Received: 13 February 2020 | Revised: 11 August 2020 | Accepted: 3 September 2020

DOI: 10.1111/bioe.12816

ORIGINAL ARTICLE

bioethics WILEY

Autonomy and social influence in predictive genetic testing decision-making: A qualitative interview study

Bettina M. Zimmermann[1,2] | Insa Koné[1] | David Shaw[1,3] | Bernice Elger[1,4]

[1]Institute for Biomedical Ethics, University of Basel, Basel, Switzerland

[2]Institute for History and Ethics of Medicine, Technical University of Munich, Munich, Germany

[3]Care and Public Health Research Institute, Maastricht University, Maastricht, the Netherlands

[4]Center for Legal Medicine, University of Geneva, Geneva, Switzerland

Correspondence
Bettina Zimmermann, Institute for Biomedical Ethics, University of Basel, Bernoullistrasse 28, 4056 Basel, Switzerland.
Email: bettina.zimmermann@unibas.ch

Funding information
Freie Akademische Gesellschaft Basel; University of Basel; Open access funding enabled and organized by Projekt DEAL.

ABSTRACT

Beauchamp and Childress' definition of autonomous decision-making includes the conditions of intentionality, understanding, and non-control. In genetics, however, a relational conception of autonomy has been increasingly recognized. This article aims to empirically assess aspects of social influence in genetic testing decision-making and to connect these with principlist and relational theories of autonomy. We interviewed 18 adult genetic counsellees without capacity issues considering predictive genetic testing for cancer predisposition for themselves and two counselling physicians in Switzerland. We conducted a qualitative analysis, building on a grounded theory study about predictive genetic testing decision-making. We found that some participants agreed to predictive genetic testing predominantly because relatives wanted them to do it, with some even acting contrary to their own convictions. Others, in contrast, based their decision on purely individualistic reasons but expressed difficulties in explaining their decision to their social environment. Healthcare professionals had a critical influence on decision-making in many cases without being manipulative, as perceived by counsellees. Still, cases of coercion and social pressure occurred within social relationships. In conclusion, predictive genetic testing decision-making includes relational and individualistic aspects, and both are compatible with autonomous decision-making. While the principlist and relational notions of autonomy compete on a theoretical level, they are two sides of the same coin when used as analytical lenses for genetic testing decision-making. Social acceptance of refusal of testing should be improved to mitigate social pressure. Individuals should be encouraged to decide for themselves how much their social environment influences their decision regarding predictive genetic testing.

KEYWORDS

decision-making, genetic counselling, genetic screening/testing, principle of respect for autonomy, relational autonomy

1 | 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'.[1] Even though Beauchamp and Childress acknowledge that social influences are unavoidable and permissible to a

[1]Beauchamp, T. L., & Childress, J. F. (2013). *Principles of biomedical ethics* (7th ed.). New York, NY: Oxford University Press, p. 101.

certain extent,[2] 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 America[16] and the UK.[17] More recently, such studies have been conducted in other European countries such as Italy[18] or Spain[19], 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.

[2]Ibid: 104–105.

[3]Donchin, A. (2001). Understanding autonomy relationally: Toward a reconfiguration of bioethical principles. *Journal of Medicine and Philosophy*, *26*, 365–386; Mackenzie, C., & Stoljar, N. (Eds.) (2000). *Relational autonomy: Feminist perspectives on automony, agency, and the social self*. New York, NY: Oxford University Press.

[4]Stoljar, N. (2018). Feminist perspectives on autonomy. In E. N. Zalta (Ed.), *The Stanford encyclopedia of philosophy* (2018 ed.). Stanford, CA: Metaphysics Research Lab, Stanford University.

[5]Christman, J. (2009). *The politics of persons individual autonomy and socio-historical selves*. UK: Cambridge University Press.

[6]Friedman, M. (2003). *Autonomy, gender, politics*. *Studies in feminist philosophy*. UK: Oxford University Press.

[7]Stoljar, op. cit. note 4; Christman, J. (2004). Relational autonomy, liberal individualism, and the social constitution of selves. *Philosophical Studies*,*117*, 143–164.

[8]Jennings, B. (2016). Reconceptualizing autonomy: A relational turn in bioethics. *Hastings Center Report*, *46*, 11–16, p. 13.

[9]Ho, A. (2008). Relational autonomy or undue pressure? Family's role in medical decision-making. *Scandinavian Journal of Caring Sciences*, *22*, 128–135.

[10]Etchegary, H., Miller, F., deLaat, S., Wilson, B., Carroll, J., & Cappelli, M. (2009). Decision-making about inherited cancer risk: Exploring dimensions of genetic responsibility. *Journal of Genetic Counseling*, *18*, 252–264; Gilbar, R., & Barnoy, S. (2018). Companions or patients? The impact of family presence in genetic consultations for inherited breast cancer: Relational autonomy in practice. *Bioethics*, *32*, 378–387.

[11]Andorno, R. (2004). The right not to know: An autonomy based approach. *Journal of Medical Ethics*, *30*, 435–439; discussion 439–440.

[12]Hallowell, N., Foster, C., Eeles, R., Ardern-Jones, A., Murday, V., & Watson, M. (2003). Balancing autonomy and responsibility: The ethics of generating and disclosing genetic information. *Journal of Medical Ethics*, *29*, 74–79; discussion 80–83.

[13]Lynch, H. T., Smyrk, T., & Lynch, J. (1997). An update of HNPCC (Lynch syndrome). *Cancer Genetics and Cytogenetics*, *93*, 84–99.

[14]Skirton, H., Goldsmith, L., Jackson, L., & Tibben, A. (2013). Quality in genetic counselling for presymptomatic testing — clinical guidelines for practice across the range of genetic conditions. *European Journal of Human Genetics*, *21*, 256–260.

[15]Llort, G., Chirivella, I., Morales, R., Serrano, R., Sanchez, A. B., Teulé, A., … SEOM Hereditary Cancer Working Group. (2015). SEOM clinical guidelines in hereditary breast and ovarian cancer. *Clinical and Translational Oncology*, *17*, 956–961; Stoffel, E. M., Mangu, P. B., Gruber, S. B., Hamilton, S. R., Kalady, M. F., Lau, M. W., … European Society of Clinical Oncology. (2015). Hereditary colorectal cancer syndromes: Amesrican Society of Clinical Oncology clinical practice guideline endorsement of the familial risk–colorectal cancer: European Society for Medical Oncology clinical practice guidelines. *Journal of Clinical Oncology*, *33*, 209–217.

[16]Etchegary et al., op. cit. note 10; Hamilton, R. J., & Bowers, B. J. (2007). The theory of genetic vulnerability: A Roy model exemplar. *Nursing Science Quarterly*, *20*, 254–264.

[17]Foster, C., Watson, M., Moynihan, C., Ardern-Jones, A., & Eeles, R. (2002). Genetic testing for breast and ovarian cancer predisposition: Cancer burden and responsibility. *Journal of Health Psychology*, *7*, 469–484; Hallowell, N., Ardern-Jones, A., Eeles, R., Foster, C., Lucassen, A., Moynihan, C., & Watson, M. (2005). Men's decision-making about predictive BRCA1/2 testing: The role of family. *Journal of Genetic Counseling*, *14*, 207–217.

[18]Godino, L., Jackson, L., Turchetti, D., Hennessy, C., & Skirton, H. (2018). Decision making and experiences of young adults undergoing presymptomatic genetic testing for familial cancer: A longitudinal grounded theory study. *European Journal of Human Genetics*, *26*, 44–53.

[19]Rivera-Navarro, J., Cubo, E., & Mariscal, N. (2015). Analysis of the reasons for non-uptake of predictive testing for Huntington's disease in Spain: A qualitative study. *Journal of Genetic Counseling*, *24*, 1011–1021.

[20]Swiss Federal Council. (2004). *CC 810.12 Federal Act of 8 October 2004 on human genetic testing (HGTA)*. Available at: https://www.admin.ch/opc/en/classified-compilation/20011087/index.html [accessed Jan 23, 2020]: Art 14.

[21]Swiss Federal Council. (1994). *Bundesgesetz über die Krankenversicherung: KVG*. Retrieved from https://www.admin.ch/opc/de/classified-compilation/19940073/index.html. Accessed September 30, 2020.

[22]Abacan, M., Alsubaie, L., Barlow-Stewart, K., Caanen, B., Cordier, C., Courtney, E., … Wicklund, C. (2019). The global state of the genetic counseling profession. *European Journal of Human Genetics*, *27*, 183–197.

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.[23]

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,[24] 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:

[23]Frith, L. (2012). Symbiotic empirical ethics: A practical methodology. *Bioethics*, *26*, 198–206.

[24]Corbin, J. M., & Strauss, A. (1990). Grounded theory research: Procedures, canons, and evaluative criteria. *Qualitative Social Work*, *13*, 3–21.

BOX 1 Case stories illustrating aspects of autonomous decision-making based on five of the 18 interview participants.

Case 1—Mary: Doing predictive genetic testing for her daughter.

One of Mary's grown up daughters underwent genetic counselling because of the family's cancer history. Since Mary's affected sister refused genetic testing, Mary agreed to do it for her daughters. She had been aware that cancer was common in her family, but had always refrained from **predictive** genetic testing. 'For myself, if my daughter hadn't come to me because she wanted to know, I wouldn't have had the test. I really primarily did it for my daughters.' (Mary, 60+, healthy non-carrier)

Case 2—Paul: Torn between his own interests and the interests of his daughter.

Paul is affected by cancer, and since his mother was too, a genetic test was recommended. He initially agreed, thinking mainly about the interests of his daughter, but started to brood while waiting for the test result, doubting that knowing an increased genetic risk would do him any good. 'And sometimes I started to brood, and started thinking, what consequences will this test result have for me… And then I switched sides and thought, no, it's good to know that, for my daughter it makes sense, she is young, she has children, and so on. And then you think about yourself again… […] My wife and daughter really took sides and said, listen, it's not just for you. It's for your daughter as well. And I think it is also reasonable for me to do it. […] So no, I wouldn't have stopped the testing process, that would have been too selfish. And: it wouldn't have left me alone. […] Not doing it would have made me very insecure.' (Paul, 60+, non-carrier in remission)

Case 3—Margaret: Perceiving genetic testing as her own private matter.

After genetic counselling, Margaret, who is healthy and childless, decided not to undergo **predictive** genetic testing despite many cases of cancer in her family. For her, a positive test result would not have resulted in any actionable consequences, just in anxiety and worries. Margaret described her difficulties in asking her family members for information she needed for her pedigree: 'My goal would have been to talk to my relatives in a small circle, and that got a bit out of control. But they don't know anything about my decision now, that is my personal thing, my decision. My friend knows about it, but I haven't told anyone else, that is really my personal thing, yes.' (Margaret, 30+, healthy, untested)

Case 4—Ruth: Doing predictive genetic testing to belong.

Because Ruth's cousin and sister are carriers of a pathogenic genetic variant, they urged Ruth to undergo **predictive** genetic testing, too. Ruth at first perceived this as unnecessary, since she intended to take preventive measures in any case. However, after a while she felt like she needed to know anyway: 'My sister, my cousin and I, we are like a fate community now, and that's good, because now we know we all have the same, we can talk about it, […] we are really on the same boat now. Eh, but it was like, if I didn't get tested, I couldn't have taken part in certain talks, I wouldn't have known if I have the mutation or not… […] Ehm, and that's why I actually wanted to know, too. And also to know, am I in their group or am I, am I outside.' (Ruth, 40+, healthy carrier)

Case 5—Anne: Listening to the doctor's advice.

Anne is affected by cancer, like her sister many years ago, and her treating physician recommended **predictive** genetic testing for a cancer predisposition. Even though Anne was convinced that the test would come back negative (based on her family history) she did not want to go against her treating physician and agreed to do the test. Her test result revealed a variant of unknown significance, but Anne was not interested in the meaning of this: '[My physician] simply said that the test result was somehow, eh, just so that one would classify it as negative, eh… as negative, not positive… But apparently they found something they still cannot decipher today, and they might be able to in a few years, but that was when I noticed that it was getting too complicated for me. I do not want to know all that at all, it seems to me one ends up like running around and thinking, eh, what is slumbering inside me, so… At that point I realized that that is ok for me.' (Anne, 60+)

> 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 counsellees], 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.[25] 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.[26] 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.[27] 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.[28] 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.

Previous studies also revealed that healthcare professionals influence genetic testing decision-making in the way they frame information.[29] This can support autonomous decision-making if counsellees are carefully guided in their decision-making process and adequate information is provided.[30] Relational autonomy, as well as theories on shared decision-making, support this approach.[31] However, this renders the principle of nondirectiveness impractical. While these considerations have been discussed at length in the past decade,[32] Swiss law and German guidelines still utilize this principle.[33]

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.[34] 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,[35] 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

[25]Zimmermann, B. M., Shaw, D., Heinimann, K., Knabben, L., Elger, B., & Koné, I. (2020). How the "control-fate continuum" helps explain the genetic testing decision-making process: A grounded theory study. *European Journal of Human Genetics*, *28*, 1010–1019.

[26]Hallowell et al., op. cit. note 12; Hallowell, N., Arden-Jones, A., Eeles, R., Foster, C., Lucassen, A., Moynihan, C., & Watson, M. (2006). Guilt, blame and responsibility: Men's understanding of their role in the transmission of BRCA1/2 mutations within their family. *Sociology of Health and Illness*, *28*, 969–988; d'Agincourt-Canning, L. (2006). Genetic testing for hereditary breast and ovarian cancer: Responsibility and choice. *Qualitative Health Research*, *16*, 97–118; Etchegary et al., op. cit. note 10; Foster et al., op. cit. note 17.

[27]Dove, E. S., Kelly, S. E., Lucivero, F., Machirori, M., Dheensa, S., & Prainsack, B. (2017). Beyond individualism: Is there a place for relational autonomy in clinical practice and research? *Clinical Ethics*, *12*, 150–165; Gilbar & Barnoy, op. cit. note 10.

[28]Skirton et al., op. cit. note 14; Deutsche Gesellschaft für Humangenetik e.V. & Berufsverband Deutscher Humangenetiker e.V. (2018). S2k-Leitlinie Humangenetische Diagnostik und Genetische Beratung. *Medgen*, *30*, 469–522.

[29]Scott, D., Friedman, S., Telli, M. L., & Kurian, A. W. (2020). Decision making about genetic testing among women with a personal and family history of breast cancer. *JCO Oncology Practice*, *16*(1), e37–e55.

[30]Manson, N., & O'Neill, O. (2007). *Rethinking informed consent in bioethics*. UK: Cambridge University Press.

[31]White, M. T. (1998). Decision-making through dialogue: Reconfiguring autonomy in genetic counseling. *Theoretical Medicine and Bioethics*, *19*, 5–19; Osuji, P. I. (2018). Relational autonomy in informed consent (RAIC) as an ethics of care approach to the concept of informed consent. *Medicine, Health Care and Philosophy*, *21*, 101–111.

[32]Evans, M., Bergum, V., Bamforth, S., & MacPhail, S. (2004). Relational ethics and genetic counseling. *Nursing Ethics*, *11*, 459–471; Pennacchini, M., & Pensieri, C. (2011). Is non-directive communication in genetic counseling possible? *Clinical Therapeutics*, *162*, e141–2144; Weil, J., Ormond, K., Peters, J., Peters, K., Biesecker, B. B., & LeRoy, B. (2006). The relationship of nondirectiveness to genetic counseling: Report of a workshop at the 2003 NSGC Annual Education Conference. *Journal of Genetic Counseling*, *15*, 85–93.

[33]Swiss Federal Council, op. cit. note 20, Art 14; Deutsche Gesellschaft für Humangenetik e.V. & Berufsverband Deutscher Humangenetiker e.V., op. cit. note 28, chapter 1.5.

[34]d'Agincourt-Canning, op. cit. note 26.

[35]Zimmermann, B. M., Elger, B. S., & Shaw, D. (2019). Media coverage of ethical issues in predictive genetic testing: A qualitative analysis. *AJOB Empirical Bioethics*, *10*, 250–264.

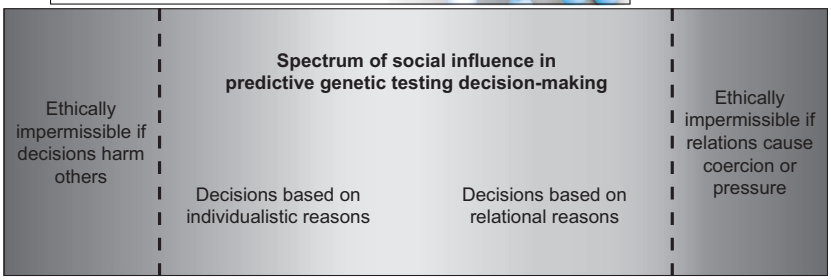

FIGURE 1 Model on the social influence in predictive genetic testing decision-making

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[36] and healthcare professionals.[37] 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.[38] 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[39] and the conceptions of relational autonomy proposed by Friedman[40] and Christman.[41] 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[42] and Friedman.[43] 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.[44] 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;[45] 'non-directive' genetic counselling is mandatory before and after predictive genetic testing,[46] 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

[36]Etchegary et al., op. cit. note 10.

[37]Gilbar & Barnoy, op. cit. note 10.

[38]Ho, op. cit. note 9.

[39]Beauchamp & Childress, op. cit. note 1.

[40]Friedman, op. cit. note 6.

[41]Christman, op. cit. note 5.

[42]Beauchamp & Childress, op. cit. note 1.

[43]Friedman, op. cit. note 6.

[44]Ibisler, A., Ocklenburg, S., Stemmler, S., Arning, L., Epplen, J. T., Saft, C., & Hoffjan, S. (2017). Prospective evaluation of predictive DNA testing for Huntington's disease in a large German center. *Journal of Genetic Counseling*, *26*, 1029–1040; Rivera-Navarro et al., op. cit. note 19.

[45]Swiss Federal Council, op. cit. note 21.

[46]Swiss Federal Council, op. cit. note 20.

person hurts other persons.[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 counsellees, 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.

ACKNOWLEDGEMENTS

We acknowledge the inputs from our colleagues at the Institute for Biomedical Ethics during our internal workshop in September 2019 and Emilian Mihailov's comments on the theoretical part. We thank the medical professionals who helped to recruit the study participants. This research was funded internally by the Institute for Biomedical Ethics, University of Basel, Switzerland, and through a personal grant to BZ from the Freie Akademische Gesellschaft Basel. Open access funding enabled and organized by Projekt DEAL.

CONFLICT OF INTEREST

The authors declare that there are no conflict of interests.

ORCID

Bettina M. Zimmermann https://orcid.org/0000-0001-7047-4496
David Shaw https://orcid.org/0000-0001-8180-6927

AUTHOR BIOGRAPHIES

Bettina Zimmermann graduated in March 2020 at the University of Basel, focusing on informed decision-making in genetic testing. She is also interested in public debates and their influence on public health ethics and currently holds postdoc positions at the University of Basel and the Technical University Munich.

Insa Koné graduated with a medical doctorate from the University of Greifswald in 2011. She currently works in Internal Medicine at the University Hospital Basel and holds a postdoc position at the Institute for Biomedical Ethics at the University of Basel.

David Shaw graduated with an MA in philosophy and English literature from the University of Glasgow in 1999, and an MSc in philosophy from the University of Edinburgh in 2001. He obtained his PhD at the University of Lausanne in 2005. Since 2012 he has worked at the Institute for Biomedical Ethics at the University of Basel, and he is also affiliated with the Department of Health, Ethics and Society at Maastricht University in the Netherlands.

Bernice Simone Elger wrote her habilitation thesis at the University of Geneva on *Ethical issues of human genetic databases: A challenge to classical health research ethics*. Since 2011, she is full professor and head of the Institute for Biomedical Ethics at the University of Basel, as well as, since 2007, associate professor and head of the Unit for Health Law, Ethics and Humanitarian Medicine of the Center for Legal Medicine, University of Geneva.

How to cite this article: Zimmermann BM, Koné I, Shaw D, Elger B. Autonomy and social influence in predictive genetic testing decision-making: A qualitative interview study. *Bioethics*. 2020;00:1–8. https://doi.org/10.1111/bioe.12816

[47]Beauchamp & Childress, op. cit. note 1.