial needs) … I've never asked the question if they need that. Don't have time for it I guess." (N2)
Patients desire to "de-medicalize" pregnancy with positive & personal approach	Q21. "Sometimes I hear friends who have been under the care of a 'regular' midwife talking to each other: "oh did you see Annie?", "Yes Annie is really nice. And then Annie is also present at the birth of your child. That's a different experience, the hospital is more clinical, more distant." (FNC, 2)
Fellow peer support, partner & surroundings important	Q22. "The ability to seek people who are facing the same choice. Because who better to talk to than people who are in the same boat, who really understand. […] it must be in small groups because it's overwhelming when a whole group of people starts shouting and sharing things, vs. a small group and you feel like personal things are going to be discussed." (MNC, 3)
Care for patients at risk	
Young patients missed group in reproductive & prepregnancy counselling	Q23. "Which group we do miss? The ones whose parents are your patients, who don't know if they have something. I report to those parents […] there is the option of having ADPKD removed from your family line […]. I do advise them to discuss that with their children […] when their children start thinking about progeny, it's wise to discuss that with us in the early stages as well." (N1)
Variable opinions & experiences follow-up advice minors at risk	Q24. "We get signals that, from adult nephrology, there is some very easy thinking about the children, that they (children) don't need close follow-up. "This only becomes a problem when you're an adult". If they (patients) hear that repeatedly, I don't think that's a good thing." (PedN2) Q25. "ADPKD is a gradual disease where the gain is in being early. When things gradually go wrong, proteinuria, hypertension […] you want to be there in time. You don't want to wait until it's already progressed. I am convinced that (monitoring) will ultimately be beneficial." (PedN2) Q26. "That's just how you view burden. I don't think an annual blood pressure check and a urine check […] are burdensome for a child. […] I think that quite outweighs […] against keeping the choice open to know or not to know." (Gen3) Q27. "That's quite a large group and you're actually going to check half of them for nothing." (N5)

(Continued on following page)

Table 3. (Continued) Illustrative quotations per theme

Theme	Illustrative quotations
	Q28. (on monitoring and not presymptomatically diagnosing children) "I think it's a very paternalistic approach. If those people (parents from child at risk for ADPKD) want to know and with a bit of luck, the child has nothing at all. Well, then you don't need to do anything further at all […] (monitoring without establishing diagnosis) might even be pointless care right? Because you're monitoring something you don't need to (in case of a non-affected child). (ObGyn3) Q29. "We're kind of doing two tracks now... If we say that check-ups are needed during childhood, then we should also deviate from the consensus of not doing presymptomatic diagnostics. (Gen2)
Need to make systematic efforts to take care of missed patients at risk	Q30. "It's actually more important to catch those kids in an earlier stage. So that you can also counsel (on reproduction) more easily. Because now I think there are children wandering around with ADPKD, who don't know it yet […] who are completely out of sight." (N1)
Inequity in access to care	
	Q31. "I think most children are not monitored... do we all advise the same nationwide? That's pretty much the question." (Gen2) Q32. "I find it quite extraordinary that it (health care) depends on the region where you live. The fact that I happen to live in (city) means that I end up in a very specialized centre, while all the care in your (other patient) area is spread out. I find it shocking, because I think everyone should be able to get the same (care)." (FNC, 2)
Multidisciplinary collaboration	
	Q33. "What is most important to me, is the alignment of care […] across all reproductive stages, alignment between all health care providers in all those hospitals you walk into. […] That everyone knows what's going on with you and what choices are made." (FPGT 3) Q34. "It's a multidisciplinary problem and I think these things (guideline, monitoring advice) are set up more monodisciplinary […] (a multidisciplinary guideline) would help. Or that you […] at least ask other involved health care providers to review. And if you implement something like that […] it should also be made known. (ObGyn1)

reproductive decision-making, because for certain reproductive choices (PGT, prenatal diagnostic testing), a confirmed molecular diagnosis of ADPKD (in a family member) was necessary (SQ8). In addition, 1 patient noted that though she appreciated receiving timely information about various reproductive choices, she felt a sense of time pressure due to her clinician's repeated message of the importance of pregnancy before clinical aggravation of ADPKD (SQ9).

Regarding reproductive options, reasons for choosing natural conception varied, including a desire not to interfere with nature, optimism about future scientific advances and treatments, and avoiding the mentally and physically demanding process of PGT (Q5–7). Patients who opted for PGT aimed to prevent passing ADPKD to their children (Q8), despite the physical and emotional strains reported (Q11 and 12). Due to these strains, some patients were not sure they would choose to undergo PGT again (SQ10). Others had no doubts, given the security of minimizing the chance of disease transmission. Men with ADPKD who chose a PGT-trajectory with their unaffected partner shared an emotional burden and concern about the impact of the trajectory on their unaffected partners.

During Pregnancy. Most patients refrained from prenatal diagnostics that could lead to terminating the

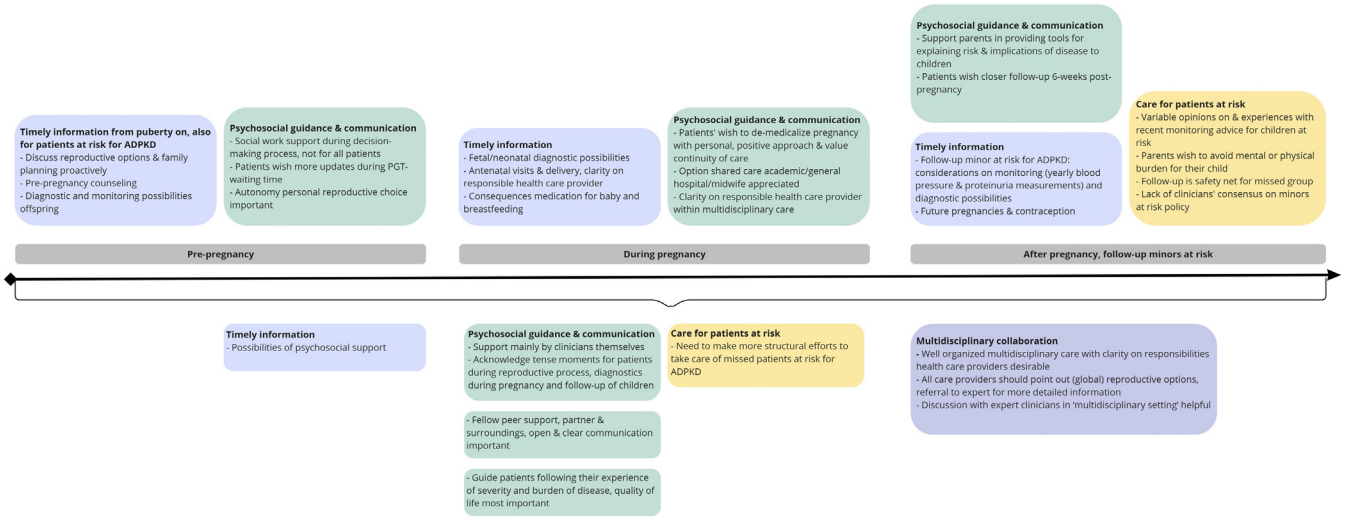

Figure 2. Timeline of reproductive care for clinicians, meeting patients' needs according to the main themes of this study. Themes above the black line are divided by reproductive phase (prepregnancy, during pregnancy, and postpregnancy). Themes below the black line are important during all reproductive phases. Regarding "patients at risk for ADPKD," it refers to individuals with a 50% risk of ADPKD that do not yet have an established diagnosis. ADPKD, autosomal dominant polycystic kidney disease.

pregnancy if the fetus was affected. This was because of the psychological impact (Q9), ethical concerns and the disease's variable expression (Q10).

After Pregnancy, Monitoring At-Risk Children. Most parents were not updated on the latest follow-up recommendations, which advice blood pressure and proteinuria surveillance starting from age of 5 years for children without establishing a genetic diagnosis. This is also reflected by the low number of patients in our study (1/4) with children aged >5 years that visit the pediatric nephrologist (Table 1). However, when we informed parents on this monitoring advice during the FGDs, parents had variable opinions regarding testing or monitoring their children for ADPKD (Q13–15). All agreed on avoiding mental or physical burden and ensuring quality of life. The perceived severity of the disease was one factor that influenced their decision regarding monitoring. Some parents were hesitant due to a desire for their children to have a normal and carefree childhood (Q14) and autonomy. Furthermore, treatment options for ADPKD in children (such as antihypertensive medication) were not clear for all parents. In contrast, other parents were in favor of early monitoring and treatment to manage disease manifestations, seeing no issue with surveillance during their child's early years (Q15). Regardless of the final decision, parents wanted to receive timely information on the follow-up advice for their children.

Theme 3: Psychosocial Guidance and Communication

Prepregnancy. Patients emphasized the importance of open and clear communication between patients and health care providers, noting that clinicians need to provide realistic scenarios concerning the advantages and challenges of different reproductive choices (Q16). Regarding the decision-making process, patients' experiences varied significantly. Whereas some struggled to reach a decision and found extra counseling support (by a specialized social worker) very helpful to align their choices with their personal values and preferences (Q17), others did not feel the need for this support (Q18, and SQ11 and 12). Some patients had no difficulty deciding in harmony with their partner, whereas others faced temporary conflicts due to differing opinions. In certain cases, decisions were influenced by others, despite a preference to make an autonomous decision (SQ13).

Lastly, patients expressed a desire for more frequent updates while on the waiting list for PGT treatment, including greater transparency about the wait times.

During Pregnancy. Some patients expressed a desire to "de-medicalize" pregnancy with a positive and more personal approach from their health care providers (Q21). For women with ADPKD, the intensified monitoring during pregnancy resulted in mixed feelings of both safety and strain. Despite the desire for regular checkups for safety reasons, patients wanted more focus on their personal experiences, the positive side of pregnancy, and continuity of care such as the care provided in Dutch standard midwifery service (Q21 and SQ14). Offering options for shared care between academic hospitals, general hospitals, and midwifery practices was well-received. At the same time, patients expressed a need for clear delineation of responsibility and communication among the various health care providers throughout the reproductive process. Clinicians pointed out the importance of not overlooking standard pregnancy care in addition to the specialized attention required for ADPKD, to provide a balanced approach.

After Pregnancy, Offspring at Risk. There was a call for more comprehensive support, especially with explaining the disease to at risk children. Some patients advocated for more rigorous follow-up during the postpartum period beyond the current policy of a single visit 6-weeks postpartum, and additional visits if clinically indicated.

During all Reproductive Phases. Participants noted the need for increased psychosocial support from their clinicians or additional sources during the reproductive process (Q19 and 20, and SQ12). The extensive number of health care providers involved in the family planning process was overwhelming for some, highlighting a need to clarify responsibilities and improve communication between providers, because patients felt insecure coordinating their reproductive care themselves (SQ14).

Although most patients did not have positive views on patient support forums, the FGDs conducted for this study, though confronting at times, were highly valued for offering information and emotional peer support (Q22), underscoring an unmet need.

Patients and clinicians both agreed that there is no "one size fits all" approach for patients with ADPKD and family planning. Individual guidance based on personal experiences of disease severity and impact on quality of life was the most important aspect to consider (SQ16). In line with this, all reproductive information needs to be tailored to each patient's needs and life phase, ensuring sufficient and fitting information without causing undue stress (SQ9).

Theme 4: Care for Missed Patients at Risk

Prepregnancy. Clinicians discussed the problem of late diagnoses of ADPKD in young people, leading to missed opportunities for early counseling and

intervention (Q23). It was noted that the new monitoring policy of annual assessment of blood pressure and proteinuria for undiagnosed individuals starting at the age of 5 years would bridge this gap, enabling early treatment and informed reproductive planning (Q24–25).

During Pregnancy. Similarly, clinicians expressed concern about women with ADPKD who are overlooked because they lacked a clear diagnosis, thus facing elevated risks for pregnancy complications and missing out on vital prepregnancy counseling.

After Pregnancy, Offspring at Risk. There were varying opinions among clinicians regarding the monitoring and genetic diagnosis of children at risk for ADPKD. A significant number were not aware of the new monitoring advice (Q24). For some, their follow-up advice was based on the severity of the family's disease. A number of clinicians considered monitoring as beneficial (Q25, SQ17, and SQ18). Although most participants thought monitoring blood pressure and proteinuria was not physically burdensome for children (Q26), there was concern about potential mental burden. Suggestions included making the process less stressful, for example, by taking blood pressure measurements at home or in a playful manner. Some clinicians were skeptical about the new monitoring policy, questioning its necessity and benefit compared to the potential stress on children, families, and the health care system (Q27 and SQ19). Others pointed out the complex issue of diagnosis timing, contrasting the autonomy parents have in prenatal diagnosis, versus safeguarding the child's autonomy after birth. Some questioned the value of monitoring disease manifestations without establishing a genetic diagnosis, advocating for widespread genetic testing in children to reduce unnecessary yearly monitoring among unaffected individuals (Q28 and 29).

Clinicians agreed on the need to make more systematic efforts to identify and provide (reproductive) care for the substantial number of undiagnosed patients with ADPKD (Q30). Proposed solutions included monitoring of at-risk children before the age of 5 years (SQ20), handing out information flyers to parents, and routinely asking their patients about potential disease symptoms occurring in families. Another option was an educational campaign for both parents and other health care providers (e.g., general practitioners, adult nephrologists) to raise awareness of the new monitoring recommendations.

Theme 5: Inequity in Access to Care

During all Reproductive Phases. Patients and clinicians pointed out that differences in how patients, clinicians, and multidisciplinary teams operate contribute to an undesirable situation of inequity in accessing reproductive care. Contributing factors include lack of monitoring or testing resulting in a high amount of undiagnosed ADPKD cases, various levels of patient proactiveness (willingness to seek medical care and information), health literacy, financial status, access to specialized centers of expertise, support systems, and information provided by their family (SQ21 and 22). In addition to regional differences among health care providers and multidisciplinary teams, there are disparities between clinicians regarding their expertise and approach to family planning. All these differences can lead to patients receiving conflicting or no information, potentially impacting their reproductive decisions and the lives of their children (Q31 and 32).

Theme 6: Multidisciplinary Collaboration

During all Reproductive Phases. Clinicians discussed the national and international differences that exist in the organization of reproductive care during the family planning process. Both clinicians and patients agreed on the need for well-structured multidisciplinary care, where the roles and responsibilities of all health care providers are clearly defined, along with clear lines of communication between them (Q33 and 34). In addition, there was a consensus on the importance of timely communication of all available reproductive options to patients, who should then have the opportunity to seek more in-depth advice from specialists if needed. One clinician noted that reproductive care for patients with ADPKD is a multidisciplinary problem and recommended establishing a platform for dialogue with a diverse range of experts to improve guidelines and patient care. Clinicians expressed support for an (inter) national, well-implemented, and multidisciplinary consensus guideline on reproductive lifecycle care for patients with ADPKD. Such guidelines, including uniform advice for follow-up of minors at risk, are necessary to provide optimized care and effective collaboration among health care providers who care for this patient group (Q34 and SQ23).

DISCUSSION

Main Results

This qualitative study describes the experiences, perspectives, and suggestions for improving reproductive care from both patients with ADPKD and health care providers throughout the reproductive journey, including follow-up of at-risk minors. We identified the following 6 key areas for improvement: (i) providing timely information from puberty on, regarding heredity, reproductive options, and family planning; (ii) understanding patient factors influencing

reproductive decision-making; (iii) improving personalized psychosocial guidance and promoting open communication with health care providers; (iv) systematically addressing the care of overlooked patients; (v) reducing inequity in access to reproductive health care; and (vi) establishing multidisciplinary collaboration and consensus.

Overall, the perspectives of patients and clinicians overlapped considerably. All participants agreed on the need for timely information provision from puberty on, before conception, and addressing disparities in access to care. Patients emphasized the need for psychosocial support, open communication, and peer support during decision-making, with appreciation being expressed for small-group interactions like in this study. Clinicians expressed the critical need to follow-up with at-risk children and debated the implications of the new monitoring policies. Annual monitoring of hypertension and proteinuria from the age of 5 years would prevent neglecting treatable disease manifestations and disease progression. However, not all clinicians were convinced to integrate this approach in daily clinic due to a perceived need for more data supporting this monitoring advice, lacking health care capacity and possible mental burden for children that will not develop the disease. Clinicians debated about when nondiagnostic follow-up ends and presymptomatic testing starts. Some clinicians advocated presymptomatic testing for minors at risk of ADPKD in the Netherlands, to prevent unnecessary monitoring of children that do not have ADPKD. This stance aligns with the most recent international consensus statement on the management of ADPKD in childhood, which endorses diagnostic testing in children after thorough counseling of both the children and their parents.[12,13]

In this study sample, only a quarter (25%) of patient participants who had naturally conceived children over 5 years of age had consulted a pediatric nephrologist or pediatrician for their child (Table 1). In the FGDs it became clear that this matched the lack of awareness about the new monitoring recommendations and their potential benefits for at-risk minors, instead of an active parental decision. Regardless of some parents being hesitant to monitor their children, parents wanted to be informed about the monitoring advice for children. Clinicians also displayed varying levels of awareness about these recommendations, revealing gaps in identifying and supporting at-risk patients.

Systemic initiatives to improve awareness and support for overlooked at-risk patients are needed, including better implementation of updated follow-up guidelines. Improvements are important in multidisciplinary collaboration and developing a multidisciplinary consensus guideline could benefit clinicians in guiding at-risk patients through the reproductive care pathway.

Comparison to Current Literature and Other Geographic Areas

This is the first qualitative study focusing on reproductive care and family planning in patients with ADPKD, offering insights into patients' and clinicians' perspectives. In the Netherlands, PGT is legal, reimbursed, and has an uptake after genetic counseling for monogenic kidney disease in a PGT-expert center of 53%.[7] Previous studies among patients with ADPKD in different geographic areas and jurisdictions, have found similar results regarding the high acceptability of PGT (50%–80% of patients with ADPKD opting for PGT were it available and funded).[18,19] The previously reported large variation in clinicians' views regarding acceptability of PGT for ADPKD was not confirmed by our study.[20] In our study, all participating clinicians viewed PGT as a valid reproductive option for patients with ADPKD. Next to the acceptability of PGT, previous studies among patients with ADPKD and other genetic diseases found similar results regarding the impact of family planning on patient's lives and challenges in accessing health care information.[32,33] Some patients desired to de-medicalize pregnancy and to receive more continuity of care like the Dutch standard midwifery care. This desire matches childbirth satisfaction studies in the general population, showing overall higher levels of satisfaction with midwife-led continuity of care compared to other models of care.[34,35]

Our study adds depth by uncovering both similarities and discrepancies between patient experiences and clinician perceptions. The discrepancies found around psychosocial guidance and communication match the findings of Baker *et al.*,[36] who reported patient distress and frustration due to insufficient acknowledgement of the personal physical and emotional impact of ADPKD by clinicians.[36]

Although recommended in the 2015 Kidney Disease Improving Global Outcomes consensus statement,[5] not all patients received reproductive counseling and information on PGT before their first pregnancy. This highlights the inequity of access to reproductive care and the need for timely information from puberty on, including for those at risk for ADPKD who do not yet have a confirmed diagnosis.

There was extensive debate on the ethical aspects of presymptomatic genetic testing of children and monitoring of minors at risk. The debated topics matched themes found in a qualitative study on presymptomatic

testing in ADPKD (in general, not specific for minors at risk): whereas presymptomatic testing allows for proactive health management, it can also bring tension due to prognostic uncertainty and psychosocial and financial impact.[22]

Although there is a trend toward a more proactive approach in monitoring and genetically diagnosing at-risk children,[12,13] this shift has not been fully integrated into Dutch clinical practice, leaving many clinicians and patients uninformed about these new perspectives.

Strengths and Limitations

To our knowledge, this is the first qualitative study fully focusing on reproductive care in ADPKD. A comparison between patients' and clinicians' perspectives on this topic, originating from the same geographic area and health care system, has not been performed before. To promote reliability, the authors were trained in qualitative research, transcripts were coded by 2 researchers independently and for investigator triangulation, prefinal themes were discussed in our multidisciplinary research team and validated with participants. Our study further benefits from a diverse purposive sample including both patients and health care providers and was conducted in a context with reimbursed and legally available reproductive options. However, generalizability may be limited for regions with different health care policies. Limitations that come with the focus group design such as overrepresentation of dominant voices, conformity due to group dynamics, moderator and confirmation bias cannot be excluded. In addition, our study has predominantly included highly educated, relatively well-informed individuals (both patients and clinicians) who were motivated to participate and spend time in this study. This selection bias should be considered when interpreting the themes of this study. Therefore, the study may have underrepresented certain perspectives, including those from patients who pursued termination of pregnancy. Nevertheless, patients and clinicians showed diverse perspectives within and across focus groups, providing a solid and rich base for the themes of this study. In addition, given the relatively well-informed participants in our current sample, we expect the identified themes for improvement to be at least as numerous if not more so in a broader sample.

Implications

We unveil valuable insights into family planning and reproductive care for patients with ADPKD, emphasizing the importance of collaborative and multidisciplinary approaches. Our findings lay the foundation for the development of comprehensive guidelines on reproductive counseling and family planning for patients with ADPKD, ensuring that their care is informed, supportive, and as effective as possible.

Future Research

Future research could explore the perspectives of patients with ADPKD who chose not to have children. Understanding their motivations could help clinicians provide support and guidance. Moreover, expanding this study through an international mixed-method design, including a larger and diverse sample of participants could offer more comprehensive and generalizable insights, and could facilitate the development of multidisciplinary consensus.

Conclusion

Family planning and reproductive counseling for patients with ADPKD can be complex due to the diversity of reproductive options, possible pregnancy complications, and the implications of disease inheritance. Improvements should be aimed at providing timely information from puberty on, improving psychosocial support and open communication, making systematic efforts to take care of missed patients at risk, minimizing inequity by improving access to reproductive care, and improving multidisciplinary collaboration. Clinicians can use the results of our study to better meet the diverse needs of patients with ADPKD in reproductive care.

DISCLOSURE

All the authors declared no competing interests.

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SUPPLEMENTARY MATERIAL

Supplementary File (PDF)

Supplementary Material S1. Online survey.
Table S1. Topic list focus group discussions.
Table S2. Supplementary quotes.

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