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# Exploration of decision-making regarding the transfer of mosaic embryos following preimplantation genetic testing: a qualitative study

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**STUDY QUESTION:** What are patients' reasoning and decisional needs in relation to the transfer of mosaic embryos following preimplantation genetic testing (PGT)?

**SUMMARY ANSWER:** This study identified four themes, which were patients' reasoning behind decision-making, their decisional needs, the influence of the mosaic embryos on the decision-making and the role of health professionals.

**WHAT IS KNOWN ALREADY:** To date, no study has investigated the reasoning of patients behind their decision-making and the influence of mosaic embryos.

**STUDY DESIGN, SIZE, DURATION:** This is a cross-sectional study using a qualitative approach. Twenty participants were interviewed, and recruitment was ceased when no new information was identified in the data analysis. It ensured a sufficient sample size for a qualitative study.

**PARTICIPANTS/MATERIALS, SETTING, METHODS:** Participants were females with mosaic embryos. Semi-structured in-depth interviews were conducted via telephone.

**MAIN RESULTS AND THE ROLE OF CHANCE:** Four themes were identified: reasoning behind decision-making, decisional needs, influence of mosaic embryos on decision-making and the role of health professionals. Potential risks of transferring mosaic embryos and prioritization of euploid embryos were the main reasons for not transferring mosaic embryos. A lack of alternatives, perceived benefits and risk tolerance were main reasons for transferring mosaic embryos. Patients reported that information on mosaic embryos, amniocentesis and termination was important to support their decision-making. Unmet needs relating to healthcare services and social support were reported. In addition, having mosaic embryos affected the patients' emotional and behavioural responses, discussions about prenatal testing, attitudes to termination and further IVF cycles and attitudes towards PGT. Health professionals were found to influence the patients' decision-making.

**LIMITATIONS, REASONS FOR CAUTION:** Participants were recruited through one clinic, which may limit the transferability of results. Also, patients' experiences in relation to financial aspects of PGT may not be relevant to other jurisdictions due to different healthcare policies.

**WIDER IMPLICATIONS OF THE FINDINGS:** The results may inform how clinicians provide healthcare services based on factors influencing patients' decision-making. Health professionals should be aware of the influence their attitudes can have on patients' decision-making and should present information accordingly. Also, providing all relevant information may help to facilitate informed decision-making. Provision of psychological support from professionals and support groups is also critical during the process of testing and transfer. Patients

have educational needs regarding mosaic embryos, and educational resources including decision aids in plain language are needed.

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**TRIAL REGISTRATION NUMBER:** N/A.

**Key words:** decision-making / decisional needs / preimplantation genetic testing for aneuploidies / mosaic embryos / qualitative study

## WHAT DOES THIS MEAN FOR PATIENTS?

This study looks at patients' reasoning for using mosaic embryos for IVF. Mosaic embryos consist of both normal and abnormal cells. The study also explores patients' needs when they consider using mosaic embryos for IVF. We interviewed 20 women, who had used or were considering using mosaic embryos for IVF. Four main themes emerged from the interviews. The first theme was reasoning behind decision-making. The risks of using mosaic embryos were the main reason for not transferring them. Not having any better choices, possible benefits and acceptance of possible risks were the main reasons for using mosaic embryos. The second theme was patients' needs in decision-making. Specifically, patients needed more information about mosaic embryos, what amniocentesis involved and the process of termination. Also, unmet needs were reported, which were follow-up healthcare services and social support. The third theme was the influence of mosaic embryos. We found that patients' emotions and behaviours were affected. Mosaic embryos also influenced patients' discussions with doctors on testing during pregnancy, their attitudes to terminating a pregnancy and assisted reproductive techniques namely IVF and preimplantation genetic testing. The fourth theme explored the influence of doctors on patients' decisions about using mosaic embryos. Our findings show that doctors should be aware of the effect of their opinions on patients' decision-making and should present information accordingly. Also, provision of all needed information may help in decision-making. Providing psychological support is additionally important during the process of testing and transfer. Educational resources in plain language should be provided to patients to meet their educational needs about mosaic embryos.

## Introduction

Mosaicism in embryos is defined as the presence of two or more distinct cell lines in an embryo, which occurs due to mitotic errors during early postzygotic cell divisions (Delhanty et al., 1993; Baart et al., 2006). Mosaicism for chromosomal anomalies (aneuploidy or large copy number changes) can be detected down to a level as low as 20%, depending on the methodology, and is routinely reported following preimplantation genetic testing (PGT), which includes PGT for aneuploidy (PGT-A), PGT for monogenic/single gene defects (PGT-M) and PGT for chromosomal structural rearrangements (PGT-SR) (Simon, 2017).

Embryonic mosaicism is associated with higher risks of adverse clinical outcomes including decreased implantation rates, reduced likelihood of establishing an ongoing pregnancy and increased risk of miscarriage (Simon, 2017). As a result, when mosaic embryos were first identified following PGT, they were not considered for transfer and were not included in the initial dichotomous reporting system; in fact, embryos with any chromosomal anomaly were not considered for transfer (Patrizio et al., 2019). However, the practice of excluding mosaic embryos from transfer changed in 2015, after two studies reported live births from mosaic embryos for the first time (Gleicher et al., 2015; Greco et al., 2015). After the first two live births had been reported, another two studies also reported live births resulting from mosaic embryos in 2016 (Gleicher et al., 2016; Maxwell et al., 2016). Also, in 2016, the Preimplantation Genetic Diagnosis International Society published a position statement to guide clinicians around the selection and prioritization of mosaic embryos for

consideration for transfer (Preimplantation Genetic Diagnosis International Society, 2016).

Despite the limited reassuring data about successful pregnancies and live births from mosaic embryos, making a decision about whether or not to transfer a mosaic embryo is challenging for couples due to the potential for adverse clinical outcomes and uncertainties regarding pregnancy outcomes. Other uncertainties relate to technological aspects of detecting mosaicism.

There is limited information about patients' decision-making regarding the transfer of mosaic embryos. To date, a study of Besser et al. (2019) is the only empirical study, which addresses this question by assessing outcomes following genetic counselling in the context of a mosaic embryo. Among the investigated participants, about 20% were undecided about what to do with their mosaic embryos even after genetic counselling (Besser et al., 2019). Based on these results, it can be postulated that a substantial number of patients may face challenges when considering the use of mosaic embryos. However, patients' decision-making in this context is not fully understood, including the reasoning behind their decisions, their decisional needs and the impact of having mosaic embryos.

To the best of our knowledge, this study is the first to explore these questions. It is expected that this study will allow a better understanding of patients' reasoning behind their decisions regarding transfer mosaic embryos, their decisional support needs and the impact mosaic embryos may have on emotions, behaviours and attitudes towards treatments. Findings from this study may lead to healthcare providers being better informed and thus being in a better position to manage patients with mosaic embryos.

## Materials and methods

### Participants and recruitment procedure

Eligibility criteria included having mosaic embryo/s identified as a result of PGT-A, PGT-M or PGT-SR. Both those who had already made a decision about their mosaic embryo and those who were still considering what to do with their mosaic embryos were included. Ineligibility criteria included having a cognitive impairment, inability to communicate in English, being under 18 years old or being unable to provide informed consent.

Patients of IVFAustralia with mosaic embryo/s were invited into the study by the service's clinical geneticist via emails. The invitation email provided a link for participants to access the online participant information sheet, a consent form and a response sheet. Prior to the interviews, a short online demographic survey was also sent to consenting participants.

### Data collection

Given that a qualitative approach provides an opportunity to gain a deep understanding of the topic of interest based on participants' experience, semi-structured in-depth interviews were conducted to collect qualitative data (Elliott *et al.*, 1999).

All interviews were conducted by L.C. via telephone, and each interview lasted about 30 min. L.C. has prior experience in qualitative research. In addition, L.C. was trained in qualitative interviewing for this study before the phone interviews and practiced qualitative interviewing with two senior researchers (B.M. and R.K.), who are highly experienced in qualitative research. L.C. has a professional background as a psychologist. She did not have any preconceived ideas about the topic at the time of interviewing, making bias unlikely. Nonetheless, to guide against potential bias, any differences of opinions between L.C., B.M. and R.K. were discussed with the research team until a consensus was reached. An interview guide was used to structure the interviews. It consisted of key questions followed by prompts with a focus on eliciting participants' reasoning behind their decision-making and decisional needs as well as the impact of having mosaic embryo/s (Supplementary Table S1). All interviews were digitally recorded after obtaining the participant's consent. Recordings were transcribed verbatim by a professional transcription company.

### Data analysis

A rigorous qualitative analysis was performed guided by the inductive thematic analysis framework outlined by Braun and Clarke (Braun and Clarke, 2006). Six phases of thematic analysis guided the analytic process to generate themes and subthemes emerging from transcripts (Braun and Clarke, 2006). Specifically, the transcripts were read repeatedly followed by collating data with similar topics to generate initial codes. These codes were then collated into potential themes and subthemes. This process was iterative and involved back and forth coding, merging, separating and refining of codes and themes. The qualitative software NVivo 12 was used to manage and organize codes, themes and subthemes throughout the data analysis. All participants were de-identified, with each participant's personally identifiable information being replaced by a study number. Data collection and data analysis were concurrent, and new codes arising from transcripts were

explored further in subsequent interviews. L.C. coded all transcripts, and three transcripts were independently coded by a senior researcher (R.K.). Codes and themes were discussed in a meeting, and another senior researcher (B.M.) joined the discussion. Then, L.C. refined the codes and incorporated complementary codes into the initial version of the codes.

Several research strategies were also employed to meet the criteria of rigour described by Tuckett (2005): credibility, transferability, dependability and confirmability. A research field journal was kept by L.C. and served as an audit trail to optimize credibility, dependability and confirmability as outlined by Tuckett (2005). This field journal recorded any reflections during or immediately after interviews, agenda points for discussion meetings with other researchers and decisions made throughout the study. L.C. continually reflected on her role in this research project. She attempted to keep her knowledge and any preconceived notions separate from the analysis. The reflexivity was also enhanced through discussions in team meetings. Credibility and dependability were further ensured by researcher triangulation through the involvement of three researchers as mentioned above.

### Ethics approval

The study received ethics approval from the Human Research Ethics Committee of IVFAustralia (Project Number: 169).

## Results

### Demographic and medical characteristics

Twenty women with mosaic embryo/s participated in interviews, after which data saturation was achieved as no new themes were emerging. There were 11 participants who used PGT-A and 9 who used PGT-SR. Whether PGT-A or PGT-SR was performed did not influence the decision to transfer a mosaic embryo. Among women who used PGT-A, four women decided to transfer a mosaic embryo and the other seven decided not to transfer. Similarly, among women who used PGT-SR, six women transferred a mosaic embryo and three did not transfer.

The majority of participants (17 participants) were in a situation where they considered the transfer of mosaic embryos. Specifically, 10 women transferred mosaic embryos and 7 participants decided not to transfer mosaic embryos after consideration. The other three participants had not needed to transfer the mosaic embryos, because two had euploid embryos for transfer, and the third was pregnant at the time of interview. This pregnant participant froze the mosaic embryo and reported that she would consider transferring it, if the current pregnancy was unsuccessful. Table 1 summarizes participants' characteristics. The mean age of participants was 40 years. Most participants were native English speakers, married and employed full-time. Half of the participants did not have children when they found out about their mosaic embryo/s.

### Theme 1: reasoning underpinning decision-making

Table 2 shows the subthemes and exemplary quotations relating to this theme.

**Table 1 Participant characteristics.**

Participant description	Mean (range)
Age in years	40 (31–45)
	<b>n (%)</b>
Sex: female	20 (100%)
Highest level of education	
TAFE or college certificate/diploma	5 (25%)
Bachelor's degree	8 (40%)
Postgraduate degree/diploma	7 (35%)
Employment status	
Full-time employed	9 (45%)
Part-time employed	6 (30%)
Self-employed	2 (10%)
Homemaker	1 (5%)
Currently on maternity leave	2 (10%)
Marital status	
Single	2 (10%)
Married	12 (60%)
De facto	6 (30%)
Language spoken at home	
English	19 (95%)
Other: Hindi	1 (5%)
Religious background	
Anglican	2 (10%)
Catholic	3 (15%)
Hinduism	1 (5%)
Orthodox	1 (5%)
None	13 (65%)
Number of oocyte collection cycles	
Cannot remember the number	13 (65%)
Two cycles	3 (15%)
Three cycles	1 (5%)
Five cycles	1 (5%)
Eight cycles	1 (5%)
Twenty cycles	1 (5%)
Number of children when patients found out about their mosaic embryo/s	
No child	10 (50%)
One child	7 (35%)
Two children	3 (15%)
When did the participant learned about their mosaic embryo/s	
Same year as interview conducted	1 (5%)
One year ago	8 (40%)
Two years ago	4 (20%)
Three years ago	1 (5%)
Four years ago	2 (10%)
Five years ago	1 (5%)
Six and seven years ago	1 (5%)
Not responded	2 (10%)

TAFE, Technical and Further Education.

### *No support for transfer of mosaic embryos*

Most participants who made this decision reported that they were concerned about the potential risks, including the low chance of implantation, risks of miscarriage and consideration of termination because of anomaly and the chance of delivering a baby with a malformation. Prioritization of chromosomally normal embryos was another common reason reported. Specifically, some participants indicated that they had frozen euploid embryos and that they would prefer to prioritize transfer of these embryos. Other participants, who had no remaining euploid embryos, stated that they would prefer to have further IVF cycles in the hope of obtaining euploid embryos before considering transfer of mosaic embryos. In addition, financial implications were considered. Given the uncertainties relating to mosaic embryos, women expressed concerns that investing time, money and emotion in such embryos might not be worthwhile. One participant felt that it would be a waste of time to spend money 'to do a transfer when having such a small chance of it'. Another participant thought that waiting for 16 weeks to obtain certainty from amniocentesis was too long, and the waiting period would be 'emotionally difficult'. Two women doubted the accuracy of PGT-A. Also, some participants reported that their fertility specialists had counselled them against transferring mosaic embryos and participants followed this medical advice.

### *Support for transfer of mosaic embryos*

Most participants who decided to use mosaic embryos had not succeeded with other options such as undergoing more cycles or transferring normal embryos. As a result, using mosaic embryos was their last available option. Others perceived transferring mosaic embryos as having a range of benefits, including the chance of success, statistics being available and discussed with their clinician, the opportunity to achieve one's desired family size, having a genetic link with a future child compared with adopting a child, and saving money. Other participants reported that they had established a reasonable understanding of the impact of mosaic embryos. Some women had searched for information themselves and then discussed it with the genetics team. Subsequently, participants weighed benefits and risks. Many of them expressed a willingness to take the risks associated with transferring a mosaic embryo. Also, advanced maternal age was a frequently mentioned reason for opting for transfer of a mosaic embryo. Participants felt that advanced age would lower the possibility of success of additional cycles. Hence, they preferred transferring mosaic embryos so that the opportunity was not wasted. In addition, participants were encouraged by the successful experience of other women in similar situations. Some participants stated that they would feel guilty due to their religious beliefs if they wasted mosaic embryos. Another reason for wishing to transfer a mosaic embryo was confidence in technology and science. For example, one participant said she transferred a mosaic embryo, because she believed in science and thought the technology would be available to check the health status of the foetus after implantation.

## **Theme 2: decisional needs when considering mosaic embryos**

Table III shows subthemes and exemplary quotations regarding this theme.

**Table II Reasoning underpinning decision-making.**

Subthemes	Exemplary quotations
<b>No support for transfer of mosaic embryos</b>	
Potential risks of mosaic embryos	<p><i>Low chance of implantation</i> Only that they're just a little bit less likely to implant. (Participant 17)</p> <p><i>Risks of miscarriage</i> I've had a lot of miscarriages, so a lot of chromosome issues. . . So yes, I'd be concerned they all end in a miscarriage. (Participant 19)</p> <p><i>Consideration of termination because of anomaly</i> Have to terminate is too much to bear, I felt the risk associated with that stops me. (Participant 9)</p> <p><i>Delivering a baby with malformation</i> My main concern would be that the baby would be born successfully, but that she might have some problems. (Participant 15)</p>
Priority of chromosomally normal embryos	<p><i>Still have euploid embryos</i> We were fortunate that we had good solid embryos what we thought, and we didn't even need to consider mosaic at that point. (Participant 9)</p> <p><i>Prefer to have further IVF cycles</i> Maybe we'd go through multiple rounds and maybe the mosaic would be our best option, in which case we would consider transferring it. But we thought it was our first cycle and we wanted to see how we went in future cycles. (Participant 4)</p>
Lack of guarantee	<p><i>Time investment</i> We would need to wait until week 16 of the pregnancy in order to do the relevant amniocentesis. Yes, we would need to do that test to obtain certainty. . . four months, it's almost halfway into the pregnancy. (Participant 18)</p> <p><i>Financial implications</i> It is pointless using the mosaic one, you know, spending that much money to do a transfer when having such a small chance of it. (Participant 14)</p> <p><i>Emotional investment</i> The time and the stress of having to wait 16 weeks is obviously emotionally quite difficult. (Participant 18)</p>
Doubt about testing accuracy	<p><i>Misdiagnosis in previous experience</i> I'm an example of the testing going wrong, okay. So, we transferred a perfectly tested embryo and it turned out that it wasn't. (Participant 9)</p>
Influence of fertility specialists	<p><i>Suggestions from fertility specialists</i> My fertility specialist thought it was a lost cause, that there's no point transferring the mosaic. . . So, he just dismissed any conversation about it. (Participant 17)</p>
<b>Support for transfer of mosaic embryos</b>	
Run out of options	<p><i>Failed in trying other options</i> We're sort of at the point after so many failures that we're running out of—well, I feel like we're running out of options. So that's what's contributed to us actually using it. (Participant 1)</p>
Perceived benefits	<p><i>Possibility of success</i> There was a chance that it would be successful, even if it was a smaller chance. Then actually it could have been successful. There's always a chance. (Participant 12)</p> <p><i>Statistics were available and being discussed</i> You would always depend on them on the mosaic and what type of mosaic is involved. . . We felt pretty safe from the get-go once we got explained actually the statistics and the information that we had so far about this type of mosaic. (Participant 10)</p> <p><i>Chance to achieve desired family size</i> We have to try it all and I really, really wanted to have another kid and for my son and like I said, we're here by ourselves. We it's just the three of us so I didn't want my son to grow up alone so I thought, I have to try it. I have to try it for him. (Participant 10)</p> <p><i>Having genetic link with future child</i> Knowing that there's an embryo somewhere that could potentially give me another baby and that is my own genetic material, I would take that chance in a drop of a hat. (Participant 7)</p> <p><i>Saving money</i> Well, if we didn't, we would not have any more embryos to transfer like we don't have any more. I can't make embryos. We'd have to try and get more embryos donate like eggs donated. So, which is costly. It's expensive. (Participant 17)</p>

(continued)

Table II Continued

Subthemes	Exemplary quotations
Established reasonable understanding	<p><i>Searching information by themselves</i></p> <p>I started reading like scientific papers and also just, you know trying to understand what the problem was. . . So, like I got to the point where I had a reasonable, I think, reasonable understanding about what the risk was and for our particular embryos. (Participant 3)</p> <p><i>Discussion with genetics team</i></p> <p>Speaking to the genetic counsellor who explained it to me, and it gave me some statistics and gave me, you know, explained she has to say that that is what completely changed my mind. (Participant 14)</p>
Risk tolerance	It would either result in a miscarriage or completely healthy child. And I was willing to accept those risks. (Participant 5)
Age	We may transfer it because as a consequence of my age, I'm 43 at the moment and next month I turn 44. I think that my partner and I have decided that we won't do any more IVF cycles. (Participant 18)
Successful experience	I'd seen a lot of people's successes and so I was just feeling like I should, we should definitely give this a try. (Participant 13)
Religious reason	I guess it also was the reason that helped me decide yes to use it because I feel like it was, I felt less guilty because I wasn't wasting – I didn't waste any of my embryos, I tried with every single one of them. (Participant 14)
Confidence in technology and science	I was quite confident. I think that like the century that we live in like there's so much technology, the science behind all of this IVF, I was very confident in transferring it because I knew there's stuff out there that can be test. . . you know can be test what's happened if I'm going to have a viable pregnancy or not. (Participant 2)

### Information about mosaic embryos

When considering transferring mosaic embryos, most participants said they wanted to know potential risks related to their particular mosaic embryos and how to manage such risks. Many participants expressed a need for research studies reporting results on mosaic embryos, including short- and long-term outcomes of babies born from mosaic embryos. Women also wished to know about the success rates, including the possibility of implantation and the chances of a live birth. In addition, some participants felt unfamiliar with the concept of mosaic embryos before speaking to clinicians and thought general information on mosaic embryos would be helpful, including an explanation of what constitutes a mosaic embryo. In contrast to general information, some participants thought detailed information such as information on mosaicism classification would be helpful to patients' decision-making.

### Technical information about testing

Several participants mentioned that they did not know how the test was performed on embryos and that they would like to know more about it. Also, amniocentesis was mentioned by some participants. One said that it had been recommended to her during genetic counselling, but that no details had been provided to her and she needed to read about it online before she felt comfortable about it. In addition, one participant expressed concern about the possibility that amniocentesis may not be accurate and could result in a misdiagnosis; she expressed a wish for more information about this test.

### Unmet needs in considering mosaic embryos

Unmet needs were identified in relation to healthcare services. Several participants felt that they had received ambiguous opinions from the genetics team and would prefer a definite answer and being told what to do. Several participants preferred receiving statistical and scientific details about mosaic embryos during genetic counselling rather than searching for this information by themselves. Given that a mosaic embryo may result in an affected foetus, participants said that termination of pregnancy was an option they would consider. Participant 12 said that she had not received sufficient information on what was involved in termination. Also, Participant 11 said she could not understand medical terminology in reading materials provided by health professionals and thought using plain language would be helpful. In relation to other unmet needs mentioned, Participant 10 said she wanted follow-ups from the genetics team checking her pregnancy after a transfer of a mosaic embryo, including being informed about the next steps. Similarly, Participant 14 said it would be beneficial if a referral to a genetics team was mandatory after the diagnosis of a mosaic embryo. In addition, Participant 4 expressed a preference for empathetic counselling rather than solely information-giving. Other participants mentioned their preference for receiving advice from specialists who had experience in managing patients with mosaic embryos.

Unmet needs were also identified in relation to social support. Participant 1 reported that she felt isolated, since she did not know anyone who had been through a similar situation. Therefore, she thought that social support from peers, such as attending support groups, would be helpful in addition to other support she had

received. In fact, a Facebook group named 'My Perfect Mosaic Embryo' was mentioned by five participants. However, most participants were unaware of any support group, which indicated the lack of information on available support groups.

### Theme 3: impact of having mosaic embryos

Table IV shows subthemes and exemplary quotations relating to this theme.

#### *Emotional responses*

Several emotional and behavioural responses were expressed in relation to having a mosaic embryo identified. Many participants described feeling stressed, disappointed and frightened when they learned that a mosaic embryo/s had been identified. In contrast, several participants described a sense of hope because transferring the mosaic embryo 'might be successful'. After the transfer of a mosaic embryo, participants' emotional responses varied depending on the results of the transfer. Specifically, participants who were waiting for results of the transfer felt anxious, stressed and worried. Participants with a successful pregnancy described that they felt positive, excited and fortunate, while those whose transfer was not successful felt disappointed and upset. However, two participants also reported some relief after an unsuccessful transfer, since this outcome pre-empted the uncertainties, which would have ensued had the transfer been successful.

#### *Discussion regarding prenatal testing*

After the finding of a mosaic embryo, all participants were informed that amniocentesis was recommended if they were to get pregnant. Many participants said they would follow such advice, because they wanted to have certainty as to whether the foetus was chromosomally abnormal. They also said that, although there was a risk of miscarriage associated with amniocentesis, it was low and that it was important to get tested. In contrast, several participants would not have an amniocentesis because of the risk of miscarriage. Others decided against an amniocentesis, because they would not terminate the pregnancy even if there was something wrong with the foetus. Some participants were unsure about whether to have an amniocentesis and believed that their decision would depend on the specific context.

#### *Considering termination of pregnancy*

In a scenario where the foetus resulting from the mosaic embryo was malformed or affected with a genetic condition, most participants said they would choose a termination of pregnancy. One reason was that they did not want to bring a child with a low quality of life into the world. Also, some would terminate, because a child with a chronic medical condition would be a burden for their family. Another reason in favour of termination was the foetus' perceived low chances of survival. Unlike these explicit attitudes towards termination, several participants said that whether to terminate would depend on the specific situation, such as type of mosaicism or anomaly identified. One participant expressed the view she would not terminate because of her religious beliefs, as she wanted to 'keep some things in God's hands'.

#### *Facing more IVF*

Many participants underwent another IVF cycle, because they preferred chromosomally normal embryos, and all of these had been

used up. One participant decided to have another IVF cycle, because she achieved a child successfully from her previous cycle.

Participants who did not pursue new IVF cycles expressed various reasons for this choice. One was advanced age, given that oocyte quality deteriorates with age. Cost was another reason for not trying more IVF cycles. Also, several participants mentioned that they had accepted their current situation. Some said they were not mentally prepared to have additional cycles. Other reasons were side effects of drugs, prevention of physical and emotional stress and the choice to transfer a mosaic embryo.

#### *Changed attitudes towards PGT*

Participants interviewed had PGT either for testing a known genetic condition or for aneuploidy screening to increase the likelihood of implantation. However, the diagnosis of a mosaic embryo went against the reasons for having PGT, as the ambiguous result meant they were less confident in the likelihood of having a healthy baby. Given the presence of mosaicism, nearly half of participants changed their attitude towards PGT and decided not to have PGT in the future.

### Theme 4: role of health professionals

Participants observed that their clinicians had different attitudes towards mosaic embryos, including encouragement or discouragement of transferring or being neutral. Participants reported that they were influenced by health professionals in their decision-making. Many participants indicated they had insufficient knowledge about mosaic embryos and, therefore, relied on their treating clinicians to decide whether or not to transfer a mosaic embryo. One participant said, given her clinician's positive attitude and encouragement, she decided to transfer her mosaic embryo. Another participant said that, because the clinician appeared to have a neutral attitude, she tried to identify clues during her discussions with the clinician/s to guide her as to whether to transfer a mosaic embryo.

#### *Other themes*

Secondary themes are listed in [Supplementary Table SI](#) and include the most challenging parts of decision-making about mosaic embryos, the people who contributed to decision-making and other information sources and impacts of mosaic embryos on patients' lives. Notably, women acknowledged the role of their partners in decision-making.

## Discussion

Previous articles describe the factors influencing decision-making about transferring mosaic embryos. Specifically, one opinion article mentioned the key factors when considering the transfer of mosaic embryos, such as the degree of mosaicism, the affected chromosome and a woman's medical history (Simon, 2017). One study investigated what patients did with their mosaic embryos and suggested the factors that influenced decision-making about transferring mosaic embryos, such as advanced parental age, physical and psychological burdens and financial reasons (Besser et al., 2019). These factors were among those identified by our participants. In addition, our study identified religion as a factor in supporting the transfer of mosaic embryos and as influencing attitudes to termination. The results indicated the influence



**Table III** Decisional needs when considering mosaic embryos.

Subthemes	Exemplary quotations
Information about mosaic embryos	<p><i>Potential risks and risk management</i></p> <p>For me understanding the specific mosaic in which how each chromosome was affected was the important information, and so like therefore what the possible outcomes could be. (Participant 3)</p> <p>Obviously, the risks and how to have then to manage and monitor those risks. (Participant 3)</p> <p><i>Research studies reporting results of mosaic embryos</i></p> <p>I don't think there's that much literature about it. . . I probably want information that doesn't exist yet, like I want results from like a pretty well-conducted trial with as many people who've transferred mosaics. . . Sort of long-term results of the baby, I guess implantation rates and then what the results were and then what the final result was with the baby. (Participant 4)</p> <p><i>Success rate</i></p> <p>What are the chances that yes, it's going to be an embryo, it's going to be a mosaic embryo, but what are the chances that it's going to be a positive um, pregnancy that I'm going to have at the end of the day, a child after nine months? (Participant 2)</p> <p><i>General information on mosaic embryos</i></p> <p>I obviously want to know what a mosaic is. . . you know, no one talks about it. I had no idea what it was, for starters. (Participant 2)</p> <p><i>Detailed information on mosaic embryos</i></p> <p>The report that came back from the laboratory said that that our particular mosaicism was 'low grade' we didn't really understand what that meant. . . It would be helpful to know if the mosaicism was detected in all of the cells that were tested or just one of them or two of them or so, I think that that level of information. (Participant 18)</p>
Technical information about testing	<p><i>Information on preimplantation genetic testing process</i></p> <p>I guess, to know like how they do the testing, the fact that they only test from the trophectoderm, which then becomes the placenta. (Participant 17)</p> <p><i>Details of amniocentesis process</i></p> <p>I looked up what was involved in an amnio and I felt a little bit more comfortable with that than previously. That was actually something actually that's probably a throwback comment, but I feel like giving a little bit more information on that during that genetic counselling session would be helpful. (Participant 4)</p> <p><i>How reliable and comprehensive are the amniocentesis results</i></p> <p>I felt that there wasn't much information on was I was actually concerned about, you know, if everything was shown to be okay so the embryo implanted amniocentesis didn't flag a really significant problem, the baby reached full-term, what was the likelihood of there being a genetic issue. And there was actually no data really on that case of it. (Participant 12)</p>
Unmet needs in considering mosaic embryos	<p><i>Health care service: Lack of certain information</i></p> <p>I wish they could stand around and said to me, 'This is what I could do'. But obviously, from a legal point of view that they can't do that sort of stuff from a professional point of view, but I wish they could have told me if it was them, they wouldn't try or, you know, there is not pretty much next to no chance. (Participant 11)</p> <p><i>Health care service: Lack of statistical and scientific details</i></p> <p>I like the statistics and the scientific details. So, yeah, but for me that yeah that would I guess that would have been helpful if they provided that rather than me kind of go elsewhere to find it. (Participant 3)</p> <p><i>Health care service: Lack of details on termination</i></p> <p>I hadn't really got during the conversation with the counsellor and genetic sciences was around, actually what is involved with a termination following amniocentesis at that point in the pregnancy. (Participant 12)</p> <p><i>Health care service: Prefer plain language</i></p> <p>They sent me this study they gave me, you know, I didn't understand it at all. I mean, a lot of it was scientific. Just medical terminology that I didn't understand. (Participant 11)</p> <p><i>Health care service: Prefer follow-ups from the genetics team</i></p> <p>I think it would have been good to, to have more of a follow-up with you guys, with the geneticists, or the embryologists. I don't know who is the one, but maybe, you know. I don't know, I like to be more on top of what is going on. (Participant 10)</p> <p><i>Health care services: Prefer to connect with genetics team mandatorily once learn about mosaic embryos</i></p> <p>I think at that point it could be very beneficial that, you know, the IVF clinic or the specialist makes it mandatory that you do speak, if you do have a mosaic that you do speak to the genetic- I don't know if it's a counsellor or a scientist, but you do speak to them. (Participant 14)</p> <p><i>Health care services: Lack of empathy</i></p> <p>But the way that I was sort of delivered. I just felt like there was something lacking. . . put herself in someone else's shoes. (Participant 4)</p>

(continued)

**Table III Continued**

Subthemes	Exemplary quotations
	<p><i>Health care service: Prefer to receive advice from clinicians with experience in mosaicism</i></p> <p>I guess, I mean, if there were information or other professionals who were, for example, more knowledgeable or had had more experience with mosaic embryos like specialists in the field. . . that would be useful. (Participant 16)</p>
	<p><i>Social support: Lack of information on support groups</i></p> <p>Wider context that is obviously a big influence by having other people to talk to about that. That certainly would be good, because certainly from my perspective, um, I don't have any friends who have been through nor are going through this sort of situation so it's, it does feel isolating. . . it would be good to have more support from somebody who's been through this or, you know, a broader support network. (Participant 1)</p> <p>So, I joined a Facebook group called, 'My Perfect Mosaic Embryo' or something. And there was a lot of information shared on there, like a lot of studies that had been done, like links to studies, um, and so I just followed up through that way, like watched a lot of videos, read a lot of studies. (Participant 17)</p>

of religion on patients' reproductive decision. Many studies have investigated the influence of religion and revealed the associations between religious affiliation and attitudes to termination and IVF (Schenker, 2005; Evans and Hudson, 2007; Larjani and Zahedi, 2007; Sigillo et al., 2012; Gebhart et al., 2016). Although only 35% of our participants reported religious affiliations, the influence of religion should be noted in relation to pregnancy and termination.

Regarding decisional needs, this study identified patients' needs for information about mosaic embryos, amniocentesis and termination. As expected, most had little prior knowledge about mosaic embryos and relevant procedures. This highlights the importance of educating and equipping patients with sufficient knowledge to achieve informed decision-making. To the best of our knowledge, no decision aids are available specifically for patients who are considering the transfer of mosaic embryos. Such decision aids could provide essential information to assist patients in making decisions about whether to transfer mosaic embryos. Clinicians should also consider patients' decisional needs as identified in this study when managing patients with mosaic embryos.

The analysis of patients' unmet needs revealed areas for improvements in the future. For example, some patients reported that clinicians delivered general information on potential risks, while statistical and scientific details of using mosaic embryos were insufficient. Such detailed information would help patients estimate the potential risks of transferring mosaic embryos. Given that not every patient favours such information, patient-centred care is recommended where provision of detailed scientific data should be based on patients' need. Also, offering information on social support like support groups may provide patients with a platform to share their feelings and thoughts with individuals in a similar situation, which may subsequently minimize their sense of isolation. Some unmet needs are difficult to meet given the current limitations of the technique. However, participants' concerns about ambiguous results relating to mosaic embryos and the ensuing uncertainties may be resolved in the future with technological advances.

Regarding the impact of having a mosaic embryo identified, negative emotions were identified at different stages: after diagnosis, while waiting for results after transfer, and when a transfer had failed. This result

is consistent with a previous study, which describes patients' anxiety levels along the PGT trajectory (Karatas et al., 2011). Therefore, it is important to provide support for women undergoing PGT, especially during the above three stages. In addition, diagnosis of mosaic embryos facilitated discussions on amniocentesis, termination and having more IVF cycles. Regarding these discussions, results showed individual differences in decisions made by women. Thus, clinicians should inform patients about all available options and the relevant pros and cons to meet various decisional needs. Also, given the identified influence of health professionals, it is recommended that health professionals should be aware of the influence their attitude can have on patients' decision-making and should present information accordingly. Future studies should also explore participants' understanding of potential disability because of using mosaic embryos and whether specific disabilities were less acceptable than others.

Although most participants (16 out of 20) learned about their mosaic embryo/s within 5 years prior to being interviewed, there is a chance of recall bias, and it is possible that women may have vague recall, or even no recall, of relevant details related to their decision-making. Other limitations of this study should be mentioned. In this study, participants were recruited through one clinic, which may limit the transferability of results, since participants' experience may be influenced by the particular practices of the clinic. Though data saturation was reached in this study, more or different themes might have emerged if participants had been recruited through more diverse sources. Another limitation of our study was that only females were interviewed. Although the decisions would have been made jointly by both partners, we acknowledge that partners' perspectives were not recorded, and this may limit the applicability of findings to couples' decision-making. Also, patients' experiences in relation to financial aspects of PGT may not be relevant to other jurisdictions due to different health care policies.

## Conclusions

This study is the first to explore patients' decision-making regarding the transfer of mosaic embryos identified by PGT-A. Results revealed

**Table IV** Impact of mosaic embryos and the role of health professionals.

Subthemes	Exemplary quotations
Emotional responses	<p><i>After an embryo was found to be mosaic</i></p> <p>I would say I would be lying if I said that it wasn't stressful because it's solidified in your mind that you went through all this testing and look, it came back that there was something wrong. (Participant 14)</p> <p>It gave us a bit of hope that, you know, we might be successful. (Participant 18)</p> <p><i>Waiting for transfer results</i></p> <p>If it does turn into a positive pregnancy, we're just going to be that stressed and worried. (Participant 1)</p> <p><i>Succeeded after transfer</i></p> <p>I feel very fortunate because I have had such a good outcome that at first, I was ready to dismiss. (Participant 15)</p> <p><i>Failed after transfer</i></p> <p>Just the disappointment that goes along with the... another failed transfer. (Participant 1)</p> <p>Obviously, we're disappointed that it failed but there was a little bit of relief as well that you know we didn't have to take the risk of transferring and not knowing whether it was going to be a healthy baby or not. (Participant 6)</p>
Discussions regarding prenatal testing	<p><i>Will take amniocentesis: To get certainty</i></p> <p>Because we understand that it's only a two per cent chance of miscarriage, so we understand that it's a very low chance in the scheme of things. And I think that we would as a couple for us personally, I, I think that we would be willing to take that chance because it's so low. Um, in order to have the certainty as to whether or not the chromosomal abnormality was actually confined to the placenta or not. (Participant 18)</p> <p><i>Will not take amniocentesis: To avoid miscarriage</i></p> <p>I guess because we don't want to do anything, not even if it's as small as the 0.5 per cent of miscarriage chances. We don't want to take that chance now. (Participant 10)</p> <p><i>Will not take amniocentesis: Will not terminate</i></p> <p>I'm just a little bit anti-testing, you know, like, I just don't know I just kind of think I'm not the kind of person that would like to get rid of the baby if there was something wrong with it. (Participant 13)</p> <p><i>Not sure: Will depend on the real context</i></p> <p>That's a question I don't 100 per cent know the answer to yet to be honest, because um, as I said, like I'd it's not that I'm not necessarily against it, I know there's risk involved with it, and I think the risk is quite small. But I think- I would possibly consider it in the sense that if we had a mosaic embryo and for example, it was there was a chance that the baby could have some kind of disability. (Participant 16)</p>
Considering termination of pregnancy	<p><i>Terminate: Future child's low quality of life</i></p> <p>But if we if I had an embryo, that was. You know, going to have like trisomy 18 or trisomy 13 or severe intellectual disabilities or something like that, then that's like, and no quality of life, then I would have a termination. (Participant 17)</p> <p><i>Terminate: Burden for family</i></p> <p>I think that's something that we will we felt really strong about like we couldn't cope with having a kid with disabilities and being alone here in Australia with all our families overseas. (Participant 10)</p> <p><i>Terminate: The foetus is unlikely to survive</i></p> <p>The foetus wouldn't survive the pregnancy or even if even if I did give birth, that it would pass away not long after, not long after birth. (Participant 18)</p> <p><i>Not sure: Will depend on the actual situation</i></p> <p>Because of the nature of the, you know, the deletion itself, if it was something else. It would depend on what type of mosaic embryo was, what the defect was. (Participant 20)</p> <p><i>Not terminate: Religious belief</i></p> <p>I would like to keep some things in God's hands and not everything in the doctors and the medical professionals, in their hands. So, at this stage, I would definitely be saying, 'No, I wouldn't terminate'. (Participant 14)</p>
Facing more IVF	<p><i>To have another IVF cycle</i></p> <p>So, when we ran out of euploid embryos, we tried to do some more egg collection cycles. (Participant 3)</p> <p>Our previous IVF cycle had been really successful... So, we thought it was worth trying another cycle because it you know, previously it had been quite a lot more successful. (Participant 12)</p> <p><i>Not to have another IVF cycle</i></p> <p><i>Age</i></p> <p>If I could have, I would have, but I'm too old and I don't have any more money to do it. (Participant 15)</p> <p><i>Accept the current situation</i></p> <p>Yeah, we moved on. And I think, you know, we're in a very lucky situation that we already have two children. I'm sure this you know, most patients probably trying for their first and would keep trying where we felt like this is this is enough, now we've tried, and we have to move forward. Hmm. (Participant 8)</p>

(continued)

**Table IV Continued**

Subthemes	Exemplary quotations
	<p><i>Not mentally prepared</i></p> <p>I wasn't at that stage I wasn't mentally prepared to do a second cycle and then I knew this was my only option. (Participant 14)</p> <p><i>Side effects</i></p> <p>Well, it's hard to say because all the drugs have side effects anyway, so whether I feel pregnant or not, I've given up trying. (Participant 1)</p> <p><i>To avoid potential stress</i></p> <p>So, we kept on going for cycles after cycles and we could have prevented doing that, we could have prevented the hardship of the you know, the physical stress and the emotional stress. (Participant 10)</p> <p><i>Decided to transfer the mosaic embryo</i></p> <p>I wouldn't try to produce any more euploid embryos. And because I know that that mosaic is actually quite a good candidate to transfer. (Participant 3)</p>
Changed attitudes towards PGT	<p>But we just felt like we try in future, and we also made the decision that we wouldn't do any more genetic testing because we felt that it had been so unhelpful knowing that it was a mosaic. Like, it didn't really it didn't help us understand what to do. (Participant 4)</p> <p>I didn't want to get the testing done because I don't know like how much they could, how much information they'd be able to provide me and what that would mean and how sure they'd be about it and how much of an impact you know it would be. And like, I just don't know that's why I kind of probably wouldn't have done the testing because I don't want to be put in that situation. (Participant 13)</p>
The role of health professionals	<p><i>Encouragement</i></p> <p>Um, encouraged by our doctor. I would say in that they were trying to explain you know that there had been relatively positive outcomes with mosaics, so um, so yes. So, he was feeling not too bad at the time. (Participant 12)</p> <p><i>Discouragement</i></p> <p>My fertility specialist thought it was a lost case, that there's no point transferring the mosaic because he was unsure if it would either result in pregnancy or result in a malformed baby. . . So, he just dismissed any conversation about it. (Participant 7)</p> <p><i>Non-directive</i></p> <p>She doesn't really have her opinion. . . it's not something she encouraged or discouraged. (Participant 9)</p>

PGT, preimplantation genetic testing.

the factors influencing decision-making about whether to transfer and patients' decisional needs, which may inform health professionals regarding important aspects in managing patients with mosaic embryos. In addition, given the individual differences in making decisions and the weight placed by patients on the views of their doctors, clinicians should be aware of the influence of their attitudes on patients' decision-making when discussing the pros and cons of potential options. The results of this study also showed patients' educational needs for knowledge about mosaic embryos. Educational resources including decisional aids regarding mosaic embryos should be developed and offered in plain language.

Given the patients' emotions such as feeling stressed and anxious throughout the testing and transfer procedure, psychological support should be offered to those in need. Also, having informing for patients about available psychosocial support as well as potential support groups may alleviate their negative emotions such as feelings of isolation.

## Supplementary data

Supplementary data are available at *Human Reproduction Open* online.

## Data availability

The data underlying this article will be shared on reasonable request to the corresponding author.

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## Authors' roles

L.C. designed the study, constructed the interview guide, interviewed the participants, analysed the data and wrote the article. B.M. designed the study, constructed the interview guide, supervised the interpretation of data and reviewed and edited the article. D.K. designed the study, constructed the interview guide, recruited participants and reviewed and edited the article. E.K. designed the study and reviewed and edited the article. K.B.-S. designed the study, constructed the interview guide and reviewed and edited the article. R.K. analysed and supervised the interpretation of data and reviewed and edited the article.

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## Conflict of interest

The authors declare no conflicts of interest.

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