INTRODUCTION

According to Beauchamp and Childress, an autonomous action requires intention, understanding and the absence of controlling influences, meaning it must be made ‘freely in accordance with a self-chosen plan’. Even though Beauchamp and Childress acknowledge that social influences are unavoidable and permissible to a

certain extent, feminist philosophers have criticized the underlying conception of autonomy as too individualistic, disregarding the influence of social relations in human actions and decisions.3

Their alternative conception of relational autonomy embeds social relationships and their influence on decisions and actions. Relational autonomy theories can be classified as substantive, meaning value-laden, or procedural, meaning content-neutral.4 We here use the procedural conception of relational autonomy, thus examining the decision-making process in predictive genetic testing irrespective of the decision. In John Christman's procedural conception, decisions are autonomous if individuals are cognitively and normatively competent during the decision-making process (i.e. rational and self-controlled), and their decision is self-reflective.5 Marilyn Friedman has a similar conception of procedural autonomy with a lower threshold, in the sense that she considers a decision as autonomous as soon as a person is minimally self-reflective.6 Both Christman and Friedman emphasize the influence of the social environment for this self-reflection but refrain from the claim that social relations constitute autonomy, meaning that social relations are necessary to act autonomously.7 Accordingly, Bruce Jennings observed a 'relational turn' in the conception of autonomy in biomedical ethics: 'The challenge of respecting the autonomy of persons is not to avoid relationality but to distinguish those forms of relationship and life worlds that are functional for the maintenance of communal meaning and integrity from those that are not.8

Particularly in clinical genetics, the influence of social relations has been increasingly recognized.9 Empirical inquiries have confirmed that family considerations often influence genetic testing decision-making.10 Particularly in predictive genetic testing, the decision to test not only uncovers information about the tested individual but also reveals facts about blood relatives. Therefore, relatives also receive predictive information about disease risks and thus may face the decision of whether or not they want to perform such a test. Because each person has a right to know, but at the same time also a right not to know genetic risk information,11 this leads to potential dilemmas if individuals from the same family have divergent preferences.12

Here, we examine autonomy in the context of decision-making in predictive genetic testing for hereditary breast and ovarian cancer syndrome (HBOC) or Lynch syndrome, which increases the risk of colon cancer.13 Predictive genetic testing is, per definition, performed in asymptomatic individuals.14 Those carrying a risk-increasing genetic variant can opt for more regular preventive screening, or, in the case of HBOC, for preventive, risk-reducing surgery.15

Empirical studies on genetic testing decision-making mainly come from North America16 and the UK.17 More recently, such studies have been conducted in other European countries such as Italy18 or Spain19, but not in Switzerland. The legal and clinical context in Switzerland differs from other countries. Genetic counselling before and after predictive genetic testing from a specialized medical doctor is legally required.20 Moreover, Swiss health insurance covers the costs of genetic testing and genetic counselling under certain conditions, including genetic testing for cancer predisposition in the case of relevant family history or early-onset cancer.21 Only a few genetic counsellors work in Swiss clinics,22 and it is not an officially accredited profession.

This article aims to explore aspects of social influences in predictive genetic testing decision-making in Switzerland and discusses consequences for the conception of autonomy in this context. This is the first Swiss study providing empirical evidence about the social influence of genetic testing decision-making.

2 | METHODOLOGY

This study is part of a grounded theory analysis based on an interview study with people undergoing genetic counselling for cancer predisposition (HBOC or Lynch syndrome) in a clinical setting in Switzerland. We asked participants in semi-structured face-to-face interviews about their reasons, attitudes and information strategies regarding predictive genetic testing and analysed the interviews in terms of the decision-making process and information-seeking behaviour. For data triangulation, we also interviewed two counselling physicians who helped with patient recruitment and had counselled some of the study participants regarding the topics that came up during analysis. For this study, we reanalysed the codes obtained from the analysis of the decision-making process from the theoretical perspective of relational and individualistic autonomy.

Inclusion criteria were participation in at least one genetic counselling session, healthy mental state, non-pregnancy and 18–70 years of age. We included both healthy individuals and cancer patients, as genetic testing for HBOC and Lynch syndrome also provides cancer patients with predictive information about risks of other cancer types. Both acceptors and decliners of genetic testing were eligible. Participants were recruited from several German-speaking university hospitals in Switzerland through their counselling physicians. To gain theoretical saturation we targeted recruitment to male participants and people deciding against predictive genetic testing during the study, as preliminary analyses had shown that those characteristics could add additional insights to the analysis. The first author (BZ) conducted all interviews between September 2017 and January 2019 in either German, Swiss-German dialect, or French. The quotes were translated to English by BZ and proofread by IK and DS.

Following the grounded theory approach, BZ and IK first openly coded the interviews 'line by line', and started building concepts by combining similar codes. Data analysis was an ongoing process and kept in the original language of the transcripts (mainly German, one French transcript), and we adapted the interview guide and refined the inclusion criteria during the study to reach theoretical saturation. We constantly compared codes within and between interviews and also compared different codes to identify connections between them, similarities and contradicting elements. At an advanced stage of analysis, we started to interrogate the data to maintain our openness towards rebuilding and discarding existing concepts, wrote memos in English and involved DS in the analysis for additional insights and interpretations. Throughout the analysis process, we wrote memos and exchanged our interpretations with each other. For this part of the study, we systematically examined our concepts and categories for their connection to relational and individualistic aspects of autonomous decision-making. The study was approved by the ethics committees of Northwest and Central Switzerland and Bern (ID number 2017-00316). All participating at-risk individuals signed an informed consent form before the interview.

3 | FINDINGS

We conducted 18 interviews with counsellees who have a family history of cancer or were diagnosed with cancer at a young age themselves, and two with counselling doctors. Counsellees’ age ranged from 27–70 years of age, 14 of them were female, and 12 had blood-related children. Six of them had a cancer diagnosis when genetic testing was offered. In five cases, a pathogenic genetic variant was already diagnosed in a relative. Three participants refused genetic testing. We use five case stories from our sample to illustrate different aspects of social influence in the decision-making process (Box 1).

3.1 | Responsibility towards relatives

The first aspect is the responsibility that at-risk individuals feel towards their relatives, particularly offspring. As illustrated by Mary’s and Paul’s (names are pseudonyms) case stories (Box 1), some participants even valued their children’s interests higher than their own and agreed to a genetic test even though they were not convinced about its usefulness for themselves. Even some of the childless participants emphasized the importance of predictive genetic testing for offspring, criticizing family members with children who were not interested in predictive genetic testing. Some participants tried to persuade family members to consider testing for their offspring: ‘I think it would be useful that my sisters would… sacrifice themselves for their children and get tested, to just know for certain, to protect their children, because children are the greatest good for a family, in my opinion.’ (Jack, 50+, carrier in remission).

This focus on the interest of family members sometimes led to dilemmas if family members had competing interests. For example, Mary explained how her second daughter refused to learn about the predictive genetic test result while her first daughter wanted to know her genetic risk:
And one of my other daughters said she didn’t want to know anything about it, that she didn’t want to know my test result […] And because I suspected that she didn’t want to know anything about that, there was some ambiguity, I had to inform her that I intended to do such a test, but in fact, she really doesn’t want to know all that. That is just a tricky situation. (Mary, 60+, healthy non-carrier)

3.2 Healthcare professionals’ influence

Secondly, participants mentioned the role of healthcare professionals in their decision-making process. As in the case of Paula, physicians usually recommended predictive genetic testing if it influenced cancer treatment. However, healthy participants also described how medical professionals influenced their interest in predictive genetic testing on different levels:
Because my gynaecologist had retired, I had to look for someone new at that time and I think that was an input for me somehow; it's a young practice, and it came up, also from her side; that she would like to send me there [to genetic counselling]. Ehm, not necessarily to do the [genetic] test but just so that she would know a little bit more about how to organise [cancer] prevention, even if I didn't do the test. [...] And then I thought, yes, I'll go to genetic counselling. And listen to what they say. (Jakobia, 30+, healthy, untested)

During the genetic consultation, I knew quite quickly that I wanted to be tested. [The genetic counsellor] rather tended towards me doing it, he also wrote down my family history and said he would recommend that I do it. That it was up to me, but, ehm, that I wouldn’t block myself from anything. (Fabia, 50+, healthy carrier)

One participant explained how she and her counselling doctor decided together how to proceed with genetic testing:

She [the counselling physician] is really passionate, she's really interested... And... she speaks frankly with me, openly. So I don't feel like she's hiding things from me or making decisions for me […] She was prepared, she had already thought about it herself, okay, we're looking for other [genes], which ones, she had already done all the research according to what the geneticist had given her, and what she knew. So she was ready to offer me… so that we could discuss [which genes to test further], the options we had. (Helen, 30+, non-carrier in remission)

Even though none of the participants explicitly expressed that they were treated or counselled paternalistically, healthcare professionals considerably influenced their decision-making process. Still, healthcare professionals related to the principle of nondirective counselling:

Ideally, genetic counselling should be nondirective [...] Nondirectiveness concerns the decision regarding genetic testing itself rather than recommendations for cancer prevention. The latter for me has nothing to do with nondirectiveness. Nondirectiveness mainly concerns the genetic test. To say one should absolutely do genetic testing. Because there should be no such thing, shouldn’t it, there are good individual reasons to reject a genetic test. (geneticist 1)

3.3 | Individualistic decisions and social relationships

Another aspect participants described was the tension between individualistic reasons (not) to perform genetic testing and their social relationships. Not all at-risk individuals took relational aspects into account in their decision-making process. Margaret, for example, based her decision against predictive genetic testing on individualistic reasons. However, she and another participant refusing genetic testing expressed difficulties talking about their decision to family and friends because they feared the judgment of others: 'I am reluctant to discuss my decision with others. Because they all have their history, their opinions... I'd be afraid someone would try and persuade me, influence me, and judge me...' (Michaela, 40+, healthy, untested).

Moreover, as Paul’s case illustrates, some at-risk individuals had contradictory feelings because, from an individualistic perspective, they would prefer not to know their genetic risk, but they felt an obligation towards their offspring to agree to predictive genetic testing. Paul said that once he was informed about the option of predictive genetic testing, he felt obliged to do it (Box 1).

Social influence was often implicit and did not influence all individuals to the same extent. As Eva’s case illustrates (Box 1), knowing one’s genetic risk can be a means to know in which group (‘positive’ or ‘negative’) one belongs. While Eva’s description could refer to implicit social pressure, she perceived her decision to have predictive genetic testing as autonomous. One of the geneticists confirmed that social influence was a common phenomenon that he frequently discussed in genetic counselling:

What is not actually perceived [by counselees], but what we also address in predictive testing is, um, it's actually a well-known phenomenon that if whole families get tested [...] then, of course, there are those who have it and those who don’t. And [...] there is a bit of grouping in that sense. That those who have it are more likely to stick together at family gatherings. (geneticist 1)

Several participants stated that it was important to them that their grown-up children had a genetic test. Some participants even admitted having nudged or persuaded their adult children to be tested. This was especially the case for participants who perceived predictive genetic testing as beneficial and who found it difficult to understand why someone would refuse it.

I think my daughters just got tested because I said so (laughs). I don’t think they knew what that meant at the beginning. Because it went so fast... I think I overruled them a little bit, to be honest. Well, I don’t regret it, I think it is important. (Rose, 50+, carrier in remission)

4 | DISCUSSION

We interviewed people in Switzerland undergoing genetic counselling for a predisposition to cancer (hereditary breast and ovarian
cancer syndrome or Lynch syndrome) about their decision-making process and analysed how social relationships influenced their decisions as part of a grounded theory study. Our findings indicate that both relational and individualistic reasons influence decision-making, and in both cases, individuals might face implicit or explicit social pressure. The following sections explore the theoretical and practical implications for autonomous decision-making.

4.1 | External influences allowing for autonomous decision-making

In line with previous studies, we show that family considerations can dominantly influence predictive genetic testing decision-making. Additionally, our results suggest that the wish to belong to a social group can be a decisional factor. In both cases, decisions are still autonomous when applying a relational conception of autonomy. The impact of relational aspects in genetic testing decision-making has been increasingly recognized. Still, clinical guidelines hardly reflect on this aspect, as they only superficially recommend making psychosocial aspects and communication to the family a subject of discussion. Moreover, the whole process of obtaining informed consent in a clinical setting is inherently focused on individual consent, neglecting relational aspects in the context of genetic testing decision-making. While consent is obtained from an individual for good reasons, it should be acknowledged that relational aspects in genetic testing decision-making tend to be neglected in the clinical routine of the informed consent process. Because of the apparent importance of relational aspects in genetic testing decision-making, we think this should be explicitly mentioned in clinical guidelines.

Some participants felt torn between their feeling of responsibility towards relatives and their own interests. This feeling of inner conflict illustrates the potentially contradictory elements of relational and individualistic aspects in genetic testing decision-making. Such cases are particularly delicate and need special attention to ensure autonomous decision-making, as those individuals might be particularly susceptible to undue influences from their social environment. If family members accompany at-risk individuals to genetic counselling, it might be helpful to have an additional session without family members present.

4.2 | Undue social pressure preventing autonomous decision-making

We observed that decliners of genetic testing did not want to justify their decision to others. They perceived their decision as concerning only their own life and thus made it based on individualistic reasons. A previous study showed that decliners might also anticipate negative consequences for their relatives if they proceeded with predictive genetic testing, which to them was another reason to decline. While decliners perceived that their decision needed social justification, those agreeing to genetic testing for individualistic reasons did not express such social pressure. This indicates that declining genetic testing might provoke social pressure. The implicit nature of this pressure is problematic, as it might leave some individuals with the feeling of having no alternative but to test. To enable true freedom of choice, which we think is crucial in predictive genetic testing, social acceptance of refusal of testing should be improved. As mass media coverage has a predominantly positive narrative when it comes to genetic testing for actionable diseases, reasons to decline should be made more transparent in public discourse.

Additionally, some participants expressed incomprehension about family members who refused genetic testing. These participants had a relational perception of genetic testing and perceived genetic testing...
for cancer predisposition as beneficial. This indicates a potential dispute between at-risk individuals with an individualistic view, as opposed to those with a relational conception regarding genetic testing decision-making. In some cases, this even led to coercive situations, which has also been reported and discussed in previous studies with test takers and healthcare professionals. Anita Ho argues that healthcare professionals should first carefully examine the family context and talk to the patient and family, before making a hasty judgement about family coercion. We propose that genetic counsellors should make counsellees proactively aware of the possibility that relatives might invoke their right not to know. This is especially important if people display unreflective approval of any genetic testing. Because our interviews revealed that most potentially problematic situations of social pressure arose out of unawareness, reflecting on the situation in genetic counselling might improve this issue.

4.3 | A model to illustrate social influences in predictive genetic testing decision-making

To illustrate the role of individualistic and relational reasons in predictive genetic testing decision-making, we propose an integrative model of the aspects of social influence on autonomous predictive genetic testing decision-making (Figure 1). This model reflects the empirical data in this context and is an extension of both the principle of respect for autonomy of Beauchamp and Childress and the conceptions of relational autonomy proposed by Friedman and Christman. We propose that there is a continuum between individualistic and relational aspects of decision-making and that individuals place themselves in different places on this continuum for their decision-making regarding predictive genetic testing. Where they place themselves on this continuum is context-dependent. Contextual factors can include the family situation, the character of the person, the cultural background, and other aspects. Individuals should not be unduly influenced when positioning themselves on the continuum. Furthermore, the model sees autonomy as a matter of degree, following Beauchamp and Childress and Friedman. Consequently, there is a threshold beyond which a decision is no longer autonomous. This threshold is also context-dependent: it depends on the specific decision at stake, but also on cultural settings, as in some non-Western cultures where autonomy is to a much larger extent associated with the family’s well-being.

4.4 | Generalizability and limitations

The data informing the model we are proposing are based on considerations regarding predictive genetic testing for HBOC and Lynch syndrome. Previous studies on predictive genetic testing decision-making for Huntington’s disease, where to date no meaningful medical actions are available, show that relational considerations (especially regarding offspring and other relatives) also influence decision-making to different degrees. While the influencing role of healthcare professionals might differ from our findings, we propose that our model can also generally be extended to such applications of predictive genetic testing. As the family has a distinct role in predictive genetic testing decision-making where the predisposition is heritable, applying the model proposed here to other contexts, such as predictive genetic testing in minors or unborn children or predictive testing for X-linked conditions, would need further investigation.

This study was conducted in Switzerland, where genetic testing decision-making has not been investigated before. Swiss law requires that predictive genetic testing is covered by health insurance if medically indicated; non-directive genetic counselling is mandatory before and after predictive genetic testing and medical doctors are responsible for providing such counselling. As such, the context differs from well-investigated countries, such as the USA and the UK.

We emphasize that our model focuses solely on how to enhance autonomous decision-making, focusing on social influence, but autonomy can legitimately be restricted if it comes into conflict with other ethical principles, for example, if the autonomous action of a
person hurts other persons.\textsuperscript{47} Besides, our model operates under the premise that there are true choices available for individuals regarding predictive genetic testing decision-making, excluding other factors such as funding constraints.

Finally, this study had some methodological limitations. Reports about persuasion and coercion mostly came from third-party descriptions and often did not concern the person interviewed, but his or her family members. This is because people refusing genetic testing were challenging to reach with our sampling strategy, which did not allow for snowballing due to ethical considerations. Those who participated and refused genetic testing still at least considered testing as an option, but our study lacks participants who refuse even to learn about the option of predictive genetic testing. Also, participants explained their decision-making process retrospectively, and their perception might have changed unconsciously after learning the outcome of the test. Still, our data set is rich in a variety of contexts and opinions and our results are in line with previous studies, which increases the credibility of our empirical findings. As with every secondary analysis, our primary purpose was not to collect information on relational aspects of autonomous decision-making; instead, they occurred spontaneously in our data.

4.5 | Conclusion

This study presented a qualitative analysis of Swiss genetic counsellors, analysing social influences in decision-making for cancer predisposition genetic testing. We suggested that both relational and individualistic reasons play a role in predictive genetic testing decision-making, which affects the conception of autonomous decision-making. While the principlist and relational conceptions of autonomy are competing concepts in the theoretical debate, they are two sides of the same coin when using them as lenses of analysis for predictive genetic testing decision-making. Still, we showed that those declining genetic testing based on individualistic reasons might face implicit or explicit social pressure and that some tested individuals might persuade family members to test out of a sense of duty. However, individuals should be able to freely decide how much their social environment influences their decision for or against predictive genetic testing. Genetic counsellors should raise awareness of these issues. At the same time, public debates should focus more on the pros and cons of predictive genetic testing, avoiding positive bias.

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CONFLICT OF INTEREST

The authors declare that there are no conflict of interests.

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\textsuperscript{47}Beauchamp & Childress, op. cit. note 1.