Costs, burdens and the prevention of genetic disorders: what role for professional influence?

Martina C. Cornel¹ · Angus Clarke²

Published online: 23 September 2021

© The Author(s), under exclusive licence to Springer-Verlag GmbH Germany, part of Springer Nature 2021

Community genetics aims to apply ‘genomics knowledge and technologies in human populations and communities *to the benefit of individuals therein*’ (Ten Kate et al. 2010). Contentious ethical issues faced in community genetics include how to manage the tension between a focus on the individual and family on the one hand and the community on the other. Should genetics professionals—and other healthcare practitioners who draw on genetic information or technologies—see their role in terms of the information and choices they bring to individuals or the benefits they bring to communities?

Of course, one response is to say that genetics should bring benefits both to individuals and families and to the communities they constitute. However, this merely evades a difficult question by failing to address it. There are occasions where the interests of the individual and the community may differ. This can happen in either of two ways. One way is for individuals to be ‘persuaded’—or coerced—to behave in a way that is thought to benefit society. In the context of genetics, this might mean that decisions about marriage, having children and the management of individual pregnancies may be guided by society’s wish not to have children with genetic conditions that are viewed as a burden.

The second type of interaction between individuals and society is the reverse. The cumulative decisions of many individuals and families may have consequences for society that no one desires. The collective (public) consequences of individual (private) decisions can be damaging, as with the decisions of many parents in some (especially Asian) countries to terminate pregnancies that would result in a girl being born (Madan and Breuning 2014). The distorted sex ratio at birth is in no one’s interests, but that does not stop gendercide of so many female foetuses, given that the

technology (foetal ultrasound for decades and now also NIPT foetal sexing) is so widely available.

Here, we wish to focus on the first problem, often referred to as the problem of eugenics, although that does not do justice to the web of issues it raises (Raz 2009). In some developed western countries, in Europe and North America in the first half of the twentieth century, this took the form of society making judgements as to who should or would be permitted to marry, with recommendations that certain categories of people (the ‘insane’, the ‘feeble-minded’, alcoholics, ...) should not marry and should not have children. In the later twentieth century, antenatal screening programmes were established to identify and then enable the termination of pregnancies in which the foetus was affected by neural tube defects or Down syndrome. Entry to these programmes was sometimes so routinised that they were difficult to avoid, and women felt pressured to take part and, indeed, often did not experience any element of choice. The pressure exerted was often applied both by the professionals involved, such as midwives and obstetricians, and by society at large, with the parents of infants affected by Down syndrome (for example) being liable to stigma and blame for not having prevented such a birth. We would regard such pressure—by professionals or by neighbours—to be offensive and unethical, an affront to the principle of autonomy of the parents and to the respect and dignity due to the affected individuals. The worst excesses of that time have now passed, we believe, although a degree of inappropriate routinisation may persist in many settings, especially in relation to foetal ultrasound anomaly scanning.

A related type of decision arises in communities that practice customary consanguinity, especially where marriages are arranged by the parents of those to be wed. In such communities, it is well documented that the birth prevalence of serious genetic and congenital disorders affecting children is substantially raised, when those to be married are first cousins or more closely related. Should ‘community geneticists’ (interpreted broadly) concern themselves with this

✉ Martina C. Cornel
mc.cornel@amsterdamumc.nl

¹ Amsterdam UMC, Amsterdam, The Netherlands

² Cardiff University, Cardiff, UK

pattern of marriage? Should they actively seek to discourage consanguinity, or at least close consanguinity, or should this be regarded as a matter for the individuals involved (Bittles 2020)? This formulation of course suggests one way through the problem, if the choice of marriage partner is not being made by those to be married. For professionals to educate the older generation of prospective grandparents or professional matchmakers in the facts of genetics, so as to help them at least somewhat reduce the risks of genetic disorders in the new couple's future children, may be much more acceptable than direct pressure on a young couple not to be married.

How would this work out in societies where the carrier frequencies for specific recessive conditions are high and where carrier screening tests are available, e.g. for the haemoglobinopathies, beta-thalassaemia and sickle cell disease? If we consider only beta-thalassaemia, where the effective treatment of affected children brings major benefits, in effect resulting in normal life, the cost of such treatments may be unaffordable in low- or middle-income countries, especially as the numbers of those to be treated would expand continuously as testament to the effectiveness of the treatment (Sinha et al. 2020). For many countries, treatment is only affordable if the numbers to be treated remain small, when this can be achieved either by steering marriage choices so that a carrier would not marry another carrier, or by identifying carrier couples, who could then be offered prenatal diagnosis and the selective termination of affected pregnancies. What does 'individual autonomy' mean in such a context, and how does one minimise the births of affected infants—to enable the treatment of those who have been born—without unethical, 'eugenic' coercion? The answer has to be through community education, so that those making the decisions—the young couple, or their parents, depending on who chooses whom—understand what is at stake.

In describing such arrangements, and the results of carrier screening programmes where these are vital for the treatment of those affected to be feasible, the language used can raise sensitivities (Modell 2020). If we hail as a great success the low birth prevalence of beta-thalassaemia, are we colluding with an unacceptable eugenics (Dive and Newson 2021)? Or are we celebrating that a (now very small) cohort of affected children is being treated effectively with regular transfusion and iron chelation? How do we present this, to ourselves and to the world?

Departing from a focus on the benefit of genetics to individuals, and instead aiming at benefits to communities at the expense of individuals, would not be seen as acceptable clinical practice. Arguments on the basis of economic gains or eugenics have not been considered appropriate (Ten Kate et al. 2010), and coercion to follow the advice of geneticists in light of increased risks would go against respect for autonomy. There are clear dangers if the effectiveness of

services is to be measured in terms of the reduction of birth prevalence for specific, costly, disorders as the measures may drive the way in which services are delivered. This could result in the ethos and quality of services being distorted and marred by a drive to maximise this metric of effectiveness.

While the journal in the first decade of its existence has been firmly rooted in the cultural tradition of the 'Western individualism', in 2020 it was decided to open the possibility to discuss different stances (Schmidtke and Cornel 2020). In the number 1 issue of 2020, consanguineous marriages were discussed, with on the one hand traditions favouring consanguineous marriages and on the other hand medical practices to avoid infant mortality and morbidity due to autosomal recessive diseases (Bittles 2020). While it can be argued that both strict marriage traditions and medical programmes can endanger autonomy, the editorial proposed that it could also be seen as 'a proper first step to help people finding a way to combat the negative consequences of their traditions' (Schmidtke and Cornel 2020).

In a recent contribution from Iran (Hasanshahi and Khanjani 2021), the authors investigated why couples who both carry beta-thalassemia married despite screening and prevention programmes. They conclude that 'providing thorough and accurate information at the appropriate time can help prevent marriage among couples with thalassemia minor in Iran'. In the contribution, we perhaps also find a diversity of arguments from a country struggling with the question how to help people who carry the condition to act appropriately on this information. The disease burden is large, but participants did not perceive a serious personal threat. They loved each other and were simply unaware of many aspects of the disease and the difficult choices, the stress and anxiety involved in raising an affected child.

Another contribution reports experiences with sickle cell disease in Tanzania, where parents favour premarital carrier screening, some even insisting that it become compulsory (Kisanga et al 2021). They understood sickle cell disease once it had been diagnosed in their child and they attended the paediatric sickle cell clinic. Awareness is limited in the general population, and some parents see a compulsory program as a way to better inform future parents.

The editors and reviewers have read these contributions and considered whether they should ask for a different wording, avoiding the implied coercion and promoting individual autonomy. However, in light of the earlier invitation to talk about contentious ethical issues, it was decided to publish the papers as they are. Meanwhile you are invited to submit your thoughts: how do we help people to use a knowledge of thalassemia and sickle cell disease in their culture and country? We could also consider how wealthy, highly developed countries will respond to a similar challenge, the challenge of very costly treatments for rare genetic disorders. Once enough such diseases have rational, gene-based but very expensive therapies available, will the treatments be made available in practice, or will their potential availability mean

that pregnancies are now screened, with the expectation that an affected pregnancy would be terminated so that the treatment would not be needed? Paradoxically, with a serious, early-lethal disorder, there may be little to be gained (from this reductionist, ‘eugenic burden’ perspective) by population screening until an effective therapy has become available so that the screening can then save money by avoiding.

References

- Bittles A (2020) Population structure and inherited genetic disorders in the Bedouin of the Negev. *J Community Genet* 11:11–12. <https://doi.org/10.1007/s12687-019-00432-9>
- Dive L, Newson AJ (2021) Reproductive carrier screening: responding to the eugenics critique. *J Med Ethics : medethics-2021-107343*. <https://doi.org/10.1136/medethics-2021-107343>
- Hasanshahi F, Khanjani N (2021) Investigating the reasons for marriage among couples with thalassemia minor, in Iran. *J Community Genet*. <https://doi.org/10.1007/s12687-021-00540-5>
- Kisanga E, Mutagonda R, Myemba DT et al (2021) Premarital genetic screening and care of Tanzanian children with sickle cell disease: a qualitative study on parents’ views and experiences. *J Community Genet*. <https://doi.org/10.1007/s12687-021-00539-y>
- Madan K, Breuning MH (2014) Impact of prenatal technologies on the sex ratio in India: an overview. *Genet Med* 16:425–432. <https://doi.org/10.1038/gim.2013.172>
- Modell B (2020) Haemoglobin disorders—a point of entry for community genetics services in India? *J Community Genet* 11:7–9. <https://doi.org/10.1007/s12687-019-00441-8>
- Raz A (2009) Eugenic utopias/dystopias, reprogenetics, and community genetics. *Sociol Health Illn* 31(4):602–616
- Schmidtke J, Cornel MC (2020) Contentious ethical issues in community genetics: let’s talk about them. *J Community Genet* 11:5–6. <https://doi.org/10.1007/s12687-019-00444-5>
- Sinha S, Seth T, Colah RB et al (2020) Haemoglobinopathies in India: estimates of blood requirements and treatment costs for the decade 2017–2026. *J Community Genet* 11:39–45. <https://doi.org/10.1007/s12687-019-00410-1>
- Ten Kate LP, Al-Gazali L, Anand S et al (2010) Community genetics. Its definition 2010. *J Community Genet* 1:19–22. <https://doi.org/10.1007/s12687-010-0007-z>

Publisher’s note Springer Nature remains neutral with regard to jurisdictional claims in published maps and institutional affiliations.