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Chromosome surveys of human populations: Between epidemiology and anthropology

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ABSTRACT

It is commonly held that after 1945 human genetics turned medical and focussed on the individual rather than on the study of human populations that had become discredited. However, a closer look at the research practices at the time quickly reveals that human population studies, using old and new tools, prospered in this period. The essay focuses on the rise of chromosome analysis as a new tool for the study of human populations. It reviews a broad array of population studies ranging from newborn screening programmes to studies of isolated or 'primitive' people. Throughout, it highlights the continuing role of concerns and opportunities raised by the propagation of atomic energy for civilian and military uses, the collection of large data bases and computers, and the role of international organisations like the World Health Organisation and the International Biological Programme in shaping research agendas and carving out a space for human heredity in the postwar era.

It is often claimed that after 1945 human geneticists—in an attempt to disentangle their science from racial thinking and its eugenic past—turned to medical questions and focussed on the individual rather than on the study of human populations (Kevles, 1995, pp. vii–xi; Reed, 1974). This account is currently being revised in at least two ways. Firstly, new scholarship indicates that eugenic aims were compatible with, and indeed dependent on, clinical objectives and that this entanglement continued in postwar medical genetics (Braslow, 1996; Comfort, 2012).[1] Secondly, taking a broader look at the study of human heredity, it becomes apparent that population studies, using old and new tools, prospered in the postwar era. This indicates that the postwar history of human heredity and the vexed question of the continuities with the eugenic and racial theories of the past are more multi-layered and complex than sometimes suggested. It also encourages us to take a closer look at the human population studies of the postwar era.

The principles of population genetics were worked out in the 1920s and 1930s in an attempt to combine the principles of Mendelian genetics with Darwin's theory of natural selection. Studies of blood group distributions in human populations and gene linkage studies provided fruitful approaches for the exploration of human evolution. The respective role of genes and environment in relation to mental diseases provided another area of study in human population genetics (Kevles, 1995, pp. 193–211). The results were used to support as well as to criticise eugenic policies. Human population studies continued in the postwar era but some questions shifted, with concerns produced by the nuclear age moving centre stage.

A central question debated by the geneticists Hermann Muller and Theodosius Dobzansky in the 1950s and 1960s concerned the evolutionary role of allelic variation produced by mutation. While Muller argued for the deleterious effect of an increased mutation rate, Dobzansky defended the position that mutation can increase the fitness of populations. In his investigation of the debate, also known as the classical/balance controversy, historian of biology John Beatty has argued that the course of the controversy cannot be

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[1] Largely supporting this claim, Diane Paul has argued that in the context of genetic counselling, the shift from a focus on the gene pool of the population to a concern with the individual only occurred in the 1970s in response to a sharpening of the notion of reproductive autonomy and a move to non-directive counselling in this period. According to Paul, links to eugenic concerns persist even beyond this shift in current practices of medical genetics and reproductive medicine (D. Paul, 1998).

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adequately understood without taking into account the policy considerations regarding the permissible level of radiation exposure with which the debate on the mechanism of evolution was deeply intertwined. Beatty here pointed to the close link of evolutionary debates, human population studies and concerns about the mutational effect of radiation in the postwar era (Beatty, 1987).

Turning Beatty's argument around, I would like to propose that population thinking framed the permissible dose discussion (as well as Muller's notion of the 'genetic load' produced by atomic fallout) and with it human genetics at the time.[2] With mutations being rare events, the mutational effect of radiation could only be established through population studies. Large scale experiments were set up with fruit flies and mice. However, experiments with the two animal populations produced different results and the question remained whether the effects were the same in humans. Data on human populations were provided by studies of the atomic bomb survivors in Japan and by follow-up studies of large patient populations exposed to radiation treatment.[3] In turn, recommendations for permissible doses were established taking into consideration the size of the exposed population. Nowhere does this become clearer than in the recommendation, adopted by various protection agencies, that the permissible dose of radiation exposure for the general population should be 1/10 of that for workers in nuclear establishments. This recommendation was based on the reasoning that the size of the general population would generate a larger number of affected individuals and would therefore represent a more serious public health problem. The fact that the general population was more difficult to monitor bolstered the case.[4]

This essay will further explore the connections between radiation, human population studies and genetics and their broader impact on postwar human genetics by focussing on the rise of chromosome analysis as a new tool for the study of human populations. Human chromosomes were studied from the 1910s. But human chromosome analysis only came into its own in the 1950s with the refining of the protocols for chromosome preparations in the context of efforts to visualise mutations in the atomic age (de Chadarevian, 2010). From there, human chromosome research developed in many different directions. Again, if we think of human chromosome analysis and where it matters today, we will probably think of pre-natal or clinical diagnosis. Yet in the 1950s–1970s human chromosomes played a prominent role in a broad range of projects, including such diverse fields as radiation biology, cancer research, toxicology, epidemiology, paediatrics, sex research, research into ageing, anthropology and evolution. Population studies found a place in several of these fields. We can usefully distinguish two different research directions. An epidemiological direction was aimed at correlating changes in chromosome structures with various factors, ranging from disease symptoms to the effect of mutagens. The other orientation we can describe in general terms as anthropological. It was aimed at studying the genetic structure or 'racial variation' of human populations.[5]

Chromosome analysis or karyotyping was not necessarily the most successful tool in human population research. The research projects articulated around the new tool none the less provide a useful point of entry to study the intersection of genetics and human population research in the post-World War II era. In focussing on the population studies undertaken with the new karyotyping techniques, I will highlight the continuing role of concerns and opportunities around nuclear radiation, the collection of large data bases, and the role of international organisations like the World Health Organisation (WHO) in shaping research agendas and carving out a space for human heredity in the postwar era. The first part of the essay will focus on the population studies undertaken in Edinburgh under the direction of the medical researcher and epidemiologist Michael Court Brown, who is credited with having founded the field of population cytogenetics. The second part looks at the use of karyotyping technologies for studies of human variation in world-wide population studies that were undertaken by the WHO and as part of the Human Adaptability project in the International Biological Programme (IBP) from the late 1950s to the mid-1970s.

1. Population cytogenetics

In the late 1950s and early 1960s only a handful of centres around the world had the necessary skills and equipment for human karyotyping. Among these the institution that most vigorously pursued the project to use karyotyping techniques for large-scale epidemiological studies was the Medical Research Council Clinical Effects of Radiation Research Unit at the Western General Hospital in Edinburgh, headed by the radiologist and medical researcher Michael Court Brown. The unit soon changed its name to MRC Clinical and Population Cytogenetics Unit to better describe the actual work of its researchers.

Court Brown made his name with a study that established that patients that had undergone radiation therapy for the treatment of ankylosing spondylitis, an arthritic condition, showed a tenfold increase in the incidence of leukaemia cases. The massive investigation that followed up over 14,000 patients who had received X-ray treatment between 1940 and 1954 in over 80 radiotherapy centres throughout the United Kingdom was a classical epidemiological study. The epidemiologist Richard Doll, who was already well known for his work on the link between smoking and lung cancer, collaborated on the statistical analysis of the data. A preliminary report appeared in the Government White Paper *Hazards to Man of Nuclear and Allied Radiations*. It was followed by a full report in the following year (Court Brown & Doll, 1956, 1957).[6] The study made an immediate impact. Together with the studies of the atomic bomb survivors, it became the most important data set to establish the carcinogenic

[2] On Muller's notion of 'genetic load' and his position in the classical/balance controversy, see Paul (1987).

[3] On the studies of the atomic bomb survivors and the reasons for the predominance of genetic investigations, see Beatty (1991) and Lindee (1994). Epidemiological studies on the effect of radiation treatment on patient populations were provided by Michael Court Brown and Richard Doll as well as by Alice Stewart in Britain, although her studies on the increased incidence of leukaemia in children whose mothers had undergone X-ray analysis only found recognition later (Greene, 1999; Smith, 2007). On Court Brown and Doll's study, see more below.

[4] See for instance the 1956 Medical Research Council Report to the British Parliament on the issue as well as the guidelines published by the International Commission on Radiological Protection (Medical Research Council, 1956, pp. 63–64; *Recommendations*, 1959).

[5] On the contested disciplinary status and the changing practices of anthropology in the 1950s, see below.

[6] For an excellent account of the background to the study as well as its impact, see Smith (2007). Doll considered his paper with Court Brown on the dose–response relationship between radiation and leukaemia as the 'best-designed study I have ever participated in and possibly my best work' (Darby, 2003).The study marked the beginning of a long collaboration between the two researchers. Their common work included studies on the cancer mortality of British radiologists and on the incidence of leukaemia after diagnostic exposure in utero. In 1959 Court Brown and Doll travelled to Japan to study the leukaemia incidence in atomic bomb survivors. On that trip Court Brown also tried to convince the US-funded Atomic Bomb Casualty Commission, which had been studying the survivors since the mid-1940s, to undertake large-scale cytogenetic studies, a project that was eventually pursued (Bloom, Neriishi, Kamasa, Iseki, & Keehn, 1966).