Chapter 27

Disability in DOHaD and Epigenetics

Towards Inclusive Practice

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27.1 Introduction

Developmental Origins of Health and Disease (DOHaD) and epigenetic research that investigate causal mechanisms and predictive biomarkers have often occurred in the absence of discussion of ethical, legal, and social implications or engagement with disability communities. This has often led to maternal blaming, labelling, stigmatisation, and ableism. Considering the debate on different models of disability by disability activists and social scientists, this is a timely opportunity to optimise the design of epigenetic research into conditions labelled as disabilities. Research aims should address the needs of disability communities, acknowledge diversity, and move away from medical to social models of disability.

Our chapter considers the implications of epigenetics research, as a mediator of DoHAD, for people with autism, an example of a condition some label a disability. We discuss how views on epigenetics and autism have changed over time, including how research can enhance the lived experience of autistic people through contributions to understanding how autism develops and how the strengths and needs of autistic people can best be identified and supported. We argue there is a need for researchers, including those with autism, to work with autistic people and their supporters to co-design studies promoting this understanding, centring autonomy and the provision of information to autistic individuals, including whether to engage with current and future epigenetic tests, particularly those available direct to consumers. In summary, we urge researchers planning such studies to first engage meaningfully and non-tokenistically with disability communities and continue to engage through to the writing and dissemination phases of their research.

27.1.1 On Terminology

Genetics research and autism studies have a complicated history, so we begin by establishing our choice of terminology and rationale for this. We acknowledge that there are strong and often polarising views about the issues presented in this chapter; however, we hope that we can contribute to meaningful discussion.

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The principle 'nothing about us without us' communicates that decision-making that impacts a particular group should not take place without the full and direct participation of its members [1]. Thus, it is crucial that individuals be referred to using their preferred terminology. Person-first language evolved in the 1970s to separate the person from the descriptive trait, for example 'a person with autism' rather than 'an autistic person', and to give primacy to their identity as a person. Although this is well-intentioned, some disability activists have noted that this forced separation between person and trait reinforces the idea that disability is inherently negative and ignores the integral role that disability plays in shaping a person's character and experience. As such, there has been a move towards identity-first language.

This terminology is by no means ubiquitous. A person-centred approach to language recommends that on an individual level, words that people use to self-describe should be prioritised.[1] For coherence, we have chosen to use 'autistic person' here, except where a direct quote incorporates other terminology. This is reflective of the preferred language identified by many autistic individuals and autism self-advocacy organisations [2].

27.2 Disability Politics in the Framing of Health

Disability studies emerged in the 1980s and engage with the concepts and consequences of disability, exploring, among other topics, what it means to be disabled in relation to the self and society [3]. Critical disability theory, which focuses on analysing and dismantling systems of ableist oppression, sits at the intersection of academia and activism [4]. Systems that privilege able-bodied people over those with disabilities are not only concerned with understanding the impacts of pathologisation but also undoing them. While there is no single approach to disability studies or politics, both consider the importance of centring and uplifting the stories, voices, and perspectives of disabled individuals in all disability work [1].

27.2.1 Models of Disability

Two dominant models of disability are often contrasted in literature and practice. The medical model ties disability directly to the body, focusing on possible interventions to bring it to a particular type of functioning [5]. The social model situates disability in the social context and physical environment of the individual and is focused on identifying barriers that prevent full participation in society. The latter model differentiates between impairments – attributes impacting how the body and brain operate – and disabilities – restrictions imposed by societal standards that reflect normative ideas of how bodies *should* function. A third model is 'neurodiversity', a term first coined by autistic sociologist Judy Singer and popularised by Steven Silberman in his book, *Neurotribes,* in which he defines it as follows:

> the notion that conditions like autism, dyslexia, and attention-deficit/hyperactivity disorder (ADHD) should be regarded as naturally occurring cognitive variations with distinctive strengths that have contributed to the evolution of technology and culture rather than mere checklists of deficits and dysfunctions. [6]

[1] Of relevance here to this topic that this might include a wide-ranging list of terms, including 'autistic person', 'person with autism', 'Autie', 'Aspie', 'person with Asperger's', 'person on the spectrum', and many more.

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Like the social model of disability, neurodiversity emphasises the disabling nature of stigmatisation and the prioritisation of brains classified as 'normal'.

27.2.2 Disability in Research

To de-pathologise disability requires engagement with disability communities and scholars in developing frameworks from research design to knowledge translation. Sometimes referred to as participatory or community-engaged research [7], evidence indicates this approach contributes to better health and social outcomes. It is critical to respect the contributions of disabled scholars, activists, and organisations and promote collaboration between disabled and non-disabled researchers, and disabled participants and their advocates. Participatory research means an increasing understanding of disabled individuals as co-creators of scientific knowledge, rather than passive subjects.

There is, understandably, hesitation in disability communities regarding participation in medical research. As with many vulnerable communities, the history of unconsented research and other research harms is long and fraught [8]. Community members are quick to spot ableist rhetoric and stigmatisation in research documentation and are reluctant to participate if their bodies, lives, and experiences may be used to pursue goals not aligned with the expressed needs of disabled individuals. Thus, participatory research does not begin with inviting disabled individuals as research subjects, but rather, with listening, learning, humility, and trust-building on the part of non-disabled researchers, using the principle of co-design and through participant advisory groups.

27.3 Mapping Disability onto DOHaD and Epigenetics

As discourse shifts from the medical model, bioethicists and clinicians have begun to recognise how social factors play a primary role in the treatment of disabled individuals. The DOHaD model represents a particularly fruitful opportunity for this shift, focused on a bio-psycho-social model of health and disease [16]. Similarly, epigenetics' attention to the role of environment, exposures, and stress moves away from the biological determinism of the genomics era [17] towards a more holistic understanding of health. Nonetheless, researchers in DOHaD and epigenetics should refrain from importing potentially harmful presumptions into these emerging fields, with bioethicists and disability scholars already expressing concerns that applying the medical model to these areas risks intensifying rhetoric around responsibility and blame for social and environmental exposures, particularly when associating maternal exposures with future disability [18, 19].

The DOHaD phenomenon is supported by ample animal and human evidence but has an intrinsic focus on 'health vs disease'. This neglects natural variations not classified as 'health' or 'disease', including a wide range of ongoing or recurring behaviours, cognitions, and health conditions that are multidimensional. These include neurodiverse conditions such as autism, whose communities refute the labels of 'disease' and 'disability', similarly to the deaf community. This is relevant when attempting to apply epigenetic models of disabilities to traits that cannot reasonably be classified as 'disease symptoms'. Therefore, we have a responsibility to be careful with terminology when engaging with participants from the disability community, including when planning and reporting epigenetics research.

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27.4 The Case of Autism

Autism presents a valuable case study to explore the intersection of disability, DOHaD, and epigenetics, as a condition that has long oscillated in medical and public imagination between having social, environmental, or biological origins. DOHaD research has shown that early-life exposures to social, biological, and environmental factors can influence fetal development. Influential biological factors include maternal infection and inflammation, which can lead to a state of maternal immune activation where immune regulatory mediators are expressed in higher-than-normal ranges, a possible risk factor for autism and other neurodevelopmental and psychiatric conditions [20, 21]. Suboptimal nutrition before or during pregnancy, particularly vitamin B9 (folic acid), has also been implicated [22], as well as prenatal exposure to traffic-related air pollution and some insecticides [23]. Factors that cannot be explained by shared genetics and environment have also been associated with autistic traits, for example in one twin from a genetically identical pair.

Despite a strong genetic influence, there is considerable genetic heterogeneity across autistic individuals [24]. Around 5 per cent are also diagnosed with a clinically and genetically diagnosable syndrome, and around 15 per cent can be attributed to simple genetic changes such as single gene mutations or copy number variations. For the remaining individuals, evidence points to autism as a polygenic condition, that is resulting from genetic differences spread across hundreds, possibly thousands of genes [24]. These genes appear commonly involved in brain development, epigenetic regulation of gene activity, and metabolism, suggesting possible causal mechanisms for autism. Since the early 2020s, autism-associated variants have been grouped together to form a 'polygenic risk score', with a higher score theoretically indicating a higher likelihood of autism [25].

There are more genetic differences in genes encoding components of epigenetic mechanisms in autistic people as a group compared to non-autistic people. As their gene products are likely to act at multiple genomic regions, some autism-specific epigenetic differences will likely have strong genetic components, [26] increasing their likelihood of being stable over time and therefore useful as diagnostic and prognostic biomarkers. Associations between epigenetic states in the sperm of fathers of autistic children compared to those with neurotypical children [27] are more likely to be explained by genetic factors, unless genetics are controlled for, for example, in identical twin studies.

Epigenetic studies of autism have identified similar genes and gene functions to genetic studies, including those involved in epigenetic regulation and synaptic function. However, far more immune system genes have been identified in epigenetic studies of autism diagnoses [34]. Epigenetic studies have also investigated specific dimensions of autism, for example social communication [28], potentially predicting biomarkers at birth [29], and risk scores [30]. However, these findings have yet to be replicated.

27.4.1 The Social Construction of Autism

Criteria for autism in the Diagnostic and Statistical Manual (version 5) include but are not limited to 'persistent deficits in each of three areas of social communication and interaction plus at least two of four types of restricted, repetitive behaviours' [31]. This

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deficit model, at times focused on the external viewer's perception of autistic experiences, typically shapes research seeking to minimise these behaviours and accompanied distress. By contrast, neurodiversity-focused groups frame autism as a constellation of strengths and challenges across social and sensory spectra and focus on research and resources to support autistic individuals in achieving their best quality of life [32].

Another current view considers autism as a potentially disabling condition that nevertheless may confer various positive traits [33]. However, in many cases this view has merely re-circumscribed capitalist values of productivity, for example celebrating those autistic traits, such as hyperfocus, that can be exploited by employers to improve work output. While this view has partly enhanced our understanding of neurodiversity and the need for better neuroergonomics in the workplace, the ultimate focus has not been on promoting quality of life for autistic people. Moreover, other autistic traits that are considered neutral or positive within the autism community, such as stimming to self-soothe and express emotions, are still misunderstood as negatives or viewed with discomfort.

The experiences of autistic individuals in healthcare provide an opportunity for examining pathologisation of their condition via scientific research into DOHaD and epigenetics. These research areas rely heavily on the interpretation of links between social and other environmental factors with biological outcomes. Perhaps the most widely recognised image of the autistic individual is that of a white, masculine-presenting child with an inability to make eye contact, limited or stilted speech, and a fascination with patterns or trains. This perception has recently begun to shift, prompted in part by an increase in later-in-life diagnoses in cisgender women as well as non-binary individuals and transgender men and women, who may present differently from this stereotype.[2]

Autism and diversity of gender identities and experiences overlap substantially, further impacting access to appropriate support and care. Gendered differences in presentation have led autistic girls and women to be underdiagnosed, misdiagnosed, or diagnosed at a later age, sometimes only after their own child's diagnosis [34]. This discrepancy has contributed to a lack of understanding of key mental health conditions that co-occur alongside autism, including eating disorders, depression, anxiety, and suicidality. Similarly, disparities in the impact of race and ethnicity on the timing and frequency of diagnosis have led to a paucity of resources and support for racialised/ethnic minority autistic youth, who at the same time experience increased risks and rates of police and other state-sanctioned violence and incarceration [35]. There is an urgent need for an intersectional approach to all disability research, but particularly epigenetic studies examining the social and environmental contexts for the lived experiences of autistic and other disabled individuals.[3]

[2] Cisgender refers to the experience of having a gender identity that aligns with the sex that is assigned at birth, whereas transgender refers very broadly to the experience of these elements not aligning in some way.

[3] Intersectionality is a term coined by Black critical theorist Kimberlé Crenshaw to describe how oppressive institutions (e.g. racism, sexism, homophobia, ableism, etc.) are interconnected and cannot be examined separately from one another.

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27.5 Reframing Epigenetics Research to Address the Needs of People with Disabilities

27.5.1 Biomarker Development for Conditions Classed as Disabilities

We are still far from having reliable predictive or diagnostic genetic or epigenetic biomarkers for conditions such as autism. One major factor that clouds the interpretation of such research is study design. Most classify autism as one entity, whereas it is a highly heterogeneous condition. Furthermore, co-occurring conditions such as ADHD are largely ignored in such studies. Some researchers have turned away from a categorical to a dimensional approach to the origins of autism, using continuously variable dimensions such as anxiety, attention, sensory processing, specific interests, repetition, social interaction, and communication [36]. We suggest that this method is preferred because it targets traits that can be clinically defined and can identify areas of strength as well as areas in which autistic individuals may require understanding and assistance. This approach also captures intersecting dimensions of co-occurrences, such as ADHD, for example, sustained attention.

We suggest that future studies be based on dimensions of autism with a view to meeting the self-determined needs of autistic individuals and the autism community. A dimensional approach also reflects the spectrum of neurodiversity within and outside the autism community and the reality of the social model of autism rather than the medical model.

As the field of epigenetics moves towards identifying more biomarkers for conditions and associating these with developmental, social, and environmental correlates, the rhetoric surrounding curative approaches to disability could increase. This rhetoric is closely tied to medical and deficit models of disability, with their foundational assumptions that people with disabilities wish to be rid of the disabled parts of themselves. For some, this may be true; the existence of the social and neurodiversity models does not detract from the struggles precipitated by certain features associated with disability, such as chronic pain, anxiety, loss of quality of life, or early mortality. Rather than attempting to categorise disabilities wholesale as 'bad' (e.g. where we may aim to repair or alter the body or brain) or 'good' (where we may instead target disabling factors in the society or environment), a more useful account would examine components of disability that are *unwanted by the individual who experiences them.* Again, following the principle of 'nothing about us without us', it is important to differentiate between calls for prevention and cure that come from researchers and healthcare providers, and policies based on the lived experiences of disabled individuals and their advocates. In doing so, a stark divide can appear between the expectation of the disabled experience and the reality.

It has long been argued that health economic metrics, such as the quality-adjusted life year (QALY) or disability-adjusted life year (DALY), are not sensitive to the real experiences of disabled people and ignore the significant adaptive ability of individuals [37]. Inviting more conscious consideration of disabled peoples' own experiences of their disability can also avoid the tendency to objectify disabled persons' bodies and view them as separate from the disabled experience. This helps avoid the risk of ignoring meaningful needs assessments conducted by the community. In other words, embracing a neurodiversity model does not mean neglecting to provide support for autistic individuals who consider certain traits to be personally disabling or undesirable. Ethically, it is

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important to remember another classic phrase in the disability community, coined by autism advocate Dr Stephen Shore: 'If you've met one person with autism, you've met one person with autism' [38]. Again, the diversity of manifestations and personal experiences can only be incorporated effectively into epigenetics and genetics research if studies are co-designed and guided by diverse members of the autism community.

27.5.2 Direct-to-Consumer (DTC) Epigenetic Tests

In the past five years, there has been a growing number of companies selling epigenetic tests directly to consumers, that is without the need for a referral from a healthcare practitioner [39]. Despite being unable to define a 'healthy' epigenome, DTC companies focus on identifying epigenetic biomarkers in consumers' blood or saliva samples with the promise of enabling consumers to improve their health outcomes. An 'altered' epigenetic status could indicate early-life exposures that increase the likelihood of developing certain conditions, which could be targeted for intervention due to the potential reversibility of epigenetic changes. Identifying environmental risks for the development of a condition also means that prevention strategies could be adopted to reduce the chance of its development, for example, via diet and exercise changes. In the case of autism, there are currently tests being developed to facilitate diagnosis in children as young as 18 months old [40]. Here, the promise is to provide biological data to complement more subjective analyses to expedite autism diagnoses and access to early intervention and resources.

However, DTC epigenetic testing raises various ethico-legal issues, related to the core technical issue surrounding the precision of epigenetic biomarkers for diagnosing complex conditions. Marketing that overestimates the reliability of epigenetic test results could exploit consumer trust in science to sell a product that falls short of its promises. Test results could also affect an individual's access to insurance policies, particularly life insurance, as the reporting of test results is often a legal obligation of the applicant. With a focus on environmental risks, there is also a tendency to blame individuals for the development of associated conditions. Here we acknowledge the long history of blaming mothers for autism, a fact well demonstrated by the term 'refrigerator mothers'[4] often used to describe them [19]. While parents of children with autism seem to support the development of epigenetic testing for improvement of the diagnosis process [41], there is a need for clear regulations of the DTC market to protect consumers, especially vulnerable populations.

27.5.3 Ethical, Legal, and Social Implications of DoHAD Research for People with Disabilities

From an ethical perspective 'respect for persons' is one of the fundamental tenets of Western biomedical ethics. Its application in DOHaD research is often more complex than in standard clinical care [42]. Core ethico-legal issues here include maintaining confidentiality and privacy, and gaining informed consent for medical interventions, which work together to promote autonomy. While protecting sensitive information such

[4] "Refrigerator mothers" refers to a discredited mid-20th-century theory that cold and unemotional parenting, particularly by mothers, was the cause of autism.

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as medical diagnoses and treatment decisions may be relatively simple within the practitioner–client communication paradigm, genetic information, for example, might be problematic for the individualistic Western ethico-legal model. (See Karpin in this volume.) From a DOHaD perspective, genetic and epigenetic information might best be conceived of as *family* information, making privacy concerns more complex. However, the rationale behind protecting this information remains the same: promoting autonomy, including through avoiding potential coercion from those who would misuse sensitive information to discriminate against individuals. The latter is relevant for accessing employment, health and life insurance, and healthcare services. Whether DOHaD and epigenetics research should aim for family or community, rather than individual consent, falls beyond our scope here, but we recognise that when a test impacts more than the individual, there is potential for social harm against others who are impacted by the results. For research on communities with disabilities, especially those with a potential genetic contribution, this suggests the co-design of research studies is important to ensure knowledge about inheritance is not weaponised against the community or used to engage in blaming or labelling of parents or offspring.

Confidentiality is a key pillar in the doctor–patient relationship protected under common law and statutory regulation. For example, privacy laws in Australia governed under the *Privacy Act 1988* (Cth) have a broad reach, protecting a range of information, including health information. According to the 'For your information: Australian Privacy Law and Practice' ALRC Report No 108) [43], 'privacy' covers several aspects, including data protection, such as medical and government records; bodily privacy, such as invasive procedures that may include genetic and epigenetic tests; and communication, such as emails.

The disclosure or privacy of sensitive genetic information in some instances might be problematic. For the disability community, it is possible that epigenetic data collected from one consenting individual or family may have immediate relevance to other community members. For this, the right 'not to know' might be as important as the right to know the genetic factors involved in the development of autism. Importantly, once these data exist, they may have wide-ranging impacts on members of the community who did not consent to the research.

27.5.4 Incorporating Perspectives from Multiple Stakeholders

A goal of disability activists has been to reframe conversations about disability, health, and disease away from views that centre concepts of 'normalcy' and 'functionality' and to instead centre the disabled individual as the core stakeholder in the discussion of their own body and experience. Throughout the twentieth century in particular, the concept of 'wellness' came to be equated with 'virtue', situating the body as a 'site for moral action' [44] with regard to the pursuit of health. The medical model, in addition to enforcing the idea of a 'normal' state of the body to which its owner should aspire [45], increasingly pushed a 'functionality' argument that privileged a body's capacity to contribute to labour [46], and disdained disability precisely because of the implication that the disabled individual is of inherently reduced worth under capitalism.

The term 'stakeholders' is suggestive of a consumer-driven approach to health and well-being that places disabled individuals immediately at a disadvantage [47]. As a result, the stakeholders most often centred on disability research have been medical

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practitioners and families of disabled individuals. While both caregivers and practitioners have a significant interest in the disability conversation and valuable experiences to contribute, at times, this has come at the expense of the voices and narratives from disability communities and their advocates. In autism research, this has contributed to frustration and conflict. As this research moves forward, it must include autistic individuals and their caregivers where necessary (as participants, researchers, and scholars) at the centre of the conversation from research development to knowledge translation.

27.6 Conclusion: Recommendations for Engagement with Disability Communities in DOHaD and Epigenetic Studies

It is essential to engage with disability communities and their supporters at every step from research design to knowledge translation. Previous experiences within these communities highlight the risks that genetic research can lead to discrimination and stigmatisation, and in the case of DOHaD, this extends not only to individuals with the condition of interest but also to their parents [48]. For this reason, we advocate for more inclusive research practices that build trust with disability communities, listen to their needs, and promote support, while maximising autonomy, dignity, and respect for all members of the community.

In the case of autism, we call on researchers to reflect on their motivations when planning epigenetic studies of autism, considering whether predictive testing prior to the typical onset of symptoms would allow for early modes of support [49]. We urge researchers to seek advice from the autistic community when studying environmental contributions to autism to consider structural frames aimed at policy change in addition to those focused on the agency of individuals. Researchers should also be mindful of the language they use in planning and reporting research findings and of adopting a dimensional framework for cognitive assessment.

Future studies may look at the ethical implications of handling and releasing wide-scale epigenetics research data on autistic communities to ensure knowledge is used to meet the needs of this community and improve the quality of life. In summary, we urge researchers planning DOHaD and epigenetics research to listen to and engage with disability communities when they say, 'nothing about us without us'.

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