



VIEWPOINT

Origins of human genetics. A personal perspective

Eberhard Passarge ¹

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Abstract

Genetics evolved as a field of science after 1900 with new theories being derived from experiments obtained in fruit flies, bacteria, and viruses. This personal account suggests that the origins of human genetics can best be traced to the years 1949 to 1959. Several genetic scientific advances in genetics in 1949 yielded results directly relating to humans for the first time, except for a few earlier observations. In 1949 the first textbook of human genetics was published, the *American Journal of Human Genetics* was founded, and in the previous year the *American Society of Human Genetics*. In 1940 in Britain a textbook, entitled *Introduction to Medical Genetics* served as a foundation for introducing genetic aspects into medicine. The introduction of new methods for analyzing chromosomes and new biochemical assays using cultured cells in 1959 and subsequent years revealed that many human diseases, including cancer, have genetic causes. It became possible to arrive at a precise cause-related genetic diagnosis. As a result the risk of occurrence or re-occurrence of a disease within a family could be assessed correctly. Genetic counseling as a new concept became a basis for improved patient care. Taken together the advances in medically orientated genetic research and patient care since 1949 have resulted in human genetics being both, a basic medical and a basic biological science. Prior to 1949 genetics was not generally viewed in a medical context. Although monogenic human diseases were recognized in 1902, their occurrence and distribution were considered mainly at the population level.

Introduction

With the completion of the Human Genome Project in 2004 [1] human genetics moved into a new era of exploring the whole genome and its relation to the causes of genetic disorders. New approaches based on numerous new technological advances, such as different automated DNA sequencing methods [2], the elucidation of different types of individual genetic variation [3] and others, allow high resolution analysis of the human genome in various genetic etiologies of diseases [4, 5] in a great number of individuals in different geographic populations [6–9] or analysis of single cells [10]. Earlier genetic studies in human genetics were aimed at individual genes or groups of linked genes. In contrast, during the first 4–5 decades of increasing

knowledge of general genetics since 1900, aspects relating to humans could rarely be considered [11–17]. The term “human genetics” has only been in wide use since 1949 on. “Man is one of the most unsatisfactory of all organisms for genetics studies.” One sentence later: “Obviously no geneticist would study such a refractory object, were it not for the importance that a knowledge of the subject has in other fields.” Thus wrote Alfred H Sturtevant in 1954 [18], expressing an opinion widely held among geneticists before the advent of human genetics (Extended Text #1 in Supp. Mat.).

How did human genetics arise? Here I propose that the origins of human genetics as an independent scientific field can best be traced to the years between 1949 and 1959, when genetic advances could be applied to humans. Several scientific events took place in 1949 that support this idea. In addition, I will briefly review advances relating to human genetics as they apply to medicine and patient care before and after 1949, much of it as a personal witness since 1963.

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Eberhard Passarge
eberhard.passarge@uni-due.de

¹ Institut für Humangenetik, Universitätsklinikum Essen, Essen, Germany

The year 1949

Two new important insights in 1949 serve as hallmarks in the development of early human genetics. James V Neel

Genetics A Human Perspective

Xiaolong Qi



Genetics A Human Perspective:

Genetics Linda R. Maxson, Charles H. Daugherty, 1992 *Sex, Cells, and Same-Sex Desire* David A Parker, 2014-02-25
This fascinating new book explores the myriad aspects of biological theories of sexual preference Sex Cells and Same Sex Desire describes reviews and questions recent biological research on sexual preference from the point of view of knowledgeable scientists and of scholars in the social sciences and humanities representing the emerging field of gay studies The issues involved have a vibrant history are wide ranging and remain the objects of much controversy This book demystifies biological research on sexual preference and makes it accessible to readers unfamiliar with biological and medical research Sex Cells and Same Sex Desire is divided into several sections each of which is introduced by an explanation of key concepts and terms found in that section The book begins with a discussion of the history of biological theories and sociocultural concepts of gender and sexuality The next 3 sections explore specialized areas of biological science and related issues genetics and evaluation hormones and the endocrine system and brain physiology and structure A final section discusses social stigma science and medicine A glossary of terms used by the authors is included so readers may look up those that may be unfamiliar **Current Catalog** National Library of Medicine (U.S.), 1979 Includes subject section name section and 1968 1970 technical reports *Environmental Health Perspectives*, 2004 **A Cladistics Approach to the Genetic Analysis of Human Quantitative Data** Martha Balliet Haviland, 1993 The Genetic Approach to Human Disease Vincent M. Riccardi, 1977 **Human Biology** Raymond Pearl, 1929 Includes section Recent literature useful in the study of human biology Perspectives on Human Biology Im/T Fox, 1991 *Human Biology* Sara Stinson, Barry Bogin, Dennis H. O'Rourke, 2012-03-19 This comprehensive introduction to the field of human biology covers all the major areas of the field genetic variation variation related to climate infectious and non infectious diseases aging growth nutrition and demography Written by four expert authors working in close collaboration this second edition has been thoroughly updated to provide undergraduate and graduate students with two new chapters one on race and culture and their ties to human biology and the other a concluding summary chapter highlighting the integration and intersection of the topics covered in the book **Magill's Medical Guide: Fracture and dislocation - Paralysis**, 2002 **Organic and Biochemistry** Caret, 1993-02 **Index Medicus**, 2002 Vols for 1963 include as pt 2 of the Jan issue Medical subject headings *The Genetic View of Berkeley's Religious Motivation* Granville Stanley Hall, 1912 Perspectives on Science and Christian Faith, 1999 The Pedagogical Seminary and Journal of Genetic Psychology, 1924 Vols 5 15 include Bibliography of child study by Louis N Wilson Paperbound Books in Print, 1992 *Recording for the Blind & Dyslexic, ... Catalog of Books*, 1996 **An Exposition of Fallacies in the Hypothesis of Mr. Darwin** Charles Robert Bree, 1872
Perspectives in Medical Sociology Phil Brown, 1996 Phil Brown has designed this book to make discussion of the issues of medical sociology interesting accessible challenging Molecular Genetics and the Human Personality Jonathan

Benjamin, Richard P. Ebstein, Robert H. Belmaker, 2002 In the 1960s and 1970s personality and mental illness were conceptualized in an intertwined psychodynamic model. Biological psychiatry for many unweaved that model and took mental illness for psychiatry and left personality to psychology. This book brings personality back into biological psychiatry not merely in the form of personality disorder but as part of a new intertwined molecular genetic model of personality and mental disorder. This is the beginning of a new conceptual paradigm. This breakthrough volume marks the beginning of a new era an era made possible by the electrifying pace of discovery and innovation in the field of molecular genetics. In fact several types of genome maps have already been completed and today's experts confidently predict that we will have a smooth version of the sequencing of the human genome which contains some 3 billion base pairs. Such astounding progress helped fuel the development of this remarkable volume the first ever to discuss the brand new and often controversial field of molecular genetics and the human personality. Questioning critical and strong on methodological principles this volume reflects the point of view of its 35 distinguished contributors all pioneers in this burgeoning field and themselves world class theoreticians empiricists clinicians developmentalists and statisticians. For students of psychopathology and others bold enough to hold in abeyance their understandable misgivings about the conjunction of molecular genetics and human personality this work offers an authoritative and up to date introduction to the molecular genetics of human personality. The book with its wealth of facts conjectures hopes and misgivings begins with a preface by world renowned researcher and author Irving Gottesman. The authors masterfully guide us through Chapter 1 principles and methods Chapter 4 animal models for personality and Chapter 11 human intelligence as a model for personality laying the groundwork for our appreciation of the remaining empirical findings of human personality qua personality. Many chapters 6 7 9 11 and 13 emphasize the neurodevelopmental and ontogenetic aspects of personality with a major emphasis on the receptors and transporters for the neurotransmitters dopamine and serotonin. Though these neurotransmitters are a rational starting point now the future undoubtedly will bring many other candidate genes that today cannot even be imagined given our ignorance of the genes involved in the prenatal development of the central nervous system. Chapter 3 provides an integrative overview of the broad autism phenotype and as such will be of special interest to child psychiatrists. Chapters 5 8 and 10 offer enlightening information on drug and alcohol abuse. Chapter 14 discusses variations in sexuality. Adding balance and mature perspectives on how all the chapters complement and sometimes challenge one another are Chapter 2 written by a major figure in the renaissance of the relevance to psychopathology of both genetics and personality. Chapters 15 17 informed critical appraisals citing concerns and cautions about premature applications of this information in the policy arena and Chapter 18 a judicious contemplation by the editors themselves of this promising and to some alarming field. Clear and meticulously researched this eminently satisfying work is written to introduce the subject to postgraduate students just beginning to develop their research skills to interested psychiatric practitioners and to informed laypersons with some

scientific background

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