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LETTER TO EDITOR

Letter to the Editor: Time to update the language of genetics from the nineteenth to the twenty-first century: a response to Schmidtke and Cornel

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Promoting respect for the autonomy of individuals, pursuing equity in any advances by health-related genetics and being cognizant of the sensibilities of those communities impacted by either genetic diseases or those policies designed to address them must be keystones of the development of genetics in the 2020s and beyond (Schmidtke and Cornel 2020). But there is an additional, more prosaic agenda to address. We need to update the language of genetics to make it fit for the public discourses it needs to engage with.

We have been very pleased to be part of the wide ROHgen collaboration that reports the effects of autozygosity on a broad range of human phenotypes (Clark et al. 2019). Our contribution involved making data available from the birth cohort study, Born in Bradford (www.borninbradford.nhs.uk). Integral to the long-term relationship we have with the families in our cohort is a commitment to share findings that we have directly generated, or that arise from our collaborations. The findings reported in Clark et al. 2019 offer much that enriches understanding of key areas of concern in relation to autozygosity and human phenotypes. It does, like much research in this field, present challenges in reframing a complex analysis for a lay public. But additionally, and also familiar in the field, the paper generated some anxiety in our cohort research team about the reaction we might get when we share its insights with our cohort participants. We are anxious about the language of genetics and specifically those terminologies that reflect the animal models that have been important in its evolution as a discipline, including "in-breeding", "mating" and "pedigree".

Advances in the understanding of genetics in medicine have seen a shift from a focus solely on discovery into a wider focus that now includes application. When such a shift occurs, different constituencies of interest are engaged and different questions raised. The insights of genetic science need to be translated into the language of clinical medicine and, if actions that impact on patients are to result, into the language of risk and of human costs and benefits.

Interdisciplinary interactions are challenging because of the different assumptions that underpin the dominant paradigms of practice. But the interactions between genetic scientists and clinical scientists are facilitated by a shared scientific epistemology. To fulfil the potential of genetics in terms of wider implementation, a bigger challenge exists in engaging with the general population (Table 1). The tropes of our contemporary debate on genetics centre on its potential to illuminate lifetime propensities and risks and to make possible personalised medicine. In this context, at the very least, the general public need to understand the potential and the limitations of genetic science and clinical genetics if they are to exercise informed consent as they participate in research or avail themselves of treatment. Furthermore, the public need to support the scientific endeavour by making themselves available as research subjects—agreeing to provide genetic material for example—and by supporting the political will that will ensure appropriate resource allocations for scientific and medical advance. Achieving the shift in scale that twenty-first century genetics seeks requires a language that invites the public in, not a language that invokes their fears or stigmatises them. There are many precedents that illustrate the importance and the impact of shifts in the language of medicine. It is not such a long time ago when terminology included "cripples" and "the subnormal"; we talk of seizures not fits because "fits" is not precise and because it has a connotation that is stigmatising; we have disabled people rather than "the disabled", and we

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Table 1 Constituencies of interest engaged and language needed for wider implementation of genetics

Shifts in focus/ paradigm shifts	Disciplinary focus	Key personnel	Language needed
Discovery	Genetic science (in animals and humans)	Academic and commercial scientists/funders	Scientific precision/adding to established understandings
Utility/implementation	Clinical science	Clinicians/doctors/regulators/commissioners	Benefits (and costs)/risk
Acceptability	Behavioural science/social science	Public health/health education/voluntary sector	Inclusive/understandable/non-stigmatising

talk of labour not delivery because we are reminded of who the most important party in the birth is!

That part of our research agenda that most pertains to these issues concerns autosomal recessive disorders in the context of a population with high levels of consanguinity (Sheridan et al. 2013.) Achieving any behaviour change that builds on an understanding of genetic risk requires our target population to accept the veracity of what we say, to recognise our benign intentions and to be clear about the actions that follow the insights of genetic and clinical science that we seek to impart. To do this, we have to invoke a language that is non-stigmatising. A service review by Salway et al. (2016) captures the challenges in accessing target communities for genetic counselling and testing, citing the need for approaches that engender trust and that are not seen as stigmatising. Conversely, when communities are approached with sensitivity, using culturally appropriate and accessible language, initiatives are responded to positively (Darr et al. 2016). In an analogous area attempts to encourage weight loss via approaches that are seen as objectifying, shaming and judgemental are counter-productive (Muenning 2008).

The scientific precision and legacy importance of terms like "pedigree" or "in-breeding" may benefit one phase of genetics evolution but do not serve well the shift into a population-based discourse. Nor does a too easy conflation of genetic risk with a social practice (a preference for marriage between blood relations in this case). While there are not easily available synonyms for legacy terms, we might consider "ancestry" or "inheritance" instead of pedigree and "related by blood" instead of in-breeding. But the development of a new terminology would benefit from the wisdom of crowds. We invite colleagues to engage in a debate about an appropriate language for the genetics of the twenty-first century—a language that is both precise and inclusive.

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