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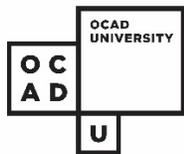
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# Ideas About Heredity, Genetics, and 'Medical Genetics' in Britain, 1900–1982

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### **Pre-Publication Draft**

## **Ideas About Heredity, Genetics, and ‘Medical Genetics’ in Britain, 1900-1982**

*Abstract:* The aim of this paper is to understand how evolving ideas about heredity and genetics influenced new medical interests and practices and, eventually, the formation of ‘medical genetics’ as a medical specialism in Britain. I begin the paper by throwing highlight on the social and institutional changes through which these ideas passed. I argue that, with time, there was a decisive convergence in thought that combined ideas about the familial aspects of heredity and the health needs of populations with an omnibus ‘genetic’ approach to health and illness that focused on the structures and activities of chromosomes and genes in individuals. I show how this convergence in thought was spurred on, first, by innovations in genetic science and technology in the after 1960, and, second, by negotiated protocols and standards of medical practice worked out by bodies such as the relevant royal colleges, the linked associations and societies for medical professionals, affected training and research authorities, and the state. The notion of ‘medical genetics’ in Britain consequently gained a semblance of unanimity over its basic reference points and arrived at a meaning directly tributary to current acceptance of the term in the context of a medical specialism.

*Keywords:* Heredity; Genetics; Medical Genetics; Professionalisation; Medical Specialisms in the UK.

## 1. Introduction

The history of ideas concerning heredity, genetics, and medicine in Britain has mainly been studied in relation to eugenics in the first half of the twentieth century and to advancements in molecular biology in the final third. With regard to the former, the history of eugenics in Britain has been well traversed by historians.<sup>1</sup> In the latter case, by contrast, historians of molecular biology have produced excellent studies of the discovery of DNA and the significance of the larger effort to map disease-causing genes.<sup>2</sup> But what has been left out of the picture is the increasing medical interest in human genetics after the Second World War and the enormous amount of work that went into organising a new medical specialism: ‘medical genetics’. Accordingly, the focus of this paper is on ways genetics and new ideas about heredity were taken up in mainstream medicine and the circumstances under which the clinicians and scientists involved set about turning their work into specialty work based on a ‘genetics-based approach’ to health and illness.

To fully appreciate the changes in medico-scientific conceptions about the relationship between heredity and heritable disease we must remind ourselves that ideas about heredity predate whole vistas of medical science including genetics, epidemiology, immunology, endocrinology, and laboratory diagnostics.<sup>3</sup> There are already more than a few good studies that show how ideas about the relationship between heredity and heritable disease were linked to an assortment of debates about rising crime and criminality, poverty and pauperism, temperance, and the declining birthrate in Britain after 1870.<sup>4</sup> Science writers shared a common language with other educated readers and writers, drawing openly upon literary, philosophical, and historical references as part of their arguments. Ideas moved ‘rapidly and freely to and fro between scientists and non-

scientists: though not without frequent creative misprision'.<sup>5</sup> Thus, deeply rooted concerns about the quality of bloodlines appeared in the personal observations, anecdotes and case descriptions of scientists and physicians who could be equally critical of 'excessive inbreeding' amongst peers of the realm and of generations of pauper families to be found in the slums of Victorian Britain.<sup>6</sup>

By and large, the hereditarily 'tainted' families of the nineteenth century were amongst those groups of individuals who became collectively viewed as counterpoint to Victorian optimism about industrialism and the inevitability of social progress. Legislative reforms to, variously, encourage and suppress the growth of segments of the population came to rely on a series of interrelated claims surrounding theories of demographic transition and evolving systems of social relief introduced by the Poor Law Act of 1834. As Dorothy Porter has pointed out, ideas about the 'health' of populations during this period were 'founded upon a political economic philosophy which intended to use statutory regulation to enhance the free operation of market relations'.<sup>7</sup> The declining British birthrate reported after 1870 in the demographic literature here became an added source of anxiety to what was perceived as the challenges to Britain's economic pre-eminence in the world.<sup>8</sup> By the time of Queen Victoria's death in 1901, the annual crude birthrate in the British population had fallen from a recorded high of 36.3 births per 1,000 persons in 1876 to 28.5 births, a decline of more than 20 percent.<sup>9</sup>

Generations of Victorians had associated the ideas of high fertility and large families with vitality and the progress of the nation. But, according to early-twentieth century demographers, the pressure for high fertility that characterised the 'country way of life' was giving way, on the one hand, as the costs of rearing a 'large' family became

an increasingly heavy burden for city dwellers and, on the other, as city dwellers succumbed to ‘unhealthy’ lifestyles of urban existence.<sup>10</sup> As a consequence, fears about ‘depopulation’ and ‘race suicide’ sparked a proliferation of prolonged, often contentious national debates in the years leading up to the First World War. The dwindling of the British family, it was feared, would bring a halt to economic growth, followed by a relentless decline. Additionally, the demographic literature indicated that fertility among the middle- and upper-classes was substantially lower than that of the labouring poor. This raised uncertainties about the future ‘quality’ of the race; fears that the numbers of the ‘pauper class’ might overtake those of the ‘civilised’ classes. Thus, emphasis was placed in Britain not upon the Malthusian consequences of unchecked fertility but upon the implications of a prolific labouring poor and low fertility among the middle- and upper classes (i.e., a differential birthrate). Edwardian advocates of Social Darwinism, in turn, endorsed pro-active ‘restrictive practices’ and policies designed to curb the reproduction of those ‘less evolved than their betters’.<sup>11</sup>

The subsequent rise of the eugenics organisations and schemes to prevent the decline of the British race through the ‘scientific management’ of populations grew alongside, on the one hand, new directions in social reform to deal with issues of fertility and class and, on the other, the development of new policies and health services to improve ‘child health’ and the ‘fitness’ of families. On the one hand, eugenicists seized on the tenets of Darwinian evolutionism to claim that the ideas of *socially* containing the ‘pauper’ class and *biologically* maintaining the race were mutually antagonistic. The belief in ‘progressive’ evolutionism – propped up by the unproven assumption that many physical, mental, moral, and behavioural dispositions were hereditary – asserted that

social provisions for improving the living conditions of the (biologically) disadvantaged only postponed their inevitable deterioration. An argument followed that the indigent urban underclass was beyond moral instruction and if the prolific breeding of this group was not controlled, pauperism and its associated undesirable qualities – intemperance, criminal recidivism, ‘weakness of constitution’, insanity, disease – would necessarily keep on increasing until the progressive evolutionary direction of the British race was reversed.

Alternatively, others argued that since the poorer and less well-nourished classes were inevitably more prolific, policies to educate and improve the living conditions of the labouring poor would ensure enough people of ability that could climb the social ladder and fill the gaps left by their less fecund ‘betters’. So, for example, policy debates surrounding the Notification of Births Act of 1907 stimulated the development of a health visiting service to instruct and advise mothers on the care of young children.<sup>12</sup> The discovery of poor standards of health among army recruits for the Boer War led, correspondingly, to the establishment by government of an Interdepartmental Committee on Physical Deterioration, whose report, published in 1904, made a series of recommendations aimed at improving child health. Two of the outcomes were the 1906 Education Act, which provided the basis for school meals services, and the 1907 Education Act, which led to the development of the school medical service. A third outcome, the *Interim Report of the Consultative Council on Medical and Allied Services* of 1920, drew upon policies designed after 1902 for the reorganization of the military and for universal elementary education as well as child health and ‘the unity of preventive and curative medicine’. The ideas of hierarchical regionalism and ‘primary health

centres' were here promoted alongside a wide range of educational and athletic activities, designed to keep the population in an 'optimal' state of mental and physical fitness.<sup>13</sup>

As W. A. Robson noted, the idea of a hierarchical regionalism of health care was not implemented until the Second World War with the adoption of a regional basis for Civil Defence. This resulted in the creation of a centrally administrated integrated hospital service with schemes for the administrative separation of various specialised functions, such as the Public Health Laboratory Service and the Blood Transfusion Service, and specialised services such as those for patients with mental handicap.<sup>14</sup> Years later, and highly pertinent to the subject of the present study, this was followed by a set of ten-year plans prepared for local health authorities which provided the backdrop for a series of preliminary investigations into the efficacy of services for the diagnosis and management of congenital and hereditary disorders in families. The plans affected a range of services previously provided by the local authority welfare and children's departments, as well as some of those previously administered by the health departments. Specifically with regard to 'medical genetics', a decisive convergence in ways of thinking about the subject emerged in the 1970s that combined ideas about the familial aspects of heredity and the health needs of *populations* with an omnibus 'genetic' approach to health and illness that focused on the structures and activities of chromosomes and genes in *individuals*. This laid the foundation for the notion of an 'integrated genetics service' in the 1980s that embraced a wide set of service relations in both curative and preventive medicine.

I provide, in what follows, background information for understanding the origins of the British genetics community after 1900 and early physician interests in heredity and

genetics. I briefly examine a range of scientific and clinical interests that were brought together, first, by advances in human cytological research and the development cytogenetic laboratory tests, second, by innovations in the biochemical analysis of metabolic disease, and, third, by the availability of mid-trimester amniocentesis and the advent of national-wide antenatal diagnostic services. This provides a framework for an understanding of medical genetics as applied human genetics. The main focus of the paper, however, is the question of why and how British geneticists subsequently created an expert role for themselves in the delivery of diagnostic and patient care services.<sup>15</sup> I show how medical genetics as a service specialism was assessed and configured to fit local health service requirements, spurred on by negotiated protocols and standards of medical practice worked out by bodies such as the relevant royal colleges, the linked associations and societies for medical professionals, affected training and research authorities, and the state.

## **2. Genetic Science in Britain**

The available evidence suggests that although scientific work on genetics gained acceptance between 1915 and 1930 in Norway, Sweden, Denmark, North America and the Soviet Union, it was less well received in Germany, and quite poorly received in Britain and France.<sup>16</sup> In his study of genetics in the early twentieth century, Jonathan Harwood draws attention to the comparative institutional strength of American geneticists in terms of the sheer scale of expansion in the educational and research sector between 1880 and 1919. Harwood suggests that although older life science programmes at American universities were dominated by traditional natural history foci, often working with the broad theoretical problems associated with evolution, newer

programmes were established after 1890 that were more likely to promote experimental research. Genetics, which happened to be emerging in the midst of such growth, obtained the widest possible support in America by concentrating on the development of improved forms of pedigreed plants and animals.<sup>17</sup> Little distinction was made in this context between what was called ‘genetics’ and ‘practical breeding’. After 1918, nonetheless, it is clear that genetics in the United States ‘began to take the form of a sanctioned normative practice with its own well-defined methods and explanatory standards’.<sup>18</sup> The agricultural connection served to provide geneticists with an institutional setting in which they could meet quasi-professionally and publish.<sup>19</sup>

By contrast, the slow growth of genetics in Britain has been attributed mostly to rivalries among competing groups in the life sciences.<sup>20</sup> Briefly, it is argued that the ‘rediscovery’ of Mendel’s laws of inheritance in 1901 occurred during a period when the study of heredity in Britain was linked to an array of inquiries and disputations concerning how physical characteristics are transmitted between generations and the manner in which an organism grows, develops, and ‘evolves’. Each line of inquiry provided a range of possibilities for investigating these matters. But, as previous studies have shown, the ‘possibilities were defined not only by the current theories or beliefs about heredity, but by the nature of the objects accessible to investigation, the equipment available for examining them, and the methods of observing and discussing them’.<sup>21</sup> The resulting struggle has been represented as a significant roadblock in the development of the British genetics community.<sup>22</sup> The salient point, however, for the purposes of the present study, is that genetics represented a kind of unorthodox scientific research and was not well-positioned within British scientific academia for many years.

The origins of the British genetics community can be traced to a small group who pursued, under the leadership of the zoologist William Bateson, hybridisation experiments with animals and plants. Bateson had left a post in zoology at Cambridge University in 1910 to become director of the John Innes Horticultural Institution at Merton, London. In June of 1919 he gathered together twenty-six interested people at the Linnean Society's rooms in Burlington House to form the Genetical Society of Great Britain. The first meeting in Cambridge on July 12 included staff members of the horticultural facilities at the John Innes, Rothamstead, Long Ashton, Wisley and Kew; and representatives of the Ministry of Agriculture and of commercial horticultural firms. The stated remit of the Society was 'to promote the advancement of genetics and intercourse among persons interested in that science'.<sup>23</sup> Their purpose was to unravel the complexities of the so-called compound characters and to enquire into the generality of the property of dominance in Mendel's laws of inheritance. There were 108 members in 1924: 42 were private individuals and plant or animal breeders, 3 in agricultural administration, 37 were at research institutes, 25 were at universities, and 1 was engaged in medical research.<sup>24</sup> Membership grew very slightly within the imposed limit of 120 until 1936. Then it increased more rapidly through to the outbreak of war in 1939. The percentage of members in the universities rose sharply between 1945 and 1949, most likely due to the general rehabilitation of the universities after the war and, more particularly, an increase in opportunities in the areas of medical research and education.

Indeed, by the start of the 1930s, there were very few genetics courses in British universities: Reginald Ruggles-Gates at King's College, London included genetics along with botany. Lancelot Hogben was appointed Chair of Social Biology at the London

School of Economics in 1930, and, in 1933, John Burdon Sanderson Haldane transferred from the John Innes Horticultural Institution at Cambridge to take the Chair of Genetics at University College, London. Two sympathetic departments outside of London were Alexander Carr-Saunders' School of Sociology in Liverpool and Frank A. E. Crew's Institute of Animal Breeding in Edinburgh. Cambridge University became the main centre for British genetics studies in the interwar years. The journal, *Annals of Eugenics*, published for the Galton Laboratory by the Cambridge University Press after 1925, served as an organ for the dissemination of genetics research until the Genetical Society of Great Britain created its own journal, *Heredity*, in 1947. The steady increase in genetics-related articles in *Annals of Eugenics* is reflected in modifications to its subtitle over, roughly, twenty-five years: 'a journal for the scientific study of racial problems' (1927-1934), 'a journal devoted to the genetic study of human populations' (1934-1945), 'a journal of human genetics' (1946-1954).

#### **4. Heredity, Genetics and Medicine in Britain**

By all accounts, the average British physician was not terribly interested in genetical explanations about the relationship between heredity and disease causality until well after 1960.<sup>25</sup> Indeed, David Lewis's survey of the membership lists of the Genetical Society of Great Britain shows a sharp increase in members involved in medical research after 1959, rising steeply to 1969 when nearly 12 per cent of the 900 members of the Society were working in medicine.<sup>26</sup> Lewis attributed this increase to technological advances in human cytological research and work being done in the area of chromosomal abnormalities in humans. He also cited the research surrounding the Rh blood group and haemoglobin

variants being done at the National Institute for Medical Research, Lister Institute, Chester Beatty Research Institute; and the Medical Research Council units in Britain.

Human cytogenetics (i.e., human cytological analysis) in the 1950s involved techniques largely developed in cytological studies of animal and plant species carried out in the 1920s and 1930s.<sup>27</sup> Improved methods during this period made it easier to count human chromosomes and to study their morphological change. This, in turn, permitted some types of chromosomal abnormalities, including missing or extra copies of a chromosome or gross breaks and rejoinings (translocations), to be detected by microscopic examination. As regards human cytology, the presence of an additional small acrocentric chromosome in typical cases of Down's syndrome was first reported in France by Jérôme Lejeune, Marthe Gauthier and Raymond Turpin in 1959.<sup>28</sup> This was quickly followed by reports from cytological laboratories in England.<sup>29</sup>

It is important here to stress the novelty of human cytogenetics during this period: chromosomal abnormalities were mostly of unknown aetiology and this was a time of uncertainty concerning the precise relationship of congenital malformations and hereditary disease. Cytogenetics provided the first clinical tools to uncover the genetical make-up of common disorders. New ways of thinking about congenital malformations, heredity and disease followed. And the publication of findings in quick succession during 1959 caused a sensation among scientists and physicians in Britain. Alan C. Stevenson, then director of the Medical Research Council Population Genetics Research Unit at Oxford, advised readers of the *British Medical Bulletin*:

For simplicity it is convenient to consider these congenital and hereditary disorders in three groups: (i) those malformations recognizable by the naked eye that have already arisen in intra-uterine life, conventionally termed congenital malformations; (ii) disorders

or diseases determined by single-gene substitutions; and (iii) those where the genetic contribution is more complex.

Such grouping on grounds of aetiology is [however] irrational. There is overlapping in terms of age-groups affected and between macroscopic and microscopic anomalies. Further, estimates of frequencies should never be given without precise statement of which traits and disorders are included. There are many hundreds of traits and there is no detailed and generally accepted nomenclature and classification of congenital and hereditary disorders to which reference can be made.<sup>30</sup>

Again, the case of Down's syndrome provides a particularly useful illustration of changing perceptions concerning congenital malformations, heredity and disease during this period. Down's syndrome was traditionally considered to affect the offspring of 'women of advanced maternal age'. In 1960, Paul Polani and Charles Ford suggested that it could also arise from a chromosomal defect known as a reciprocal translocation.<sup>31</sup> Polani later confirmed this proposal in a paper published with Cedric O. Carter, a member on staff of the Medical Research Council's Clinical Genetics Unit at the Institute of Child Health, The Hospital for Sick Children, London.<sup>32</sup> Using evidence collected by Carter, Polani proposed that a family history of Down's syndrome was often caused by the transmission of a translocation between parent and child.

A standard system of nomenclature for human mitotic chromosomes was ultimately established in 1960.<sup>33</sup> The classificatory system facilitated greater cooperation between scientists in France, Sweden, Japan, North America, and the UK, as well as enhancing the diagnostic potential of cytogenetics in clinical medicine. At the same time, it was clear that the ability to serve increasing demands for cytogenetic diagnosis in Britain was compromised by a lack of resources.<sup>34</sup> There were approximately thirty laboratories equipped to handle requests for cytogenetic diagnosis, but they were generally small research outfits housed in paediatrics, pathology and haematology departments.<sup>35</sup>

With the benefit of hindsight, it can be argued that new kinds of occupational roles and working relationships were appearing in medicine in the 1960s as a result of such innovations in genetic science as cytogenetic analyses of chromosomal abnormalities. It is nonetheless important to stress that the impact of these innovations on clinical practice evolved slowly, with wide disparity and divisions in how the individuals involved thought about themselves and the ways they characterised their activities.<sup>36</sup> This point is made abundantly evident in another innovative area during this period: biochemical analysis of metabolic disease. The case of phenylketonuria is particularly instructive in this regard; it is frequently cited in the genetics literature as an exemplar of modern hereditary disease management.

The cause of phenylketonuria was identified in 1934 by the Norwegian physician and biochemist Asbjorn Følling. The study of the disease was taken up in England in the mid-1930s by Lionel Penrose, a science graduate of Cambridge and physician, employed by the Medical Research Council (MRC) to conduct a detailed study of the origins of mental deficiency amongst patients at the Royal Eastern Counties' Institution at Colchester. Juda H. Quastel, a biochemist and a collaborator in the study, coined the name 'phenylketonuria', which was contracted to PKU.<sup>37</sup> Penrose, like Følling, proposed that abnormal metabolism of the amino acid, phenylalanine, was the cause of mental deficiency. Studies by the American physician and biochemist, George Jervis, confirmed that the disorder originated in the liver during infancy and was caused by an inherited inability to convert (metabolise) phenylalanine into tyrosine. And a treatment based on a low phenylalanine diet was subsequently developed by a team of biochemists and paediatricians at the Birmingham Children's Hospital. Population screening of newborns

for the prevention of PKU followed in the 1960s, beginning with the testing of infants' urine for a metabolite of phenylalanine.<sup>38</sup> Nation-wide neonatal screening for PKU was established in 1969 with the aim 'to reduce morbidity by complete and timely detection and treatment of affected cases'.<sup>39</sup>

The salient point here, for the purposes of the present study, is that because neonatal screening services were provided throughout the country at university children's hospitals by paediatricians, obstetricians and other specialists in the field of neonatal medicine, they remained managerially and operationally separate from the type of services associated, years later, with genetic diagnostics and counselling in regional genetic centres. Likewise, the role of specialist geneticists in the diagnosis and management of Rh incompatibility, the thalassaemias, and other haematological conditions remained marginal in relation to that of haematologists, obstetricians and specialists in blood transfusion and serology.<sup>40</sup> Thus, it is clear that, in the 1960s and 1970s, a number of different possibilities could be conflated under the heading of genetics and medical responses to hereditary disease.<sup>41</sup> But, at the same time, an alternate conception of the field came to light involving what Robert Platt, physician and Professor of Medicine, University of Manchester, called 'a kind of general practitioner of human genetics who, though not able to use all the techniques himself, can nevertheless maintain a general view of the fields in which they can be applied'.<sup>42</sup> In the remainder of the paper I focus on medical activities utilising genetic knowledge and techniques that were subsequently recognised professionally as the work of a new medical specialism.

## 5. British Medical Genetics

By 1964 there was sufficient medical interest in a genetical understanding to disease to sustain both a specialist journal and a new society. In September the first volume of the *Journal of Medical Genetics* was published under the editorship of Arnold Sorsby. The *Journal* was mostly London-based: Sorsby worked at the Wernher Research Unit on Ophthalmological Genetics (MRC), and was accompanied on the editorial committee by G. R. Fraser of the Godfrey Robinson Unit (RNIB), Department of Ophthalmology at the Royal College of Surgeons of England and the Royal Eye Hospital, London. Other members of the editorial committee included John Alexander Fraser Roberts and Cedric O. Carter, both members of the staff of the Medical Research Council's Clinical Genetics Research Unit at the Institute of Child Health, Hospital for Sick Children. The Child Health Institute, on its inception in 1949, had taken on and integrated Britain's earliest heredity counselling clinic, established in 1946 under the auspices of the Hospital for Sick Children. The *Journal* was designed to provide opportunities for academicians and practitioners in the new field of medical genetics to demonstrate, through peer review, that they were involved in scientifically legitimated modes of practice. In his inaugural editorial Sorsby asserted:

The considerable periodical literature in English on the various fields covered by present-day genetics includes two journals devoted exclusively to heredity in man. Both these – the *Annals of Human Genetics* and the *American Journal of Human Heredity* – aiming, as they do, to cover all aspects of heredity in man, carry contributions on medical genetics, but most genetical papers of medical interest – and these are becoming increasingly numerous – are widely scattered throughout an ever-increasing number of specialised medical journals. Things are rather better on the continent, but the *Journal of Medical Genetics* is the first to be exclusively medical and to be broadly based. As such, it is a timely venture.<sup>43</sup>

Further opportunities for collegial support and approbation came from the Clinical Genetics Society – also established in 1964. This was an academic society and functioned

primarily as a forum for presenting patients and case studies. Like the *Journal of Medical Genetics*, the Society was London-based, run mainly out of the Child Health Institute. In 1970, the membership of the Society had risen to 164 individuals and it was decided that a more formal approach to meetings would be taken.<sup>44</sup> Cedric O. Carter and Sarah Bunday of the Child Health Institute acted as Chair and Secretary of a steering committee. Other London committee members were Paul Polani and Martin Bobrow (Paediatric Research Unit, Guy's Hospital) and Michael Laurence (University of Wales, Cardiff; formerly at the Child Health Institute). National representation on the steering committee included: Alan Emery (Western General Hospital, Edinburgh), Malcolm A. Ferguson-Smith (Royal Hospital for Sick Children, Yorkhill, Glasgow), Alan W. Johnston (Woodend General Hospital, Aberdeen), Norman C. Nevin (Institute of Clinical Science, Belfast), Cyril Clarke (Alder Hey Children's Hospital, Liverpool), Rodney Harris (United Manchester Hospitals), M. d=Auvergne Crawford (St. James Hospital, Leeds), Derek F. Roberts (United Newcastle-upon-Tyne Hospitals), Eric Blank (United Sheffield Hospitals), Jack Insley (Birmingham Maternity Hospital, Edgbaston), Richard H. Lindenbaum (Churchill Hospital, Headington, Oxford), and Mary Vowles (Royal Devon and Exeter Hospital, Exeter).

Initially the Clinical Genetics Society represented a kind of divided nucleus within the emerging field of medical genetics; a polycentric structure with multiple bases of interest. At the most basic level, the Society was divided along lines of primary specialisation. This became most apparent with the Society's early recommendations on the training of medical geneticists. The recommendations were prepared at the request of the Executive Committee for Joint Higher Medical Training of the Royal College of

Physicians.<sup>45</sup> The issue of training was then placed under the supervision of the Paediatric Specialist Advisory Committee (SAC) of the Royal College of Physicians.<sup>46</sup> The Paediatric SAC, in turn, proposed that the Member of the Royal College of Physicians (MRCP) would be considered by them to be an essential qualification, as it was then for consultants in every other branch of clinical medicine.

The Paediatric SAC's suggestions followed a particular line of logic: parents with a family history of hereditary disease or congenital malformations were traditionally seen by paediatric specialists with expertise in the care and treatment of diseases and disorders of the newborn. Up to this point in time genetics had been useful for purposes of prognosis, i.e., predicting the recurrence or outcomes of a disease or disorder in an affected family. The new testing regimes of the 1960s for identifying chromosomal anomalies and metabolic diseases widened the remit of genetic consultation within paediatrics. And geneticists were clearly experiencing increases in workload with the work associated with mid-trimester amniocentesis and antenatal diagnosis.<sup>47</sup>

At the same time, the membership of the Clinical Genetics Society had a varied background including specialists in ophthalmology, psychiatry, obstetrics and general practice – fields that were not branches of clinical medicine under the Royal College. Having the MRCP as a training requirement, many believed, would be too restrictive and potentially hold back the growth of genetics in clinical specialty areas other than paediatrics. Further to this, concerns were raised about the evolving relations between clinical and laboratory personnel. Some members went so far as to suggest that training for medical genetics should require the MRCPath. In other words, geneticists should run laboratories under the control of departments of pathology *and* also see patients.

Alternatively, others suggested separate roles for the clinician and scientist, advocating a formal association be made between clinical genetics, cytogenetics and biochemistry. In the end, the arguments and debates resulted in a breakup within the Clinical Genetics Society. The cytogeneticists, fearing a paediatrics-dominated Society would exclude non-physicians, split off and formed their own body, the Association of Clinical Cytogeneticists. The Association became an organisation that arbitrated in matters concerning the research and service functions of cytogenetic laboratories. This set the example, much later, in 1988, for a third body, the Clinical Molecular Genetics Society, representing scientists interested in diagnostic applications of molecular genetics. A final organisation, the Genetic Nurses and Social Worker's Association, was also created that year for allied health personnel working in the field.<sup>48</sup>

In 1978, Alan Johnston, reporting on the findings of the Clinical Genetics Society's *ad-hoc* working party on the training of medical geneticists, remarked that whilst the chance convergence of different disciplines into the field of human genetics had resulted in a number of spectacular contributions 'which have had repercussions through the whole science of genetics',<sup>49</sup> medical genetics had progressed to the point where highly specialised training and professional standards of practice were now necessary.<sup>50</sup> Citing the example of the recently incorporated Canadian College of Medical Geneticists, the working party envisioned the Clinical Genetics Society establishing and maintaining standards of health service delivery. The medical geneticist of the future would have four main functions;

to contribute to diagnosis (including prenatal); to counsel patients and their relatives; to maintain genetic registers; and to act as consultant to cytogenetic, biochemical, and other relevant laboratories. In short, he or she would be responsible for organising a comprehensive genetic advisory service. Additional functions would be involvement in

teaching and research, which would be particularly important since he would be working mainly in major centres, usually teaching hospitals. Thus a joint NHS/university appointment at consultant level to a Genetic Advisory Centre is preferable.<sup>51</sup>

The role of the medical geneticist and the creation of regional genetic services were topics pursued in detail by the Clinical Genetics Society in two subsequent working parties.<sup>52</sup> The Royal College of Physicians became involved, initially through the work of the Medical Genetics Sub-Committee of the Paediatric Specialty Advisory Committee and, in 1984, with the formation of a standing Committee on Clinical Genetics. Both the Medical Genetics Sub-Committee and the standing Committee were largely made up of members of the Clinical Genetics Society.

Medical genetics in this way came to exemplify the type of specialised work Victor Thompson described in terms of ‘task specialization’.<sup>53</sup> Using Thompson’s nomenclature, specialization of tasks refers to work specificity, i.e., ‘making activities more specific’. This he contrasted with the specialization of people, referring to the adaptation of the individual to their circumstances. In the context of British medical genetics, it can be said that, prior to 1970, there was high personal specialization in the genetics community. The community was an aggregate of various primary specialisms with clinicians and scientists operating within the limits of their individual specialties, experiences, and skills. In the late 1970s and early 1980s the situation changed. Individuals gravitated into associative groupings according to the specificity of certain tasks. Each group pursued different objectives in different ways and they were more or less delicately held together under a common title of ‘medical genetics’. Internal differentiation eventually became highly structured and clearly evident in formal educational tracks, certification processes, and well-defined societies and associations.

The ‘symbolic cement’ that would hold together these groups over time was the joint NHS/university appointment first discussed in Alan Johnston’s report. This appointment provided the basis for building an integrated genetics service, in line with other consultant-led services that linked research, laboratory services, clinical work, and education/public health functions. The integrated genetics service, in turn, was made to fit the hospital-dominated service specialisms of the NHS reorganisations of 1974 and 1982.

## **6. Medical Genetics and the NHS**

A comprehensive review of precisely how genetic diagnostic and laboratory services evolved in relation to other hospital-based services under the NHS is beyond the scope of the present paper. A more limited reappraisal of the organisational concepts of ‘hierarchical regionalism’ and ‘regional health centres’ and their role in the development of an integrated genetics service will therefore be undertaken in order to suggest future directions for more detailed historical analyses.

As Charles Webster has shown, the use of the terms ‘hierarchical regionalism’ and ‘regional health centres’ in British health policy extend back at least to the *Interim Report of the Consultative Council on Medical and Allied Services* of 1920.<sup>54</sup> Aptly named the Dawson Report, the recommendations of the Consultative Council very much bore the imprint of the chair of the responsible committee, Major-General Sir Bertrand Dawson, a senior figure in the army medical service. Significantly, Dawson did not regard medical policy a special case from other aspects of welfare policy. And he was attentive to contemporary concerns about the ‘fitness’ of the British citizen and the

‘physical deterioration’ of young men entering national service. Moreover, Dawson drew upon policies designed after 1902 for the reorganization of the military and for universal elementary education:

Dawson ... gave prominence to the existing school medical service, which provided a direct link between local health and education services. It was itself created in the aftermath of the Boer War, and it was concerned with building up the physical fitness of the younger generation, as well as with early diagnosis and treatment of minor conditions. The school medical service therefore exemplified for Dawson the inseparability of curative and preventive medicine. The principle was strongly developed in his lectures and in the Dawson Report. Replacement of the terms ‘hospital’ and ‘medical centre’ by the alternative ‘health centre’ terminology signified his commitment to the unity of preventive and curative medicine. The primary health centres, in particular, were to be concerned with the widest range of remedial and athletic activities, designed to keep the population in a maximum state of physical fitness. The Dawson Report emphasized that ‘physical culture is thus concerned with education, with the maintenance of the health and recreation of the people, and the curing of disease and disability, and there is no sharp line of demarcation between these functions’.<sup>55</sup>

The Report further emphasised that all health services, both curative and preventive, be integrated in regional catchment areas and brought together in close coordination under a single health authority.<sup>56</sup> Proximity to a university teaching centre was regarded as important for functional integration. At the same time, Dawson’s Consultative Council was unable to agree on the nature of the health authority and the size of the catchment areas was not specified in detail.

The idea of a hierarchical regionalism of health care that emerged from the Dawson Report failed to garner much support in the interwar years. Local authorities resisted the idea of joint planning with one another or with voluntary hospitals.<sup>57</sup> And there was little cooperation among the various hospital administrations. Furthermore, the Report made little initial impact within the Ministry of Health. The Consultative Council that issued the Report was allowed to fall into abeyance and no subsequent reports were produced. The idea of hierarchical regionalism was nonetheless resurrected during the

period of the Second World War with the adoption of a regional basis for Civil Defence. Specifically, this resulted in the creation of a centrally administrated integrated hospital service.

One way of successfully overcoming resistance to the idea of regionalisation in health care emerged with schemes for the administrative separation of various specialised functions, such as the Public Health Laboratory Service and the Blood Transfusion Service, and specialised services such as those for patients with mental handicap.<sup>58</sup> As regards medical genetics, a set of ten-year plans prepared in 1972 for local health authorities provided the backdrop for a series of preliminary investigations into the efficacy of services for the diagnosis and management of genetic diseases. The plans affected a range of services previously provided by the local authority welfare and children's departments, as well as some of those previously administered by the health departments. The main objective was to integrate services that had previously been administered separately, and to provide for the development of a comprehensive family service.

The initial inquiries into the status of genetic diagnostic and laboratory services in Britain showed that there was considerable variation in the range of services offered from region to region and much of what made up service arrangements was largely dependent on university and research funding.<sup>59</sup> Geneticists offering these services, on the other hand, were clearly willing to cooperate with government programs for integrated services so long as sufficient resources were made available.<sup>60</sup> The Clinical Genetics Society consequently set up working parties in the 1980s who in turn endorsed an 'integrated regional genetics service' that would act as the 'focus to which the primary health care

team, hospital consultants and other specialists should direct particular problems relating to inheritance, including the diagnosis and prevention of birth defects'.<sup>61</sup> Moreover, each service centre would set up computer registration of genetic diagnostic information (genetic registers) for the express purpose of tracing, following-up, and counselling individuals at risk of having or transmitting a serious genetic disorder. These centres would, in accordance with the structure of NHS Regional Health Authorities, serve a population of 1 ½ to 3 million and would, on this basis, provide for several District General Hospitals. The rationale for the allocation of catchment areas was based on the calculation of the proportion of, first, congenital malformations and, second, chronic disease with a major genetic component. In addition, the 1983 Working Party envisaged programmes to extend counselling to district general hospitals through regular visits by centrally based whole-time clinical geneticists.<sup>62</sup> Local clinicians would prepare information on patients in advance of the 'satellite' clinics and visiting clinical geneticists would share after-care responsibilities with family practitioners.

The organisation of the genetics centres themselves would follow along the lines of the multidisciplinary centres established earlier in North America – but with noticeable differences. Whereas the North American centres were predominantly run by scientists, the UK centres would have a strict division of labour comprising separable roles for clinicians and scientists.<sup>63</sup> Consultants (i.e., clinical geneticists) would be responsible for genetic counselling and syndrome identification for all referred individuals. In a significant number of cases, this would entail further assessment following counselling and, in some cases, long term supervision of patients and their families. Correspondingly, there would be need for close collaboration between the clinical geneticists and the heads

of laboratories in the areas of, initially, cytogenetics and biochemical analysis and, later, molecular analysis. Both clinical geneticists and scientists would participate in the delivery and monitoring of services as well as in the forward planning of the local genetics services. The 1982 Working Party asserted: ‘there are obvious advantages in such a team being housed under one roof and where there is an academic interest it would be appropriate for the University to share in providing accommodation and facilities for teaching and research’.<sup>64</sup>

At a high level of generality, the formal structure for the integrated genetics service proposed by the Clinical Genetics Society fit well within the regimes of service delivery instituted between 1974 and 1982 when the National Health Service was reorganised to integrate hospital, community health care and family practitioner services under a unified management structure. By 1982 genetic laboratory services and counselling clinics were to be found in nineteen NHS regions across the UK. The ‘regional genetic centre’ became the hallmark of British medical genetics, holding regular clinics in the centre and also ‘satellite clinics’ to which clinicians would be dispatched to see patients in District General Hospitals. A decade later, responsibility for genetic services would devolve from regional administration to conurbations of districts.

Summarily, the notion of the ‘integrated genetics service’ that emerged in the 1980s embraced a wide set of service relations in both curative and preventive medicine. The regional genetics centre, though of great importance, was but the nucleus of an extended pattern of interrelations. In its simplest spatial aspect, local services were comprised of two generalised unit parts: the centre and the adjoining catchment area. The two developed together, each presupposing the other. But while the centre was compact

and readily visible, the catchment area was diffuse and difficult of precise observation. The boundaries of regional genetic services in fact appeared in varying degrees of distinctness. Relations between consultation and laboratory areas were elastic. In a large hospital setting a concentration of services combined with plenty of resources (monetary, human and otherwise) created the impression of self-sufficiency. At the same time, there was order in the movement of patient referrals and resources to and from the local centre. It is possible to observe here a series of concentric zones around each centre which differed in the degree of attachment of their occupants to the centre, of the frequency of movement of patients or patient information to and from the centre, and in the extent to which contacts with the centre were direct, involving the movement of individuals, or indirect, involving a circulation of information and specimens (i.e., patient records and test samples) rather than people. It is here, in the complex resource interdependencies of the NHS that the semblance of medical genetics can be seen as a system of network relations that transcended the local differences of the regional genetics centres.

## **7. Conclusion**

In this paper, I have tried to underscore certain themes and periodisations of direct relevance to the history of ideas about heredity, genetics, and medical genetics in Britain. First, that the British genetics community was not well-positioned within the wider domain of science academia until after 1960. With specific regard to medicine, the advent of new laboratory testing regimes of the 1960s for identifying chromosomal anomalies and metabolic disease represented a kind of turning point in ‘medical genetics’ in which the organisation of clinical and laboratory labour was transformed. This called for a redefinition of the role of the geneticist in medicine to coincide with a conceptual shift in

emphasis away from the familial aspects of heredity and the ‘fitness’ of populations to an omnibus ‘genetic’ approach to health and illness that focused on the structures and activities of chromosomes and genes in individuals. It can certainly be said that the rise and subsequent legitimisation of medical genetics, beyond showing the success of a new area of medical specialisation within its self-defined limits, presents an opportunity to observe the creation, for the first time, of a distinctly genetical corpus in medicine and of a group of self-designated *medical* geneticists. But, in relation to this, the role of the state and central government cannot simply be reduced to a reactive consideration.

What I am describing here as a turning point in medical genetics occurred at a time when national health policy became divorced from other aspects of British welfare policy, and regionalism in health care was taking central stage. Under the NHS, health centres were adopted as the basis for integrating curative *and* preventive medicine in new regional hierarchies. Health centres were intended to introduce multidisciplinary teamwork, coordination and rationalisation into the delivery of patient care on a national scale. Once appraised of this situation, the individual constituents of what I have described in this paper as a kind of divided nucleus within the emerging field of medical genetics mobilised themselves into an effective interest group which produced a series of influential publications which lay the foundations for the rationalisation of an ‘integrated genetics service’. By skilful negotiation these individuals ensured that their vision for the future of ‘medical genetics’ was effectively registered in the complex negotiations that took place between 1974 and 1982 when the NHS was reorganised to integrate hospital, community health care and family practitioner services under a unified management structure.

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## Endnotes

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<sup>1</sup> See, in particular, Waller (2001a), Paul (1998), Mazumdar (1992), Soloway (1990), Kevles (1985), Farrall (1985), MacKenzie (1976).

<sup>2</sup> See, for example, Cook-Degan (1993), Kevles and Hood (1992), Lee (1991), Holtzman (1989).

<sup>3</sup> It is noteworthy that as early as 1814, in his *Treatise on the Supposed Hereditary Properties of Diseases*, the English physician Joseph Adams had accurately – from a (contemporary) genetics perspective – distinguished between familial diseases ‘confined to a single generation, to brothers and sisters, the children of the same parent’ and hereditary diseases which are ‘traced from generation to generation’. In addition, he identified congenital illness as ‘disease appearing at birth’, noting that such conditions are more frequently familial rather than hereditary. Causal explanations for these ideas, nevertheless, were not available until the next century. It was not until the early twentieth century that genetic theories, based on experimental and statistical examination of the reappearance of visible differences between generations of individuals, were sufficiently mature to provide such explanations. (See Adams, 1985.)

<sup>4</sup> See, for example, Searle (1976), Pick (1989), Oakley (1991), Mazumdar (1992), and Jones (1998).

<sup>5</sup> Beer (1983), p. 7.

<sup>6</sup> Waller (2001b).

<sup>7</sup> Porter (1999), p. 121.

<sup>8</sup> Soloway (1990), pp. 1-5.

<sup>9</sup> Cited in Soloway (1990), pp. 3-4.

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- <sup>10</sup> See, for example, National Council of Public Morals (1916).
- <sup>11</sup> Saleeby (1910), pp. 90-93, 96.
- <sup>12</sup> Parker (1965), p. 27.
- <sup>13</sup> Webster (1990), pp. 122-23.
- <sup>14</sup> Robson (1931), pp. 62, 290, 313.
- <sup>15</sup> My analysis is based on library and archival research complemented by interviewing ninety-eight individuals in twenty clinical settings in the UK between July 2001 and August 2002.
- <sup>16</sup> Harwood (1993); Allen (1978), pp. 278-283.
- <sup>17</sup> The first reports of Mendel's laws appear in the American medical literature in 1903. See Guyer (1903). For an extensive discussion of the subject, see Cravens (1978).
- <sup>18</sup> Sapp (1983), p. 334.
- <sup>19</sup> Kimmelman (1983); Rosenberg (1976), chap. 12.
- <sup>20</sup> See, for example, Rushton (2000).
- <sup>21</sup> Bowler (1989), pp. 246-81.
- <sup>22</sup> Rushton (2000); Paul (1998), pp. 11-13; Kevles (1985), pp. 43-44.
- <sup>23</sup> Genetical Society – Rules; pamphlet in the *Eugenics Society Archive*, Contemporary Medical Archives Centre, The Wellcome Institute for the History of Medicine, SA/ EUG/ AMS/ MF/113/ D78.
- <sup>24</sup> Lewis (1969), pp. 1-7.
- <sup>25</sup> Coventry (2000); Concerted Action on Genetic Services in Europe (1997).
- <sup>26</sup> Lewis (1969), pp. 5-6.
- <sup>27</sup> See, for example, White (1954).

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- <sup>28</sup> Lejeune, Gauthier and Turpin (1959).
- <sup>29</sup> Ford, Jones, Miller, Mittwoch, Penrose, Ridler and Shapiro (1959); Ford, Polani, Briggs and Bishop (1959).
- <sup>30</sup> Stevenson (1961).
- <sup>31</sup> Polani, Briggs, Ford, Clarke and Berg (1960).
- <sup>32</sup> Carter, Hamerton, Polani, Gunalp and Weller (1960).
- <sup>33</sup> Anonymous, ‘A Proposed Standard System of Nomenclature of Human Mitotic Chromosomes’ (1960).
- <sup>34</sup> Public Record Office, MRC (1962).
- <sup>35</sup> Coventry (2000), pp. 177-193.
- <sup>36</sup> Leeming (2001).
- <sup>37</sup> Mazumdar (1992), pp. 235-6.
- <sup>38</sup> Veale (1980).
- <sup>39</sup> Streetly *et al.* (1995), p. 726.
- <sup>40</sup> Coventry (2000), pp. 57-80, 112-30.
- <sup>41</sup> Coventry and Pickstone (1999), p. 1236.
- <sup>42</sup> Platt (1961), pp. 177-8.
- <sup>43</sup> Sorsby (1964), p. 1.
- <sup>44</sup> Clinical Genetics Society, Committee Meeting Minutes, 16 October 1970.
- <sup>45</sup> Clinical Genetics Society, Committee Meeting Minutes, 23 November 1972.
- <sup>46</sup> Clinical Genetics Society, Committee Meeting Minutes, 25 October 1974.
- <sup>47</sup> Refined techniques for culturing foetal cells from amniotic fluid were developed and amniocentesis (i.e., removing amniotic fluid during early pregnancy by puncturing the

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amniotic sac with an aspiration needle), performed by obstetricians, became useful as an outpatient procedure for obtaining test samples for chromosomal and biochemical analysis.

<sup>48</sup> The four organisations eventually came together under an umbrella organisation known as the British Society for Human Genetics, formed in 1996.

<sup>49</sup> Johnston (1978).

<sup>50</sup> The ‘ad-hoc’ working party was formed in November 1973 and consisted of Alan W. Johnston (Chairperson), Derek F. Roberts, Malcolm Ferguson-Smith, Cedric O. Carter, and Alan E. H. Emery. (Clinical Genetics Society, Committee Meeting Minutes, 23 November 1973; 7 March 1975.)

<sup>51</sup> Johnston (1978), p. 260.

<sup>52</sup> Fitzsimmons, *et al.* (1982); Harris *et al.* (1983).

<sup>53</sup> Thompson (1964), p. 25.

<sup>54</sup> Webster (1990), p. 136.

<sup>55</sup> Webster (1990), pp. 122-23.

<sup>56</sup> Dawson (1920), para. 92.

<sup>57</sup> Robson (1931), pp. 62, 290, 313.

<sup>58</sup> Webster (1988), pp. 34-8, 380-8; Honigsbaum (1989), pp. 40-8; Pater (1981), pp. 34-41; Eckstein (1958), pp. 247-52.

<sup>59</sup> Medical Research Council/Department of Health and Social Security Joint Working Group on Genetics (1980), British Paediatric Association (1979), Medical Research Council Sub-Committee on Genetics (1978), Department of Health and Social Security (1977:22-7; 1976a, 1976b, 1976c).

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<sup>60</sup> See, for example, Medical Research Council/Department of Health and Social Security Joint Working Group on Genetics (1980), British Paediatric Association (1979), Medical Research Council Sub-Committee on Genetics (1978), Department of Health and Social Security (1977:22-7; 1976a, 1976b, 1976c).

<sup>61</sup> Fitzsimmons *et al.* (1982), p. 2.

<sup>62</sup> Harris *et al.* (1983), p. 22.

<sup>63</sup> For a detailed discussion of the history of genetics centres in North America during this period, see Leeming (2004).

<sup>64</sup> Fitzsimmons *et al.* (1982), p. 2.