De novo kin
sharing data, shielding persons, and forging relatedness in precision medicine
Vive, Laura E. Navne; Svendsen, Mette N.

Published in:
Journal of the Royal Anthropological Institute

DOI:
10.1111/1467-9655.13817

Publication date:
2022

Document version
Publisher's PDF, also known as Version of record

Document license:
CC BY

Citation for published version (APA):
De novo kin: sharing data, shielding persons, and forging relatedness in precision medicine

Laura E. Navne VIVE – The Danish Centre for Social Science Research

Mette N. Svendsen University of Copenhagen

Comparing, sharing, and shielding children's biological and biographical data in genetic databases and on Facebook are central moves when geneticists and families search for diagnoses for children with rare diseases. Based on ethnographic fieldwork in Denmark, we show that the work of linking children carrying the same genetic mutations forges new sibling-like forms of relatedness between them. With the concept of ‘datasociality’, we add new layers to ‘biosociality’ by capturing the ways in which biological information is increasingly mediated by digital and algorithmic processes in the genomic era. In the process of both sharing data and securing anonymity, unrelated children become related through qualities of otherness.

This article investigates issues of anonymity and relatedness which are brought to the fore in the use of international genetic databases when diagnosing children with rare diseases. The ethnographic material is drawn from Denmark, but speaks to much broader empirical discussions of the management of transnational health data and to anthropological theories of kinship. The starting point of this article is the ethnographic puzzle that for doctors to diagnose a child with a rare genetic disease, they need to compare one child’s data with the data of multiple other children, while respecting each child’s right to remain anonymous. In the process, the sharing of data in genetic databases and on Facebook comes to form recognition and affinity between the children.

Sitting in on genetic counselling sessions of children with rare genetic diseases in a Danish clinic in 2018, we learned that to connect and track family disease histories and inheritance through generations, a genetic test and a family tree were not enough to facilitate a diagnosis. For the children featured in this article, disease does not run in the family. Instead they carry de novo disease-causing genetic mutations, which appear for the first time in the history of their families and are not inherited from
their parents. Therefore, the geneticists must look beyond the family tree and consult international genetic databases and the scientific literature to confirm whether these de novo mutations are in fact disease-causing. The databases used to diagnose children with rare diseases contain information on mutations found in hundreds of thousands of children and adults around the world. By comparing genetic mutations found in these individuals, the geneticists aim to link gene and disease. This digitally mediated linkage work is a crucial building block in current strategies to make medical diagnostics and treatment more precise, and is often called 'precision medicine'.

The comparisons facilitated by genetic databases are strictly based on gene mutation (named genotype) of individuals, and sometimes also a few details about their symptoms: for example, an extra toe, a large head, a learning disability, or childhood obstipation (named phenotype). Importantly, in the database used by the Danish geneticists whom we followed, both genotype and phenotype are considered confidential and appear only in anonymized form. Respecting the right to privacy and ensuring anonymity are default bioethical standards in research and treatment. In the legal guidelines regulating health data, ‘anonymity’ means that data cannot be traced back to the person they come from (Prainsack 2019; Yakowitz 2011). Nevertheless, questions about encroachments on privacy have proliferated with developments in precision medicine (Hoeyer, 2019; Prainsack 2017; Reardon 2017). Precision medicine, in short, holds the ambition of using genomic data, in combination with many other kinds of personal data, to develop faster, better, and more precise diagnostics and targeted treatments for the individual (Chan & Ginsburg 2011; Perlman & Govindaraju 2016). Existing social science literature has addressed the potential privacy rights violations (Kulk & van Loenen 2012) as well as the public health advantages of liberal data sharing, and the responsibility that data-sharing rhetoric puts on patients to share personal data as a way of contributing to the common good of society (Prainsack 2018; Prainsack & Buyx 2016; Starkbaum & Felt 2019). To our knowledge, nobody has addressed the forms of sociality emerging within the space between shielding persons to ensure anonymity and sharing data to create more precise diagnoses. This is what we set out to do in this article by investigating the forms of relatedness emerging with the new infrastructures of precision medicine.

In the complex management of data we have studied, anonymity is not a stable state, nor a straightforward marker measured by name, face, or personal identification number. Rather, anonymity comes in various degrees and scales, and must be reinterpreted and shaped to facilitate comparisons and connections between children with rare diseases in international databases. Recent studies in bioethics and anthropology have introduced the idea of ‘relational privacy’ to recognize that consenting to genetic information involves not only individuals but also families (Goldim & Gibbon 2015; Ursin 2008). In line with these insights, we approach anonymity as a relational phenomenon. Yet, where previous studies have focused on ‘biological’ relations (how the return of genetic test results may affect biological siblings and parents), we uncover how anonymity becomes part of creating sibling-like forms of relatedness. We argue that a form of ‘datasociality’ emerges when digital databases and algorithms make geneticists and families experience recognition and affinity between strangers. Were it not for the interface between geneticist, patient, databases, and sometimes Facebook, these strangers would most likely not have been connected across huge geographical distances and strict privacy protection regulations. In this datasociality, the children are both ‘genetic strangers’ to the families they live...
with (Rapp 2000) and ‘kin’ to the anonymous others with whom they share a de novo mutation. The sociality at stake in ‘de novo kin’ is as much about strangeness as it is about familiarity and closeness.

The study
Between 2017 and 2021, we followed the emerging field of precision medicine in Denmark in a research project that set out to explore its social, ethical, and legal aspects. While the first author has studied the case of precision medicine in the field of rare disease, the second author has focused on the general consolidation of precision medicine in Denmark. Conducting fieldwork in a department of clinical genetics, the first author followed twelve families and children with rare diseases through their diagnostic processes, and interviewed them in their homes about their expectations, concerns, and experiences of new genetic technology and the knowledge it produces. The first author presented every patient and family with an information sheet about the research project and asked for oral consent for both participant observations in the clinic and interviews in their homes. The families rarely read the information sheet and all consented without any questions. This response was parallel to what we observed in conversations around consent forms between geneticists and families. The families were generally happy to contribute to research which they default expected to benefit other patients in the future. Moreover, their body language communicating ‘let’s get over with this “formal consent procedure”’ expressed their trust in state-employed professionals and their expectation that data from public registries can be shared for the benefit of the public good. For example, according to the Danish Health Law, consent to treatment presumes consent to registration and banking of health data and such data can be retrieved for administrative and research purposes without additional consent from patients. Moreover, ethics approval is not required for qualitative research in Denmark.

Observing closely the counselling work of two geneticists, the first author interviewed eight geneticists in total. From this fieldwork, we learned that connections based on geno- and phenotypical similarities were forged through an exchange of photographs, illness stories, and doctors’ accounts. Moreover, we observed that different forms of relatedness emerged when geneticists asked the families to consent to their child participating and sharing their genetic data in scientific international databases and publications, to facilitate comparison of children with the same rare disease. The second author attended numerous public seminars on precision medicine and interviewed more than twenty-five professionals in the field. In all, we have conducted more than forty interviews with families, geneticists, microbiologists, researchers, and administrators. In following the establishment of new infrastructures for collecting, analysing, storing, and sharing genetic data in a Danish context, we became aware of the central role of Facebook groups and genetic databases and the challenges related to anonymity in modern genetic diagnostics of rare diseases. While we have not conducted fieldwork on Facebook or pursued a ‘digital anthropology’ (Boellstorff 2012), in this article we include interlocutors’ reflections on their use of Facebook and experiences of feeling connected through digital databases. We also refer to interlocutors’ practices of anonymity as told to us by them in face-to-face interviews. In addition, we draw on long-term familiarity with the Danish healthcare system, in particular our previous studies of the introduction of new medical technology which we have researched together and individually since the early 2000s.
Social scientists addressing the ethical aspects of genomics have almost become default members of huge biomedical research projects (Reardon 2017). We were funded independently of the clinicians and researchers we followed, yet our regular participation in policy meetings, and the talks we gave in clinics, in medical societies, and at public events, provided us with great insights into the professionals’ creative work of navigating anonymity guidelines; and as part of our engagement, we participated in shared reflections with them. At the same time, as independent critical anthropologists, we continually created links between our empirical field and anthropological studies on sociality and relatedness in settings far removed from clinical genetics and the use of databases. It is from this engaged, yet critical, position that we weave together data practices and kinship theory.

Relatedness and anonymity
In the early 2000s, the new kinship studies approached kinship as a practice of relatedness (Carsten 2004; Franklin & Roberts 2006; Howell 2003). Following David Schneider (1984), this scholarship challenged ideas about kinship as constituted by consanguinity. When Janet Carsten first introduced the concept of ‘relatedness’ in her work among the Malays on the island of Langkawi, she investigated the processes of becoming complete persons and kin through living and consuming together in houses (1995: 223). Inspired by Carsten, anthropologists around the world came to attend to the ways people act out, conceptualize, and experience social relations instead of assuming an a priori meaning of ‘kinship’. Uncovering kin making among people who do not share genetic or sexual bonds, or a marriage contract, these studies showed how sharing space, food, and work become central to forming relatedness (Carsten 1995; Franklin & McKinnon 2001; Govindrajan 2018).

We suggest that the use of genetic databases in precision medicine presents an interesting case of the formation of relations between humans who share neither descent nor dwelling. But how are connections between children in the same database formed? What kinds of sociality do the connections give rise to? For these children, becoming complete persons and kin entails a sharing of identity and materiality vastly different from sharing food and shelter. They share genetic alterations and come to experience an affinity linked to a sense of similarity in physical appearance, biography, and imagined future trajectory.

With one exception, the children we discuss in this article only meet in the clinic in absentia and in non-identifiable ways. For instance, the children hear of each other through the geneticists’ rather detailed descriptions of intimate details about other children’s medical histories and physical appearances. And the children see each other through the photos of other children presented to them in scientific publications or on Facebook. This way of meeting has parallels to what Matei Candea (2010) describes as ‘anonymous introductions’. During his fieldwork in Corsica, he observed that his interlocutors in first-person encounters avoided the exchange of personal names to enable a social relationship. Similarly, when geneticists become involved as very specific connectors for the children – matching and comparing children with rare diseases in genetic databases and in research communities while shielding their identities – they connect people through ‘anonymous introductions’. Whereas Candea argues that name-avoidance between strangers is more about a social ideal of indirectness than the management of sensitive information (2010: 122), the anonymous introductions that geneticists make between children with rare diseases serve exactly...
the purpose of managing sensitive data. The children remain strangers to each other and geographically separated. Yet, as we shall show, a form of affinity and relatedness is established through practices of anonymity.

Drawing on anthropological studies on anonymity and kinship in transnational adoption (Howell 2003; 2006; Kim 2010) and adoption reunions (Carsten 2000), we unfold how relatedness through anonymity is linked to ‘reflections on personal biography and the completeness or incompleteness of personal histories’ (Carsten 2000: 687). Put differently, the work that geneticists and parents engage in to connect children in the database contributes to personal biography by filling out the blank spaces in the family history. In this creative work, carried out to facilitate diagnosis, the geneticist simultaneously shares the child’s data and shields the child’s personal identity in the database and beyond.

The relatedness we identify has parallels to the biosociality investigated by medical anthropologists and sociologists in the context of the life sciences. In Paul Rabinow’s concept of biosociality (1992), ‘bio’ refers to the shared DNA and illness that becomes grounds for collective identification and action. Since the end of the 1990s, studies on biosociality have documented the myriad ways people make sense of genetic ties and diagnoses (Finkler 2000; Franklin & McKinnon 2001; Gibbon & Novas 2007; Howell 2006; Konrad 2005; Rapp 2000; Sachs 2004) and mobilize political action (Navon 2019; Novas 2007; Petryna 2003; Rose 2007). Our concept of datasociality adds new layers to biosociality. First, with datasociality, we emphasize the fundamentally new ways in which biological information is mediated by digital and algorithmic processes in the genomic era, including intensified possibilities for sharing and pooling data. Second, the sociality experienced among our interlocutors is not linked to the political mobilizations of parents, scientists, and big pharma, and it is not part of a ‘genetic citizenship’ (Heath, Rapp & Taussig 2004: 152). The families we encountered did not look for political alliances with people carrying the same disease. Instead, the clinical work of linking and comparing children carrying de novo mutations by using digital genetic databases and – as we shall see – Facebook communities gives way to new forms of recognition and affinity. With the concept of datasociality, we capture the sociality emerging from data-sharing practices in precision medicine and explore the individual’s experience of comparing her or his own phenotypical, genotypical, and biographical data with those from other individuals.

**De novo mutations, genetic databases, and precision medicine**

The use of gene technology in precision medicine is not new (Reardon 2017; Tutton & Prainsack 2011), yet the falling costs and the accelerated speed of the technology has made it increasingly accessible to healthcare in the Global North. In particular, patients suffering from rare disease who carry genomic variants that lead to disease are expected to benefit from the increased use of genome sequencing technologies. However, many variants are new – de novo mutations – or extremely rare, making clinical interpretation difficult and genotype-phenotype correlations uncertain (Timmermans, Tietbohl & Skaperdas 2017).

From visiting the websites of the two mentioned databases, we learned that GeneMatcher only provides genotype data due to the absence of consent for sharing phenotype data, while DECIPHER offers both genotype and phenotype data based on consent from patients and parents. DECIPHER has data from almost 40,000 patients who have given consent for ‘broad data-sharing’. On their website DECIPHER explains
that ‘anyone can browse publicly-available patient data … and request to be put in contact with the responsible clinician’. The website further informs us that:

Each contributing [medical] centre has a nominated rare disease clinician or clinical geneticist who is responsible for overseeing data entry and membership for their centre. DECIPHER enables a flexible approach to data-sharing. Each centre maintains control of its own patient data (which are password protected within the centre’s own DECIPHER project) until consent is given to share the data with chosen parties in a collaborative group or to allow anonymous genomic and phenotypic data to become freely viewable within Ensembl and other genome browsers. Once data are shared, consortium members are able to gain access to the patient report and contact each other to discuss patients of mutual interest.

From this description, we learn that, on the one hand, data in the DECIPHER database are presented as available and viewable. On the other hand, boundaries are drawn as to how and for whom data are accessible, as only ‘members’ can access patient reports and contact other members for more details about the person.

During fieldwork, the first author observed the geneticists use these genetic databases in preparing genetic counselling. Typing in the result from the microbiologist suggesting a particular *de novo* mutation to be disease causing, the database usually showed no or only a few matches. Asked to explain the work of matching genetic variants in the database, Esther, a geneticist who specializes in rare diseases, said:

When we have the patient’s and family’s consent, we put the patient’s genetic variant [genotype] and symptoms [phenotype] in the database. … I use databases like DECIPHER and GeneMatcher to see if other doctors have reported variants in the same gene as I found in my patient. In the database, we can’t see details about the individual patient, but I can contact the doctor who has reported the patient.

Thus, in order to learn more about the potential likeness or difference between two or more matching mutations in patients, the geneticist has to write a conventional email to the corresponding geneticist who has registered the other patient’s data. This is to ask for a thicker description of the patient’s symptoms, medical history, and maybe even photos. In the following, we shall attend in greater detail to how the geneticists navigate these boundaries and availabilities in daily data-sharing practice.

**Anonymous introductions: ‘Your data … are connected to me’**

In a university hospital in Denmark, geneticist Esther sits in front of a 16-year-old boy, Simon, and his mother. With the family’s consent, the first author is sitting next to Esther, whose genetic counselling work and use of exome sequencing she has followed over the course of a year. Esther informs Simon and his mother about the process of matching Simon’s genetic profile with that of other children. She tells them that she first uses trio-exome sequencing (blood tests from mother, father, and child) to try to identify a genetic change that is suspected to be the cause of Simon’s symptoms. The exome sequencing makes it possible to search nearly all coding regions of the human genome, she says. If she comes to know the genetic change, she will search the literature to see if anyone has described a link between the gene and a disease. If such a link has not yet been made in the literature, Esther explains to Simon and his mother, she will search the international web-based database DECIPHER. ‘In this database, geneticists from all around the world have uploaded thousands of cases of genomic variants’, she says. When a geneticist types in a patient’s genetic variant in the database, she can see if others have found variants in the same gene, and contact the relevant clinicians or researchers. ‘This might help discover new disease genes, and give a diagnosis even...
if the relevant gene is not recognized in the literature as associated with disease’, she continues.

Before Simon’s genetic variation can be shared and compared in the database, his parents must consent to their child being ‘only partly’ anonymous. Esther introduces two different consent forms. The first is for the exome test. Addressing Simon’s mother, she says,

This will … be in an anonymized form where neither you nor your child will be recognized. The second consent form is for the international database DECIPHER. In the database, your son cannot be recognized, but the blood samples [from him and you] can be traced back to me through an identifier: a sample number. In order for the samples to be applicable in the database, another geneticist somewhere else in the world might contact me as the [original] geneticist representing you in the database, and this means that you are not completely anonymous. This is why the consent form says a lot about anonymity; because it is hard to assure full anonymity.

As a final comment Esther adds, ‘Your data [the child’s] reside with me, and are safe with me [in the database]. They are connected to me, and in the database my name is on them, so that the other researchers can contact me, if they need more information about you’. Simon’s mother says, ‘We don’t have any reason to stay anonymous’, and Esther responds by stating, ‘No, but these are also almost American conditions [the consent forms].’

This counselling session brings us into the world of managing personal data and anonymity in transnational genetic databases, to facilitate genetic matchmaking and eventually diagnosis. As Simon enters the database, he is placed in new constellations of belonging. We might say that, as a means to achieve anonymity, the geneticist forges a particular relationship with the child within the realm of the database. Although this relationship is far from the process of adoption, anthropological studies of adoption may assist us in understanding the process of making the child enter the database. In her work on transnational adoption, Signe Howell (2006) refers to the process of bringing family members into relatedness in the realm of adoption as ‘kinning’. She argues that adoption involves a ‘de-kinning’ of the child (Howell 2006: 4), meaning that the child is separated from his or her biological parents. Importantly, de-kinning is enabled both through anonymization of the identity of the biological parents and through physical distancing: the child moves from the country of his or her birth and arrives in a new one. In clinical genetics, children like Simon are not socially stripped or ‘denuded’ (Howell 2006: 4) of meaningful relationships: they are already kinned by nature and nurture to the parents they live with. Nevertheless, in order to achieve anonymity in the database, the doctor needs to stand in as a deputy, namely ‘a person appointed as a substitute with power to act’ (Merriam-Webster dictionary definition) on behalf of the child, in order to access the phenotype before the gaps in the biography of the child can be filled. In other words, connecting children to other children necessitates that the geneticist takes on the role of digital deputy with the aim of protecting the patient’s privacy and anonymity in the database. Whereas kinning in adoption constitutes permanent relations, the relations forged between geneticists and children in the database are situational, temporary, and instrumental.

In her book about diagnosing dysmorphology, Joanna Latimer demonstrates how ‘the family tree and other diagnostic practices … open up the possibility that those who were socially “far” become “near” because they display parts in common’ (2013b: 157). In the diagnostic practices around children carrying de novo mutations, we observed how children who were biologically as well as geographically far became near within the
database and how the geneticists became mediators as they moved the children closer to each other.

Our interpretation of genetic matching as an emergent form of datasociality that resembles a kinning-like practice is inextricably linked to the role of anonymity in data sharing. In the genetic counselling of Simon (above), geneticist Esther expresses an understanding of anonymity as a matter of degree of identification that can be bracketed into parts and wholes (‘not completely anonymous’). Every part added to the whole picture of a child – a name, a photograph of a foot, a face, or a hand, or a disease history, and their combination – will further dissolve or threaten the anonymity of the patient. Stating that the genetic variant of a child can be traced back to the geneticists, Esther explains that it is this link between doctor and child that both enables and compromises the child’s status as anonymous in the database. If the doctor did not stand in for the child in the database, either the child’s name would be directly identifiable or a possible matchmaking would not be able to reach the child. In the database, the geneticist’s representation of the child makes it possible for her to practise ‘anonymous introductions’, avoiding the exchange of names and identification numbers of children and avoiding a breach of confidentiality of each individual patient, to enable more precise diagnosis. We may say that the geneticists’ practices of anonymity – the work of keeping individuals’ identities apart – come to enable new forms of sociality in transnational data sharing.

From the conversation above, we learn that for the parents and clinicians the anonymity is seen as an almost unnecessary bureaucratic formality. By characterizing the consent form for DECIPHER as ‘American conditions’, Esther somehow establishes an alliance between her and the family, demarcating their relationship as different from the doctor-patient relationship in other places in the world. While the parents accept the formalities, they are willing to bypass their rights of privacy if it paves the way for a better and more precise diagnosis of their child. In other words, in the view of the parents and clinicians we met, the benefits of sharing data overshadow the need for shielding persons.

In a counselling session some months later, Esther announces enthusiastically, ‘Listen Simon, we’ve found an explanation for why you’re different. You know, why you find schoolwork difficult and all that …?’ Esther finds a piece of paper, writes ‘DPF2’, and says, ‘Yes, I know that it is a strange name and it doesn’t mean anything and you don’t have to remember the name’. She explains, ‘Six months ago, we still didn’t know what it was [the genetic disorder]’. Esther looks up and asks rhetorically, ‘It could be nice to have an explanation, right …?’ Simon’s mother nods eagerly. Simon smiles shyly. Esther goes on, smiling, ‘Recently, a scientific paper was published that describes mutations in a gene that looks exactly like the mutations in your gene, Simon. The article describes eight [people carrying the same mutation], and you are number nine’. Simon’s mother asks, ‘Nine, in Denmark?’ ‘No, in the whole world’, Esther replies. Placing on the table the scientific paper she is referring to and pointing at some photographs of children in profile, Esther says,

The question is: do you look like [the others] or not? When I first saw you, I noticed your ears [she points to the photograph of another child in the paper]. If I read here what other challenges these children have, it’s about developmental delays, problems in school [mother nods confirming], delayed talking [mother nods], delayed walking.

The mother adds, ‘This is absolutely correct’.
Here geneticist Esther establishes a physical and biographical continuity between, on the one hand, Simon's genetic profile, his facial traits, learning disabilities, and disease history, and, on the other hand, the profiles, traits, disabilities, and histories of eight strangers. Simon's mother recognizes these continuities. The scientific publication together with the database places Simon in extra modes of belonging. Although this act of gaining membership of the community lacks the 'togetherness' with the other database members characteristic of conventional understandings of kinship and relatedness, we suggest that it enables a datasociality. Exploring human-animal sociality, Latimer (2013a) challenges ideas that social relations and social organization are limited to interaction between persons. She introduces a distinction between the state of 'being alongside' and the process of 'being-with' to keep apart moments of relations where the constituent parts are left more temporary and tentative from more sought-out relationships characterized by persons who are connected through 'a sense of togetherness'. In other words, Latimer opposes ideas about sociality confined to relations in which two persons deliberately seek out each other's company in lasting and purposeful relations, sharing both time and space. 'Alongsideness', Latimer suggests, is an intermittent and partial connection.

The concept of 'alongsideness' helps us unpack the qualities of datasociality: a form of relatedness emerging from transnational data sharing in the field of rare disease. In the counselling session above, we might say that Esther is crafting a proximity and making tentative and partial connections between Simon and the eight other children described in the scientific paper, while they remain strangers and geographically divided. This proximity is based on the algorithm of the database bringing together – or holding alongside – the genotype and the phenotype of each of the matching members of the database. Meanwhile, the individual patients are simultaneously kept apart in the name of anonymity; they remain nameless and only remotely reidentifiable through their named doctor. This form of data sharing forges relations that simultaneously preserve connection and division. How the families experience these relations established through the database is what we turn to next.

**Genetic relations without descent**

On his way to Washington, D.C., 11-year-old Piet from Denmark is travelling to meet with Jaxon, a boy carrying the same rare disease as him: Saul-Wilson syndrome. Saul-Wilson syndrome is a rare form of dwarfism associated with a specific genetic de novo mutation, COG4, and with characteristic facial and radiographic features. Piet is famous in Denmark from the television documentary series *The Rare Danes* [*De sjældne danskere*]. Ten years ago when Piet was a baby and first came to the Department of Clinical Genetics for a medical examination, the doctors performed an exome gene analysis but they could not identify which genetic variants were causing his disease. Recently, though, Piet's mother found a baby photograph of a boy (Jaxon) looking exactly like Piet in a Facebook group for undiagnosed children. She made contact with Jaxon's parents and found out that he lived in the United States and was involved in a research project in Washington, D.C. Piet's doctors contacted the American research team, who told them about Jaxon's COG4 mutation. Once they knew the name of the disease gene, the Danish doctors knew where to look in Piet's genome.

In the last episode of *The Rare Danes*, we see Piet in a hospital bed. A doctor shows Piet clinical photographs of the hands and feet of Jaxon. Looking from the photograph to his own hands, Piet raises his hand and spreads his fingers while his face lights up in
fascination, bursting out, ‘It’s unbelievable, I have the same face, the same hands, and the same feet’. He continues, ‘Have you ever heard of anyone looking just like you when you were a baby? It’s pretty awesome’. Next we see Jaxon and his parents standing outside their house in Washington, D.C., ready to welcome Piet and his family. This is a moving scene of strangers hugging as if it were a reunion of relatives after years of separation. We see the boys playing, and Piet says, ‘It’s funny; me and Jaxon look exactly like each other. I feel like I’m looking into a mirror and that I’m reflected by two people (jeg bliver spejlet med to personer)’.

Piet’s physical encounter with Jaxon constitutes an exception from the other children diagnosed with rare diseases whom we encountered in our fieldwork but who never actually met each other face-to-face. Nevertheless, Piet’s experience of strong identification with Jaxon points to the paradoxical meanings of descent and genealogy in the field of rare disease. Whereas most children would have heard of and even seen someone ‘looking just like them when they were a baby’ (typically in a family album), Piet experiences this later in life, with an unrelated stranger, and with the help of Facebook. The relations shaped by genetic matchmaking between Piet and Jaxon exemplify Paul Rabinow’s concept of biosociality (1992), in which biogenetics enforces sociality. In spite of living geographically far apart, belonging to different genealogies and bloodlines, and being separated by anonymity and data-sharing regulations, Piet and Jaxon become connected. Seeing the photos of a stranger makes Piet feel like he is ‘coming home’, as he himself phrases it. While the relations emerging in international genetic databases somehow reaffirm a genetic grounding of kinship (Klotz 2016), what connects Piet to Jaxon, and Simon to the eight other children in the database, is not heredity, but a shared genetic mutation. They share a particular set of geno- and phenotypical traits, which sometimes include a similarity in physical appearance and/or biography that can evoke a strong experience of identification. Unlike studies of kinship and biosociality in the context of genetics (Featherstone, Atkinson, Bharadwaj & Clarke 2006; Gibbon 2009; Klotz 2016), our interlocutors’ search for genetic connections does not aim for social networking but for an experience of affinity. More profoundly, Piet’s experience of affinity with Jaxon raises questions about how anonymity and distance become drivers of relatedness, and how genetics can become a point of departure for growing relations that are not about descent, but nevertheless carry a potential for kinning.

Like Piet, many of the children we met had experienced years of alienation in relation to their biological kin. To be clear, the alienation refers neither to a general experience of lack of love, nor to an ‘alien kinship’ in the sense that parents feel their child is ‘not quite from their family’, as Rayna Rapp found in her work on amniocentesis (Rapp 1999; 2000). Instead, the experience of alienation among the children we met was a sense of feeling different (in appearance and behaviour) from their kin members. Genetic mutations are interchangeably termed ‘genetic alterities’, ‘genetic mistakes’, or ‘genetic variations’ in the clinic, and also by patients and families. All these terms denote that the child is different from, other than, and at variance with the family album and history. This constitutes an important background for understanding the strong identification Piet experiences when seeing photographs of the hands, feet, and face of a stranger, and his experience of being ‘reflected in two people’ when meeting Jaxon.

Considering recent discussions of the limits of conventional thinking on social relations which point to the integral – and not antithetical – role of otherness in social relations (Stasch 2009: 14), we could say that Piet or Simon’s experience of being other in
relation to their genealogical insiders (parents and siblings) enables new social relations rather than rules out existing ones. When Piet experiences affinity with a stranger across the Atlantic Ocean based on a striking similarity in physical appearance and biography, the reverse form of relations of otherness is forged. In Rupert Stasch’s (2009) study of the Korowai people of West Papua, Indonesia, he describes how otherness among certain kin members constitutes social relations. His ethnography is situated within a close community and in the intimate space of a house where, for example, kin members become strangers to one another by avoiding the sharing of plates or food. In contrast, the connections established between children in the database are not based on otherness in physical close relations. Rather, in this case, a relatedness is forged between others who are strangers, yet carrying the same de novo genetic variations. What appears at first hand to connect these others resembles what Rayna Rapp (1999) calls a ‘kinship of affliction’, stressing that what is shared is suffering. However, for Piet, and Simon, what was shared was not a conventional suffering of pain, but hardships caused by physical and cognitive disabilities and the absence of being able to recognize themselves in family members. This was also pivotal in the case of Peter.

Peter is 18 years old and lives with physical and cognitive disabilities. His mother tells us, ‘During the genetic counselling session, Peter was presented with profile pictures of a girl with a variant in the same gene EBF3 as himself. He exclaimed, “Mom, she looks exactly like me, except for the long hair!”’ His mother continued, describing how she herself had reacted to the photograph, thinking, ‘My son has much more in common with this stranger girl, whom we’ve never met, than with his biological siblings’.

The reactions of Peter and his mother illuminate the experience of alienation in relation to his biological family, which becomes the main reason for pursuing a genetic diagnosis and finding out why he is different from his peers and siblings. The girl with whom Peter shares a genetic mutation is simultaneously a complete stranger, a genetic sibling, and not family. Nevertheless, Peter experiences sameness in a way common among kin members who share descent. He also experiences a sense of completeness in the sense that he recognizes himself in another person. Peter’s and his mother’s experience of seeing the girl in the photograph in a scientific publication may also be interpreted as a moment of encountering the face of a relative in a family album.

Looking at photographs of children is generally considered an intimate and personal experience, associated with practices of delving into family albums in private homes. Mary Bouquet suggests that photographs are a means of both establishing and cutting genealogical relations, arguing that ‘kinship came to be significantly constituted through camera lenses during the twentieth century’ (2001: 110). Our ethnography points to the way in which the use of scientific publications and databases in precision medicine crafts a material and embodied sense of belonging among children with rare diseases by making them face anonymous others. In clinical genetics, it is in the practice of recognizing one’s own facial traits and phenotypical particularity in the photograph of a stranger in a scientific publication – not in family albums and histories as in most of Latimer’s cases (2013b) – that relations between children who do not share descent are forged. In filling in the gaps of personal biographies, they become ‘de novo kin.’ The question then becomes: what characterizes this de novo relationship?

While relations in the field of rare disease concern anonymous others, they are always face-to-face or biography-to-biography. In addition to presenting photos of other children carrying the same genetic mutation to the patient and family coming for genetic counselling, the geneticists also read the medical histories of these children
documented in scientific publications or in databases. Yet these interfaces do not necessarily involve physical nearness or intimacy. Piet's meeting with the American boy was exceptional. 'Face-to-face community' takes on a very particular meaning in the context of rare disease in the era of precision medicine and Facebook. When children in these communities sometimes meet others with whom they experience sameness and relatedness across difference and strangeness, these meetings are materially mediated by technologies such as scientific publications or databases. In the case of Peter, Piet, and Simon, relatedness emerges through a double strangeness. Their relation to de novo siblings – that is, to other children carrying mutation in the same gene, sharing their symptoms, and physical appearance – is motivated, simultaneously, by an experience of alienation, or otherness, within the (blood) family and an experience of identification with the other outside the family (genetic mutation).

As opposed to Piet, Simon does not know the eight others carrying de novo genetic mutation DPF2. He has not been in contact with them, they have not met each other, and they live far apart. Thus, the relations we see enabled by the database are not necessarily sought out, nor do they rely on intimate sharing of time, space, or substance. Rather, the patient and family experiences of genetic relations are 'situated moments of relations' (Latimer 2013b) in the sense that identification with a stranger is often bound to the clinical situation – confined to the consultation room – and then left aside when the patient and family leave the clinic to go home. These connections are provisional, contingent, and partial (Strathern 1991). Still, the genetic de novo kin significantly affect the lives of the families we met in ways that we shall delve into in the following.

Mutuality of genetic being

Rabinow's (1992) concept of 'biosociality' has prompted numerous social science scholars to map and investigate the implications of genetic knowledge and technology for how individuals understand themselves or relate to others (Gibbon & Novas 2007; Heath et al. 2004; Pálsson 2007). Many of the parents we met joined groups on Facebook to exchange stories, photographs, and symptoms of their children. While this is illustrative of the social communities and identities emerging around disease (Gibbon & Novas 2007; Rabinow 1992), for the families of children with orphan or undiagnosed diseases, joining Facebook groups was mainly a way to gain a glimpse of their child's future or, as in the case of Piet's mother, to assist in diagnosing the child. Concerned about the unknown future of their children, many of the parents we met asked questions such as, 'How would they manage?'; 'Would they graduate?'; 'Would they be able to get an education?'; 'Would they be able to take care of themselves and leave home?' This concern was evident in an interview with the father of Jenny, a 10-year-old girl:

My daughter has the Sotus syndrome. It's an extra copy of a chromosome. So nothing is missing; rather it is a duplication; in fact a rather significant duplication … I started googling and stumbled over 5Q35 … Then we contacted various Facebook groups to learn from other people's experiences with children with similar syndromes … In the Facebook group, there was a mother of a now-grown-up daughter (in her twenties) with the same chromosomal defects. She had had more than ten different psychiatric diagnoses … It would be really nice to hear what awaits us, and what we can do to help her … to anticipate, to prevent this.

Just like Jenny’s father, many parents expressed ideas that children sharing genetic name codes inevitably also take on each other’s destinies. Janet Carsten (2000) introduces time as central to the experience of kinship in adoption reunions, and in particular the link between a desire aimed at knowing where you come from and the
The establishment of lost continuities between past, present, and future life. The families we encountered in genetics were searching for the unknown future of their child, trying to establish a lost connection to this future through digital connections on Facebook.

In Facebook groups, parents may find children who are older than their own child, which allows them to read about what kind of problems the other children have encountered and what kind of diseases they have developed and at what age, as a means to prepare themselves for what might come. Moreover, for Jenny’s parents, the other children’s life courses also become a stepping-stone for making adjustments – to anticipate, to prevent – in everyday life. Sitting in the family’s home, Jenny’s parents tell the first author that Jenny struggles in school; she is a very slow learner and she has trouble ‘reading’ the social rules in the schoolyard. The parents feel uncertain about what is best for her: should they move her into special education? Reading about the adult woman with psychiatric diagnoses, who – they add – had even tried to commit suicide, Jenny’s parents feel that the best they can do is to help their daughter build self-confidence. Moving her to special education is an attempt to change her future.

In the television documentary series *The Rare Danes*, after ten years of not knowing the future progression of Piet’s disease, we hear the parents of Piet and Jaxon sharing childhood stories of broken bones, infections, hospital stays, and years of uncertainty. Jaxon’s mother says, ‘Before we got the diagnosis we never knew whether a birthday celebration would be his last’. Piet’s parents nod in understanding. From the researchers in Washington, D.C., Jaxon and Piet’s families learn that thirteen individuals carrying COG4 are described in scientific publications. Among these individuals, a 39-year-old man is living a relatively normal life in the United States. This means that from not knowing whether Piet will survive his next birthday, his parents can now imagine him living into adulthood.

This way of enacting alongsideness and establishing temporal continuities, we suggest, adds new meaning to ideas of kinship as a form of ‘mutuality of being’ that may account for the ways in which ‘relatives live each other’s lives and die each other’s deaths’, as Marshall Sahlins puts it (2011: 2). Kinship or relatedness takes the form of well-described reciprocal relations around the world, such as sharing food, work, or housing. Sharing genetic mutations and exchanging personal data on their lives, bodies, and DNA, these children with rare diseases become entangled in a form of reciprocal relation facilitated by geneticists and mediated by databases or Facebook. In the case of Jenny, her father, as with many parents in our study, searches Facebook groups and imagines that his child may come to live the life (i.e. suffering the same ten different psychiatric diagnoses) and die the death (i.e. committing suicide at a young age) of other children sharing the same genetic mutation.

In the case of Piet, his parents lived one year at a time, until they read about a man carrying the same mutation who was then in his thirties, and still going strong. His story helped Piet’s parents connect the present to an open future. Thus, the parents of children with rare genetic diseases may seek out online communities to build imagined communities of mutuality of genetic being. However, this mutuality of being differs markedly from the sharing of time, place, and experience which characterizes Sahlins’s understanding of kinship. As we have shown, the opposite is often true in the case of children sharing genetic mutations. Their relatedness is enabled through a double strangeness: anonymity and otherness. Nevertheless, when parents imagine the biographies of other children to be reflections of their own child’s potential future, we might say they come to read ‘the inscription of multiple others in the one subject’ (Sahlins 2011: 13). This was vividly illustrated in
the clinical counselling session when geneticist Esther showed Simon photographs and read the disease histories of other children out loud.

For the individual entering small groups of ‘patients like me’, group members who are older become ‘foregoers’ or ‘predecessors’ in the sense that they are people who precede the younger member in having carried the disease for more years. Relations forged between predecessors and younger children add yet another temporal component to our analytical understanding of patients sharing de novo mutations as a kind of genetic siblingship, or of having someone alongside them yet at a distance – living their lives at one and the same time separately (physically) and interwoven (imaginatively). Conventionally, such digital connections are considered fluid and evasive compared to relations bearing the qualities of permanence and endurance (Carsten 2000; Weston 1997). We do not wish here to tap into classic misunderstandings that some forms of living or relations are more authentic than others: that is, separating ‘real’ life from ‘digital’ life (Boellstorff 2012). Nevertheless, the relations established in the DECIPHER database – while situational and instrumental – may have a much longer expiration date and may survive the biographical lives of doctors and children.

Evidently, not all families are eager to realize the potential of extra kin. Morten was diagnosed by the age of 27, being one of four documented individuals in the world sharing a rare genetic variation. Morten’s case was unlike Jenny’s: he was the oldest of these four, so his parents could not anticipate their son’s life course by looking into the future of his three younger genetic siblings. Morten had no foregoer of his own, but instead constituted a foregoer of the other children. His mother told us in an interview that to her, listening to the doctor reading out loud the stories of the other three children was unnecessary and even unwanted; ‘I didn’t need to carry the burden of another family, too; I have enough in carrying our own. I came to feel very sorry for the other families, it’s unbearable’. Contrary to how Peter’s parents feel, Morten’s mother considers de novo siblingship unproductive. To her, there is no fulfilment – only a burden – in being involved in the destiny of the other children. Still, she did acknowledge that Morten’s experience may be productive in helping other families, with younger children, to anticipate their child’s trajectory. We may interpret the willingness of not only Morten’s mother, but also the families of Simon, Piet, and Jenny – who all agreed to their case stories and photographs being published in scientific papers – as an acknowledgement of a mutuality of genetic being, binding the destinies of these children together. Moreover, these families’ ability to craft relatedness to unknown strangers is grounded in their relationships to the geneticists with whom they willingly share their children’s data.

Throughout our lives, the process of kinning remains sensitive to continual expansion and shrinking. The de novo kinning we see emerge in the field of rare disease is largely casual, contingent, and imagined. These relations do not take over or substitute for existing kin relations (Klotz 2016). Whereas the long-lasting caring and intimate relationships of sharing home and care over time (nurture) are conventionally seen to solidify a kinship bond (Howell 2006; Mariner 2019), biology and genetic name codes enable de novo relations in the field of rare disease. The kinning we identify here is first and foremost biological, depending on the sharing of an embodied condition and potentially also the well-documented sharing of illness experience. However, this kinning does not depend on the intimate sharing of physical space over time. Instead, temporality here involves the anticipation of coming to live the lives and die the deaths of anonymous others.
Concluding remarks

Precision medicine promises faster and better diagnoses using advanced genome sequencing technologies and millions of biological samples and other personal data to compare the individual patient with the many. However, as our study shows, comparison is not that simple. Evoking well-documented practices of data sharing (Hoeyer 2019; Tutton & Prainsack 2011) dependent on research collaborations (Pinel 2019) and computer algorithms (Ruckenstein & Schüll 2017; Schwennesen 2019), the case of diagnosing rare disease offers new analytical insights into the creative work of anonymity and the new forms of relatedness emerging with digitally mediated data-sharing practices. As our ethnography demonstrates, diagnoses are enabled through algorithms matching children in genetic databases, which make the individual child and their disease-causing gene variant come to the fore against a backdrop of a group of patients sharing mutations as well as pathology and physiology. Importantly, in this diagnostic process, anonymity must be protected and the children kept physically separated to become algorithmically assembled (Schwennesen 2019). As the geneticists perform the seemingly impossible – both sharing data and shielding patient identities – unrelated children become related through qualities of otherness. Geneticists meticulously avoid the use of personal names by offering to enter a deputy-like relation to the children with rare diseases, which involves disclosing their own identity to represent the child in the database. As soon as the child is in the database, algorithms identify and compare children based on impersonal gene names.

In the endeavour to establish new and unacknowledged connections between children sharing the same genetic mutation, geneticists search for hereditary links outside the sphere of biological family. The children who become connected in this process are complete strangers to one another and live their lives separately, scattered across the world, growing up in heterogeneous environments. Yet they are enabled to become de novo kin. The adjectival phrase de novo shares affinity with the notion of step-, as in step-sibling, in the sense that de novo does not substitute for any existing kinship relations, rather it adds on siblings but without carrying the usual moral and social obligations of step-siblings. We do not claim that de novo siblingships take the shape of intimate sharing of time and space. Piet and Jaxon’s encounter was the exception. Nevertheless, independently of whether the families seek out the people with whom their child shares a genetic mutation, they may experience a form of affinity and belonging to an imagined community of others when seeing the photographs of other children’s faces, feet, and hands, or when listening to the biographies of others being read out to them from scientific publications. They may experience a form of ‘mutuality of genetic being’ – becoming aware of other children with the same genetic mutation. Introduced to the other thirteen Saul-Wilson patients and their life stories, Piet’s and Jaxon’s parents could suddenly look into an open future of people carrying the disease who have survived into adulthood, providing them with a new horizon. Yet, as we have shown, this mutuality of being is characterized not so much by a co-presence throughout a lifetime, as Sahlins proposed, through either descent or dwelling; rather, they constitute imaginaries of living the lives and dying the deaths of anonymous others. Meanwhile, within the infrastructure of datasociality, the doctors’ deputy-like relationships to their patients and the alongsideness of children ‘lying side by side’ in the database may survive a parent-child relationship. The possible immortality surrounding the life of data in precision medicine can in some ways make de novo kin appear more permanent than conventional kin.
Acknowledgements

Our first thanks go to the children, families, and geneticists in the genetic department who welcomed us into their life and work. We are grateful to our colleagues in the MeInWe research group for always stimulating discussions and comments on this article. We also thank the participants in the seminar Future Horizons of Medical Humanities at the University of Washington, Seattle, 2018, where the very first ideas of this article were presented. Finally, we would like to thank Janelle Taylor, Lone Grøn, Maya C.F. Jensen and not least our colleagues in the Department of Health Services Research at the University of Copenhagen for their generous feedback on earlier drafts of this article. In Denmark, ethical approval is not mandatory in interview and observation studies. According to Danish law, this study has been approved by the Danish Data Protection Agency. This research was funded by Mette N. Svendsen’s Semper Arden grant from the Carlsberg Foundation.

NOTES

1 https://genematcher.org.
2 https://www.deciphergenomics.org/.
4 https://www.deciphergenomics.org/about/overview (accessed 8 August 2022).
5 All interlocutors’ names are replaced with pseudonyms, except for Piet and Jaxon (introduced in the section ‘Genetic relations without descent’), who are publicly known from the Danish documentary series The Rare Danes. However, we keep the names of the genetic variations of our interlocutors to be in alignment with the ethics of our empirical field.

REFERENCES


——— 2013b. The gene, the clinic and the family: diagnosing dysmorphology, reviving medical dominance. Abingdon, Oxon: Routledge.


——— 2018. The ’we’ in the ’me’: solidarity and health care in the era of personalized medicine. Science, Technology, & Human Values 43, 21-44.


Reardon, J. 2017. The postgenomic condition: ethics, justice, and knowledge after the genome. Chicago: University Press.


Laura E. Navne is a senior researcher at the Danish Centre for Social Science Research and affiliated researcher at the University of Copenhagen in the project MeInWe, investigating the relationship between person and collectivity in the field of precision medicine.

VIVE – The Danish Center for Social Science Research, Herluf Trolles Gade 11, 1052 Copenhagen K, Denmark. LaNa@vive.dk

Mette N. Svendsen is a Professor of Medical Anthropology at the University of Copenhagen and the leader of the MeInWe project. She is the author of Near human: border zones of life, species, and belonging (Rutgers University Press, 2022).

Department of Public Health, Centre for Medical Science and Technology Studies, University of Copenhagen, Øster Farimagsgade 5, 1353 Copenhagen K, Denmark. Mesv@sund.ku.dk

Nouvelles parentés : partage des données, protection des personnes et apparentement dans la médecine de précision

Résumé
Quand un enfant est atteint d’une maladie rare, la comparaison, le partage et la protection de ses données biologiques et biographiques dans des bases de données de génétique et sur Facebook jouent un rôle central dans la recherche du diagnostic par les généticiens et les familles. Sur la base d’un travail de terrain ethnographique au Danemark, les auteurs montrent que le travail de mise en relation d’enfants portant les mêmes mutations génétiques crée entre eux de nouvelles formes d’apparentement qui se rapprochent de la fraternité. Avec ce concept de « socialisation des données », les auteurs ajoutent de nouvelles strates à la «biosocialisation» en identifiant les manières dont, dans le domaine de la génomique, les informations biologiques sont de plus en plus portées par des processus numériques et algorithmiques. Par le processus parallèle de partage des données et de protection de l’anonymat, des enfants non apparentés deviennent parents, du fait de leurs qualités d’altérité.

Laura E. Navne & Mette N. Svendsen

Journal of the Royal Anthropological Institute (N.S.) 28, 1139-1176
© 2022 The Authors. Journal of the Royal Anthropological Institute published by John Wiley & Sons Ltd on behalf of Royal Anthropological Institute.