Genetic Nature/Culture

Anthropology and Science beyond the Two-Culture Divide

EDITED BY

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Chapter 2

Provenance and the Pedigree

Victor McKusick's Fieldwork with the Old Order Amish

M. Susan Lindee

Provenance is defined in the *Oxford English Dictionary* as the record of the "ultimate derivation and passage of an item through its various owners." The term is most commonly used to describe the history or pedigree of a painting—who has owned it, its value at various stages—but it also has a meaning in silviculture, in which it refers explicitly to genetic stock. Provenance, for forestry professionals, is the record of where a seed was taken and of the character of the "mother trees." In this essay I explore provenance in both senses, as a textual record of the origins of a given object (in this case a blood or tissue sample) and as a record of genetic stock. I focus on fieldwork, which creates a record of origins that can certify the authenticity and reliability of a particular pedigree, which then can acquire status as a form of scientific evidence.

In the 1950s and 1960s, human geneticists undertook wide-ranging field studies of human populations around the globe. They tracked visible anomalies, such as Ellis–van Creveld syndrome in the Pennsylvania Amish and albinism in the Hopi of Arizona. They also tracked geographical anomalies, such as the presence in the Pacific Rim of small populations that appeared to be African. Identifying suitable populations, assessing their genetic status, learning their reproductive histories, and extracting from them blood, tissue, and pedigrees were important activities in postwar human genetics.

The medical geneticist Victor McKusick, of Johns Hopkins University, was among the most prominent practitioners of this genetic fieldwork in the 1960s. This essay focuses on McKusick's field practices in the early 1960s with the Old Order Amish in Lancaster County, Pennsylvania. He was tracking a rare form of hereditary disease, Ellis–van Creveld syndrome, a dwarfing condition, and within a few years he had identified as many cases of this syndrome in the Pennsylvania Amish alone as had previously been reported in the entire medical literature (McKusick 1978: 104).

As he continued to work with the Amish, McKusick found many other recessive conditions in this inbred population, and he published many papers on genetic disease in Amish populations. Though I deal here only with his early work with the Lancaster County Amish, and with his efforts to understand one rare genetic disease, this case illuminates more generally the labor involved in genetic fieldwork in this period and, by extension, in the study of genetic disease as it became the focus of scientific and medical interest after 1955. I look at McKusick's methods, his recording systems, and his data collection network, attending particularly to the ways he enrolled different social actors in his project and deployed different kinds of knowledge. McKusick drew on field methods used in anthropology, sociology, and history in order to bring human pedigrees, notoriously complicated social documents, into the laboratory. His labor turned the Amish into a medical and scientific resource. Gossip, X rays, feelings, blood tests, and social consensus were resources for the construction of the pedigree, and any data point might have more than one axis running through it, from notes in Bibles to state public health records to reports from the local undertaker. Knowledge of heredity and disease was craft knowledge, dependent on a wide range of diagnostic and social skills, documents, and practices.

McKusick was acutely sensitive to questions of legitimacy and authority—he was himself a skilled clinician and, therefore, not quite a scientist in the eyes of some of his peers—and he kept scrupulous records of his field activities. I do not claim that he invented the field methods I examine here, but rather that his field methods exemplify the ways in which human geneticists began to remake human genealogy as a scientific resource. The tabulated lists of ancestors had long been suspicious in the eyes of some geneticists, and there had been various calls over the years for human genetics to find a way to go "beyond" the pedigree (see, for example, Haldane 1942). The pedigree was burdened by its transparently social nature, its dependence on the words of the subjects describing their parents or grandparents, and perhaps even by its connection to the project of eugenics and to the questionable data collection practices of the American eugenicist Charles Davenport. Davenport organized dozens of field studies before 1924 of albinos in Massachusetts, of juvenile delinquents in Chicago, and even of the Amish in Pennsylvania, but his workers' field methods were casual and the resulting pedigrees later were considered to be of relatively little scientific value (see Kevles 1985: 55, 199–200). In the molecular era, and with the explosion of new work in human population genetics, physical anthropology, human cytogenetics, cancer genetics, and related fields, the pedigree was being remade into a resource for laboratory science.

As Yoshio Nukaga and Alberto Cambrosio point out in their ethnographic study of the pedigree in contemporary genetic counseling, pedigrees "still constitute the basic investigative tool" in human genetics. The stories people

tell about their families move from a "web of oral narratives to a sequence of visual inscriptions which, in turn, become part of larger inscriptions connecting medical pedigrees to the visual display of, say, cytogenetic or molecular biological test results" (1997). Even the most technical, machine-driven inscriptions of molecular genetics are grounded in the social complexity of the pedigree, which is nature-culture, and which represents a signal case of the employment of cultural resources to achieve erasure—of the cultural.

McKUSICK AND THE AMISH

In the fall of 1962, McKusick, then head of the Division of Medical Genetics at Johns Hopkins School of Medicine, happened to read a profile of a country doctor in Lancaster, Pennsylvania. This doctor, David Krusen, suggested that achondroplasia was frequent among the Amish. McKusick had been working with Marfan's syndrome patients and had an interest in disorders of connective tissue. Because most dwarfing conditions involve a defect in connective tissue, he was interested in achondroplasia in the Amish. He thought, however, that the rates the doctor described seemed much too high, and suspected that the Amish had some other condition.[1]

A few months later, John Hostetler, a Penn State sociology professor, submitted a book proposal to the Johns Hopkins University Press. The book was to focus on the medical, social, and cultural beliefs of the Old Order Amish; McKusick read the proposal for the press and was intrigued. He invited Hostetler to give a talk on the Amish to his research group (see Hostetler 1963, 1963–4). In his invitation, McKusick noted, "We have had occasion to become much interested in blood group and other physical anthropological characteristics of the Old Order Amish in Mifflin County"; the interest derived from "an observation of an unusual type of hereditary disorder which seems to occur with relatively high frequency in this Amish group." McKusick had not yet been out in the field but explained that he was interested in "making some arrangement to get blood samples on a representative group of individuals." He realized that acquiring these blood samples would be facilitated by a knowledge of "family structure, the attitude of the group toward illness and conventional medicine, etc., etc."[2] It is perhaps noteworthy that McKusick here explicitly construed the blood as historical and social, a material embedded in local narratives and Amish culture, and a material whose acquisition would require knowledge not of genetics but of social organization, history, and medical belief.

Just as knowledge of genetics required knowledge of social organization, so social organization and practice could produce biological qualities, bringing genetic disease out into the open through a series of cultural and reproductive choices. McKusick's later list of the qualities that made the Amish good research subjects included "great interest in illness," "clannishness,"

and a high rate of cousin marriage.[3] The cultural produced the biological. The Amish made genetic disease socially visible and easier to track as a result of their acceptance of cousin marriage, their closed breeding population, their meticulous genealogical records (records that had a religious significance but then became scientific resources), and their practice of publishing reports of diseases of all kinds in local newspapers. Indeed, the practices of this population seemed to be almost tailored to the priorities of field research in human genetics. In addition, the Amish, despite their isolation from mainstream life in Pennsylvania, were subject to the standard collection of vital statistics that applied to all residents of the state. Their births and deaths were recorded in Harrisburg. Their death certificates, with the names of attending physicians, were filed in state records, and these records, too, became a part of McKusick's information network.

McKusick's first foray into the field included two local guides: Hostetler, whose scholarly work on Amish culture he found so useful, and who had been born into an Amish family; and Krusen, the country doctor who thought he was seeing a specific genetic disease in the Amish, and who brought the Baltimore physician to meet the families in which it was present. Characteristically, McKusick pulled together various forms of local knowledge and created allies that could help him build an information-gathering network. Finding one's way through the social and cultural system, and through the country roads of Lancaster and Mifflin Counties, required many informants.[4]

Over the next year, McKusick, Hostetler, and a Yale University Ph.D., Janice Egeland, who recently had written her dissertation on the medical sociology of the Amish, conducted a formal survey of five hundred physicians who worked with Amish patients in Pennsylvania, Ohio, Indiana, and Ontario. Their first publication on the Amish as subjects of genetic study appeared in the *Bulletin of the Johns Hopkins Hospital* in 1964. This paper, "Genetic Studies of the Amish: Background and Potentialities," proposed that "the interest of many simple peoples in genealogy is a matter of note."[5] Geneticists, like "simple peoples," were interested in genealogies, of course, and their specific needs intersected with those of their subjects. McKusick, Hostetler, and Egeland stated, "In some primitive people, such as the Navajo Indians, descent, kinship, and clan identification are important in connection with decisions on whom to marry." Such details were therefore accessible and well-known to the populations under study. For the Amish, the model for genealogical record-keeping was the Bible, and "most Amish can trace their complete ancestry back to the immigrants from Europe." One family genealogy, that of the Fisher family, contained data on thirty-six hundred families and had some relation to almost all living Amish in Lancaster County.

As subjects of genetic studies, the Amish were a closed, defined popula-

tion, the authors pointed out. No one could join, and there could be "no question of who is presently Amish." The Amish were also producing large families. Seven to nine children were common, and parents did not stop having children after the birth of an ill or abnormal child. Furthermore, mentally retarded or disabled children were kept at home, which meant that they could be readily studied "in relation to the rest of the family." Populations with similar characteristics had been studied in Switzerland, Sweden, and other areas of Europe, but the authors pointed out that the most informative such population in the United States was the Utah Mormons, who, like the Amish, were genealogically inclined, relatively immobile, clannish, and closed to outsiders (McKusick et al. 1964).

An entire range of cultural and religious choices made such isolates scientific resources. But these choices could also interfere with the fieldwork. The difficulties in "realizing the full potential of the Amish for genetic studies" related to Amish suspicion of outsiders, and reactions to some aspects of medical science. Amish families, for example, were in general reluctant to agree to autopsy. Of the thirty-six deceased persons with Ellis–van Creveld syndrome reported in a separate paper in the same issue of the journal, only one was autopsied (McKusick et al. 1964).

McKusick and his coauthors reprinted a letter from a thirty-year-old Amish man who had Ellis–van Creveld syndrome and who refused to be examined by the Hopkins researchers. The man stated, "I feel I am exactly the way the Good Lord intended for me to be, even before I was born. So I feel no human hands or brains can do a thing about me or anyone like me, if it is the way the Lord wants it, no matter how highly educated anyone is. I am happy, have work, friends and can support myself. So what more do such people want?" (McKusick et al. 1964). Another prospective participant in the study sent a postcard declining: "I am not interested in going in the hospital, so don't come around for me because I am not going in. And you don't have to stop by to see me either. I am allright[,] there isn't anything wrong with me and I don't think much of those x-rays you want. So don't stop in to see me. I am not interested in your stopping by."[6] In both cases respondents were contesting their status as objects of medical interest. The first was satisfied that his condition was God's will; the second that there was nothing wrong with him. They would not participate in the medical research, nor in the construction of Ellis–van Creveld syndrome—of their short stature and extra fingers and toes—as a genetic disease, and they were resistant to the technologies that McKusick's work would require.

Over the next two years, McKusick built a sieve that could lift a specific, visible form of genetic disease out of a social network. He was trying to find all cases of short stature and extra fingers. He used public records, Amish genealogy books, birth reports, newspapers, health professionals, and Amish contacts. He began to subscribe to *The Budget,* the Lancaster County news-

paper that published reports about why someone missed church (a knee injury), tonsillectomies, birthday parties, and bad backs. He kept in touch with the undertaker who handled most Amish deaths. He surveyed the records of hospitals. He wrote to school officials to ask them to excuse students who missed school to be examined in his clinic.[7] He wrote personal letters to teachers, nurses, parents, and physicians. He was looking for extra fingers (polydactyly), which were signs that left a record in the state capital in Harrisburg years after a neonatal death in Lancaster, or that could be remembered years later by a midwife, a grandmother, a sibling. The disease that ended a life of only a few hours could be seen despite the poor resolution of the record and the temporal and cultural distance from a midwife on an Amish farm to the head of the Division of Clinical Genetics at the Johns Hopkins University years later. Every baby mattered, including those who lived only twenty minutes. "Did this baby have extra fingers?" McKusick asked a nurse present at a birth in September 1969.[8] The entire population needed to pass through the sieve. (See figure 2.1.)

Records of field trips from Baltimore to Lancaster and Mifflin Counties in McKusick's papers suggest the many kinds of information McKusick brought back. His notes describe families returning from funerals, conversations at the vegetable market, a dog bite he endured at one home, a frightened young child who "sobbed throughout our time there." He visited one family to learn that "they were no longer Old Order Amish" and another in which a teenager with Ellis–van Creveld was a "very fine boy," a junior in high school who had a job at a hardware store. "All the children work hard on the farm taking care of 27 milk cows," noted McKusick. Another boy in this family had been run over by a "wagonful of stones" but now was doing all right: "He sings a great deal and takes voice lessons."[9] On another field trip, the group "stopped by Kaufman's Orchard to get some apples and other things" and ran into one of the families they were going to visit. They also stopped by the home of a "very attractive young couple" who were "prepared for having church at their house the following day. They expected 70 or 80 people. She had made 32 pies the day before!" When the group visited an Amish school they learned that there recently had been a "discipline problem" at the school, where some of the boys had refused to wear their hats on the playground. At another home they found a blind father, a daughter with serious medical problems, and a "competent" mother: "She has to run a farm and raise a family with a blind husband." The group ended up "having a family style meal at the Harvest Drive Restaurant."[10] I mention these details to capture something of the tone and feel of this fieldwork. McKusick and his assistants were collecting many kinds of information about Amish culture and Amish people and made many kinds of observations in these field notes.

Amish participation in McKusick's study seems to have been enthusiastic. While there were a few resistant reactions, most Amish queried were willing

Figure 2.1 The "Amish Madonna." This was the second child in sibship 14 affected by polydactyly. Courtesy of Dr. Robert Weilbaecher.

to participate, and some served as informants and field-workers for his project. In one of the early papers, the authors hint at female skills in the management of awkward social approaches, noting, "Several unmarried Amish women collected information both of medical and genealogic nature and provided introductions to affected families who in many instances were relatives" (McKusick et al. 1964). Amish provided McKusick with clues, leads, and suggestions for tracking other cases. They wrote to McKusick for advice

about whether to marry distant cousins. They spontaneously reported abnormal births in their extended families, and many cases came to McKusick's attention through the Amish. His families were organized in sibships, groups composed of all the offspring of a particular pair of parents. The "mother in sibship 23" reported two other cases that became sibships 25 and 26; the "father in sibship 3" was the "informant" for a case in sibship 10. McKusick was recording exactly who told him what.[11]

Parents provided him, moreover, with descriptions of their children's bodies, recalling the morphologies of stillbirths and neonatal deaths years after the fact, so that those pathological forms could become a part of the pedigree. "Mother states no extra fingers. Four children living and well," he recorded in one case of a four-week-old infant who died; or "Mother states extra fingers were present but apparently not of type in EvC" in the case of a thirty-three-year-old daughter who died in 1959, before McKusick began his study.[12] Maternal descriptions could rule out Ellis–van Creveld syndrome or certify its presence. Amish family members could also lead McKusick to other communities, as did one Pennsylvania father who said his siblings in Ohio had children with the same condition that affected his own. This father theorized that the condition was hereditary. "Now this may sound strange," he wrote to McKusick, "but five years ago my sister had a baby girl with the same trouble as ours. And a month or so later my brother's wife gave birth to a baby girl also with this same thing. Could it be hereditary?"[13] There was what might be called a folk epidemiology in the community itself, a network of knowledge and interpretation that could help McKusick identify relevant families and relevant bodily forms.

The Amish genealogy books encoded this folk epidemiology, and McKusick used them to construct pedigrees. In the fall of 1963, for example, he encountered an Amish family in which there were three adult siblings with polydactyly and achondroplasia, Ellis–van Creveld syndrome. He found in reading the genealogy book of the family that there were two other siblings in the family who died young, one as an infant and one as a teenager. Did these children have the same traits? He made his inquiry to the family physician, but the family physician asked the father and the father reported that the infant probably did, for it was a "short, chunky little baby," but the teenager certainly did not and had died of pneumonia.[14] A genealogical text, prepared for religious reasons, helped make the family a scientific resource, and a physician consulted for his specialized knowledge simply asked the father, who diagnosed both infant and teenager. There were many kinds of knowledge in this reconstructed pedigree.

Local physicians were of course an important resource, and tracking down a new case usually began with an appeal to the attending physician. In September 1963, for example, McKusick queried a Pennsylvania physician about

a child who had died at Lancaster General Hospital in the spring of 1962, and who reportedly was born with signs of achondroplasia and heart problems. McKusick had probably learned about this death from the Amish newspaper, which reported the details of neonatal deaths, including the presence of abnormalities. He asked the attending physician questions about the size of the family, the presence of extra fingers, and the health of the parents, noting, "I have been much interested in the last year in hereditary disorders among the Amish and have been making a particular study of dwarfism. I would appreciate any information you can give me on Amish dwarfs." He enclosed a list of cases and families he already knew of and asked the physician if he knew any others, closing with a proposal that he would drop by the physician's office on his next field trip. This particular physician cheerfully answered all the questions and invited McKusick to stop by his office.[15]

As McKusick's database grew, he recorded where and how he had learned of the existence and status of each diseased person. The provenance of any given case included how it was ascertained initially, on what basis Ellis–van Creveld was diagnosed, what the health status of the affected individuals was, and how the pedigree was constructed in relationship to other pedigrees such as the Fisher genealogy. McKusick recognized fully the importance of this documentation, and he even included it as an appendix to his 1978 paper "Dwarfism in the Amish," stating that the "frequency of EvC as determined in this study is so unusually high[,] and such a large proportion of the cases had died before the study was performed[,] that it is deemed essential to outline briefly the features of each sibship, and to indicate the mode of ascertainment and basis for diagnosis in each case" (McKusick 1978: 119). The appendix listed twenty-nine sibships, and a typical listing included a description of the affected child and some indication of the reliability of the information ("polydactyly is . . . absolutely certain in the minds of the Amish informants" [McKusick 1978: 121]). In sibship 11, the affected family member was reported to "work hard with horses on farm" (suggesting perhaps that he was relatively healthy), and in sibship 3 there was a strange coincidence that led to ascertainment, a coincidence reported in the appendix. For most of his cases, and certainly for his most important pedigrees, he had multiple sources, and these were all recorded. A case might be documented in birth and death certificates in Harrisburg, a family Bible, hospital records, phone calls from a physician, letters from family members, personal observation on a particular field trip, reports from a descendant, or by one of his field-workers. As McKusick followed these signs, he collapsed distinctions between sources of information, accepting as equivalent the reports from midwives about recent births, from mothers about infants born a decade earlier, from physicians and nurses, and from a great-grandson reporting on the

health and stature of a long-dead great-grandfather. The pedigree was like a patchwork quilt, pulled together from multiple fabrics into a pattern that rationalized the heterogeneity of the sources.

His papers from this period contain hand-sketched maps and directions telling field-workers where particular houses were and where families lived. At the same time, field-workers were mapping relationships between families on similarly scribbled pieces of paper, which were also tucked away in archived files. Both forms of maps—those depicting roads and landmarks, general stores and silos and red barns, across many miles; and those using darkened and clear circles and squares to depict complex familial lines across generations—were ways of organizing the Amish.

In Edward R. Tufte's explorations of envisioning information, he distinguishes between pictures of nouns and pictures of verbs. Maps and aerial displays, he says, "consist of a great many nouns lying on the ground," while pictures of verbs involve "the representation of mechanism and motion, of process and dynamic, of cause and effect." The directional maps of Lancaster County were "nouns lying on the ground," while McKusick's tentative pedigrees, hand-sketched and tucked in folders and notebooks as the work progressed, were pictures of verbs, arguments about cause and effect, and stories about history and heredity. They depicted a flow chart that made its case using the "smallest effective difference" between diseased and not diseased, male and female, alive and dead (Tufte 1997: 73–78, 121–27). The genetic pedigree, a standardized genre by the 1960s with rules about circles, squares, shading, and arrangement, is a record of field labor. In this case the labor engaged an entire community. Both the Amish and McKusick were proficient collectors of genetic disease.

Despite their participation in his fieldwork, McKusick did not particularly want the Amish to see the scientific papers he published. When he sent a reprint in 1965 to a physician in Strasbourg, Pennsylvania, who was treating one of the Ellis–van Creveld patients, he noted, "I of course do not want it to get into the hands of our Amish neighbors."[16] He was clearly assuming that the Amish would not read the scientific journal in which the paper was published. The social and intellectual gulf between his own world and that of the Amish seemed large enough to prevent any chance encounter between his subjects and his published work. It may be that he was concerned about the photographs, which featured recognizable Amish people, both adults and children, who had Ellis–van Creveld syndrome. Their names were not included in the text, but they presumably would have been known in the community. But I wonder too if he was concerned about his translation of their own genealogies, from religious texts to secular texts, from records of relation to records of pathology.

Another individual case was pictured in one of the early papers, in the form of a reproduction of a seventeenth-century Dutch drawing of an ele-

gantly posed human skeleton (see figure 2.2). The skeleton was that of a newborn infant, drawn standing, skull tipped quizzically, arms slightly raised, in a formal posture that would have been impossible for the living child. "Thrown into the river at birth," according to the accompanying text, the newborn had been retrieved for its scientific interest. If not the focus of maternal love, it could at least be the focus of the love and desire that informed natural philosophy. It had seven digits on each hand, eight and nine digits on its feet, and some had theorized that it was an Ellis–van Creveld case. The Amish, with all their historical specificity, could be linked to a seventeenth-century Dutch newborn. Their disease bound them to a distant place and time.[17] The Dutch newborn was perhaps a record of the provenance of Ellis–van Creveld syndrome, a record of the "derivation and passage of an item through its various owners." The gene moved through human populations, leaving traces, signs, clues in the standard systems of accounting for people and recording their medical status. The pedigree highlighted these clues, brought them together, and situated them in a narrative that could make them scientific resources.

THE NATURE OF THE PEDIGREE

Finding any gene requires extracting words and blood from people, convincing them to contribute some portion of their bodies and some portion of their personal histories to science. The blood and the narrative are embedded in a larger narrative, a pedigree documenting family history, a causal model documenting the nature of a genetic defect based on inheritance patterns revealed in the pedigree, an origin story about the source of the mutation based on its population distribution, or a map upon which a particular disease can be placed in relation to all other genetic diseases. The potential inscriptions of fieldwork are multiple, complex, cumulative. But the basic inscription, the first point of translation, is the pedigree, and producing a pedigree is unquestionably social work.

Recent work in the history and sociology of science has explored the properties of the field sciences with special attention to the "chronic issues of status and credibility that derive from the social and methodological tension between laboratory and field standards of evidence and reasoning." Fieldwork involves phenomena that are "multivariate, historically produced, often fleeting and dauntingly complex and uncontrollable," as Henrika Kuklick and Robert Kohler have noted, and the field seems to be almost unsuited to the production of scientific knowledge. "It may seem astonishing that any robust knowledge comes out of fieldwork. Yet it does, abundantly and regularly." They also point out that in the field, unlike in the private space of the laboratory, scientific work is shaped by the social interactions of profession-

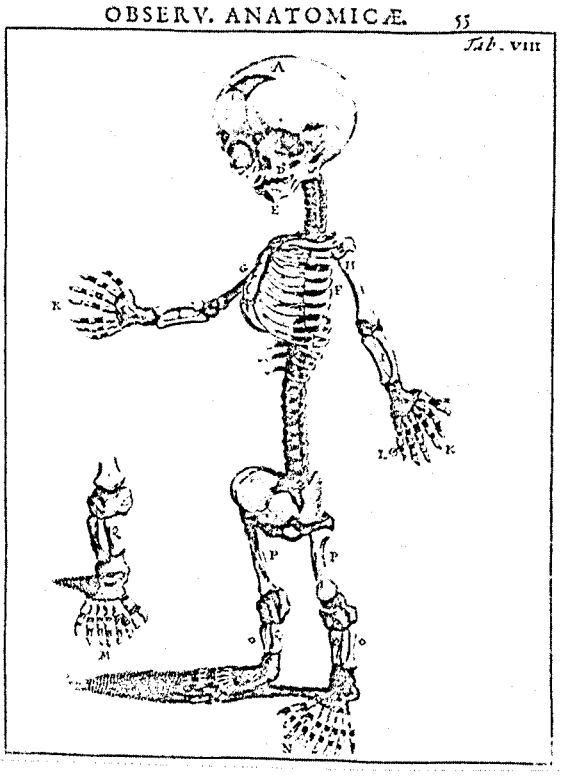

Figure 2.2 A polydactylous dwarf. Pictured in a 1670 Dutch publication, it was identified in 1940 as a possible Ellis–Van Creveld case.

als, amateurs, and local residents whose cooperation is both necessary and rarely acknowledged (Kuklick and Kohler 1996).

In a scribbled, undated set of notes in McKusick's papers, he outlined his concerns about pedigree methods. First, he said, genetic disease involves a "dynamic process": "At the time you study them you can't give the last word." The disease might develop as a person ages, and human subjects, McKusick pointed out, live as long as geneticists do. They were therefore outside the normal time frame for scientific study. Second, he said, there was a "lack of specificity" in clinical manifestations. The trait was "far from the gene." Reading down to the hereditary material from the clinical sign was difficult, complicated, and sometimes not possible. McKusick fully appreciated the complexity of moving from the body to the genotype. Finally, there was the question of familiar seeing. He knew himself that the more you looked at

anyone the more you saw. The clinician in a standard examination might simply miss something that was relevant to understanding the disease and, therefore, the pedigree: "The more [an] individual personally studied and studied from a specific point of view, the greater the reliability."[18]

Yet the pedigree, despite its difficulties, was the "cornerstone" of medical genetics.[19] The preparation of pedigrees and the listing of new genetic diseases did not amount to mere "stamp collecting," he argued. "The catalogs of simply inherited genetic traits in man are like photographic negatives from which a positive picture of the normal genetic constitution can be constructed" (McKusick et al. 1964). McKusick suggested that cataloguing genetic traits, birth defects, and diseases in human populations could transform medical practice and patient care. This idea—I have called it the cataloguing imperative—has seen its most recent formulation in the promotion of the Human Genome Project; but it predates the project itself, and it is possible to argue that the project is a consequence of this expectation, which was so important to the rise of a scientifically legitimate human genetics. The listing of traits (diseases, chromosomal anomalies, birth defects) became central to the promotion of medical genetics. The catalogue seemed to be a crucial resource, a database that could demonstrate the relevance of genetics to medical practice.

In a 1977 grant proposal, McKusick said his project would permit him to document "new recessive disorders among the Old Order Amish." He could also gain insights into "incompletely characterized Mendelian entities" by studying this population. There were at least sixty cases of cartilage-hair hypoplasia, for example, and there were some new reported cases of chondrodysplasia with severe combined immune deficiency. There were also a "presumably autosomal recessive form of osteogenesis imperfecta," many cases of cleft hand and foot, and six sibships with Kaufman syndrome.[20] The Amish were a rich treasure trove of genetic disease, and McKusick's field trips provided access to these diseases, which were called to his attention in casual conversations at the market and in trips to the local school. The fragments of detail—a child born dead, a sister who was mentally retarded—became part of grant proposals. The "medical tourist" brought back enough information to secure funding for another trip.[21]

A grant reviewer, when considering this proposal for the National Institutes of Health, wrote that "the type of research is more descriptive than innovative, but an enormous amount of data which makes a significant contribution to clinical genetics results from astute clinical observation and intelligent interpretation of the results. It is in this field that Dr. McKusick is a world leader."[22] As this assessment suggests, McKusick's own relationship to both clinical medicine and high science mirrored the complicated interplay of clinic and laboratory in the rise of medical genetics. McKusick occupied a critical borderland between science and medicine. When he was nom-

inated for membership in the National Academy of Sciences, one skeptic asked, "But is he a scientist?"[23] And was human genetics a science?

In the 1970s, McKusick kept a dwarfed miniature poodle, named Vanilla, as a pet. In March 1973 Vanilla was bred back to her father, Can.Ch.Andeches Ready to Go. The pregnancy was expected to be risky, with a potential need for a cesarean section, and McKusick made provisions for Vanilla to be kept and closely watched at the Division of Laboratory Animal Medicine at the Johns Hopkins University School of Medicine, under the care of Edward C. Melby, professor and director of this division, during the final stages of her pregnancy.[24] I could not find any documentation in McKusick's papers describing the outcome of this high-risk pregnancy. I mention the pregnant poodle because she was an animal model—and a companion animal—manifesting a condition related to those McKusick was studying in human populations. McKusick could control the breeding and pregnancy of Vanilla and could produce, through inbreeding, a biological state that mimicked Ellis–van Creveld. He was an overseer of dwarfing conditions, a manager of bodies and genomes and catalogues, and a perceptive student of the pedigree, both canine and human, in all its biosocial complexity.

Like other human geneticists of his generation, he was struggling to find a way to study human populations and human genetic traits that mitigated the chaotic conditions of the (human) field. Genetic fieldwork was the study of phenomena that were deeply disordered and uncontrollable. Managing this disorder required a keen attention to provenance, to the histories of people, blood, families, and cultures as they intersected in a single phenotype. It required following a malformation of the limbs and an abundance of fingers through the many texts, stories, and memories where they were documented, and translating these signs into an authentic record of heredity that could presume to hold its own in the laboratory. Creating the impeccable pedigree was one of the great achievements of postwar human genetics. Making its contingency functionally invisible—or irrelevant to its usefulness as a guide to the genome—was a social project of enormous complexity, as McKusick's fieldwork makes clear.

NOTES

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ongoing dialogues, and for critical comments and suggestions, I am grateful to Dorothy Nelkin, Rayna Rapp, Barton Childs, Charles Rosenberg, and Henrika Kuklick and to my coeditors Alan Goodman and Deborah Heath. Following the practices of the sociologist or anthropologist, I do not use the names of Amish subjects here because the records I have explored are basically confidential medical records, but I recognize that constructing McKusick, physicians, and other researchers as the only named people in this narrative represents a form of asymmetry that I would prefer to avoid.

1. "Since true achondroplasia is a dominant with markedly reduced reproductive fitness, one would not expect it to have a high frequency in an inbred reproductive group." McKusick, "Medical Genetic Studies of the Amish," grant application to National Foundation–March of Dimes, for 1979–80, Box R109C16, "National Foundation 1979–80," Papers of VAM.

2. McKusick's invitation to Hostetler to speak is in McKusick to Hostetler, 7 May 1963, Box R115F1–2, "The Amish Population," Papers of VAM. He offered Hostetler a fifty-dollar honorarium, and as was often his custom with invited speakers, an invitation to stay at McKusick's home. See McKusick to Hostetler, 7 May 1963.

3. McKusick discusses these advantages in many sources both published and unpublished, but a specific list, "Characteristics of Amish Society Favorable for Genetic Studies" is in Ellis–van Creveld 8005, Box R115 F1–2, Papers of VAM.

4. This first trip is discussed in David Brown, interview with Victor McKusick, p. 35, Papers of VAM.

5. McKusick, Hostetler, and Egeland 1964, reprinted in McKusick 1978.

6. January 1964, postcard to Victor McKusick, Ellis–van Creveld 8005, Box R115F1–2, Papers of VAM.

7. 13 September 1966, Ellis–van Creveld Syndrome File, Box R115F1–2, Papers of VAM.

8. Letter in Ellis–van Creveld 8005, Box R115F1–2, Papers of VAM.

9. Andrew Gale, E. A. Murphy, V. W. McKusick, and V. A. McKusick, "Trip to Lancaster County," 17 February 1975, Lancaster File, Box R110D8, Papers of VAM.

10. Notes on a trip to Lancaster with Clair Francomano and Guadalupe Gonzalez-Rivera (known as Lupita), 4/6/82, Red Binder, Box R109I18, Papers of VAM.

11. Twenty-one pages of handwritten notes describing these sibships and this informant network, probably written in July or August of 1963, are found in Ellis–van Creveld 8005, Box R115F1–2, Papers of VAM.

12. Elis–van Creveld, Box R115F1–2, Papers of VAM.

13. Letter in Amish, Box R110D8, Papers of VAM.

14. McKusick to Robert Baur, 26 November 1963, with attached handwritten notes in response, in P8005, Ellis–van Creveld, Box R115F1–2, Papers of VAM. Other correspondence with physicians who treated the Amish is also in this file.

15. McKusick to Clinton Lawrence, 12 September 1963, Ellis–van Creveld 8005, Box R115F1–2, Papers of VAM.

16. McKusick to Henry S. Wentz, 10 March 1965, Ellis–van Creveld 8005, Box R115F1–2, Papers of VAM.

17. This illustration appeared originally in Theodor Kerckring, 1670, *Spicilegium anatomicum* (Amsterdam: n.p.). It was reprinted in McKusick 1978.

18. McKusick (his handwriting), "Pedigree methods," in the Pedigree Method in Medical Genetics file, Box R109C8, Papers of VAM.

19. McKusick used this word in a grant application in 1957 to the National Institutes of Health for support for a study that could "define, in as much detail as possible, the mode of inheritance, mechanisms of clinical manifestations . . . [and] factors affecting penetrance and expressivity" in cardiovascular disease. The pedigree method, he said, would be "the cornerstone of the program," and the "machinery" for ascertainment of relevant pedigrees was already in place in Baltimore. He would need a nurse for "pedigree tracing, checking hospital records, assistance in clinical testing," part-time secretarial help, and a technician for chemical tests. He also asked for $150 for the drawing of pedigree charts. Application for research grant: "A study of the genetic factor in cardiovascular diseases," 20 February 1957 (not in folder), Box R109C8, Papers of VAM.

20. See Amish Grant, Box R110D8, Papers of VAM.

21. This term was used by a physician currently treating the Amish for genetic diseases to refer to medical researchers of an earlier era who were "less interested in the health-care needs of the community than they were in the diseases themselves." This is presumably a reference to McKusick and his field-workers. D. Holmes Morton, quoted in Stranahan 1997.

22. The summary referee report is attached to a letter. Paul A. Deming of the National Institute of General Medical Sciences to McKusick, 28 December 1977, for application 1RO1 GM 24757–01, filed with grant proposal, Amish Grant, Box R110D8, Papers of VAM.

23. McKusick was elected to the National Academy of Sciences (NAS) in 1973. See the large file on his nomination and admission, NAS, Box R109E4, Papers of VAM.

24. McKusick to Edward C. Melby, 6 April 1973; and Melby to McKusick, 7 March 1973. See correspondence and related materials in Folder M, Box R109I9, Papers of VAM.

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