The golden age of human genetics in Italy

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Summary – After the Second World War, human genetics in Italy had to cover a gap with respect to the International scientific community. Some older researchers were still tied to the old concepts of human genetics and eugenics, while a new generation of scientists slowly managed to introduce the new concepts and methods of human population genetics. The first part of the paper will discuss the “old school” of human genetics, while subsequent paragraphs will focus on three lines of research: immunogenetics, human medical genetics and human population genetics. These three strands mainly developed respectively in Turin (with Ruggero Ceppellini), Rome and Naples (with Giuseppe Montalenti), and Pavia (where the group headed by Adriano Buzzati-Traverso was). The paper describes the growth and the achievements of this small groups of researchers, characterising three different research lines that rapidly reached a level of international excellence and later became a major part of anthropological studies. Notwithstanding enormous difficulties, these pioneering scientists managed to leave important legacies.

Keywords – Human Genetics, Immunogenetics, Human Population genetics, Thalassaemia, Ruggero Ceppellini, Giuseppe Montalenti, Adriano Buzzati-Traverso, Luigi Luca Cavalli-Sforza.

Introduction

This paper stems from a research project on the history of human and medical genetics in Italy. As it is the first outcome of this attempt to map and describe the development of these disciplines in Italy, it has no aspiration to completeness, nor may it be considered a comprehensive account of how these research fields grew in Italy after the Second World War (WWII). Such a disclaimer is even more necessary given the complexity of the scenario that the historian has to confront with. Human genetics has many different bidirectional connections with a large diversity of disciplines: the vast ensemble that goes under the single name of medicine, for example, but also anthropology, with its different inclinations (biological, physical, and cultural). Finally, it shouldn’t be overlooked that the time span we will consider (roughly, from the 1940s to the 1960s), coincides with the emergence of the so-called “molecular biology”. This large and important set of techniques, theories and concepts deeply affected life sciences and greatly eased the migration of methods, ideas and scientists from one field to another. As a result, human and medical genetics overlap with many other disciplines. It thus confronts the historian with a complex mixture whose borders are extremely blurred, leading to an unavoidably partial and somewhat arbitrary selection of themes and issues.

Eugenics and the Old School

As a starting point, I will take a paper published in 1954 in the British journal Annals of Human Genetics. The title of the paper runs “Data for the study of linkage in man. Microcythaemia and the Lewis-Secretor Characters”. There are four authors: Ida Bianco, Ruggero Ceppellini, Ezio Silvestroni and Marcello Siniscalco. Their academic affiliations are the following: Bianco and Silvestroni are from the Medical Clinic of the University of Rome.
(which at the time was directed by the pathologist Cesare Frugoni), Ruggiero Ceppellini is from the Istituto Sieroterapico Milanese Serafino Belfanti (hereafter ISM), while Siniscalco is attached to the Institute of Genetics of the University of Naples. By considering these factors – authors, affiliations, subject, and journal – we can find hints to most of the key features of the history of human and medical genetics in Italy: its roots, the most important Italian research traditions, and the research fields in which these traditions ventured. Let’s explain this unusual condensation.

The journal in which the work was published is, as noted above, the *Annals of Human Genetics*. This journal was formerly entitled *Annals of Eugenics*, published by the Galton Laboratory of the University College in London. The 1954/1955 issue (volume 19, part I) was the first with the new name, still bearing the old one in brackets under the new line. An Editorial note explained this change on a pure scientific basis, since:

“From the outset, the journal contained many papers dealing with heredity and, in recent years, has consisted almost exclusively of contributions to the science of human genetics. It seems logical to recognize this trend by alteration of the title.” (Editorial Note, 1954)

It is clear however that eugenics was no longer welcome at the high table of science, and that the “scientific treatment of racial problems in man” – as Karl Pearson (1925) put it in the opening words of the first issue of the journal – was to be radically changed after the horrors of Nazi-Fascism.

The same issue confronted Italian human genetics after the fall of the Fascist regime. The whole genetic field (not only its human side) had fallen somewhat short in respect to its concepts and theories, not having kept up the pace of the development of classical genetics and the so called *modern synthesis* (the fusion of Darwinian evolution and Mendelian genetics) (Pogliano, 1999; Mantovani, 2004; Volpone, 2004). Human geneticists had the further burden of racism to get rid of. Eugenics and *racial science* in Italy had in fact a long and worrisome tradition, dating back to the end of the XIX century, that did not disappear in the aftermath of WWII, as clearly shown by recent historical works (Mantovani, 2004; Cassata, 2006a). It is therefore not surprising that the beginning of human genetics in Italy has been traced back to the pre-War period:

“The origin of human and medical genetics in Italy deserves to be credited to the school of Professor Luisa Gianferrari, who had initially taught the course on “General Biology and Zoology including Genetics and Race Biology” since the mid-twenties in the Medical School, University of Milan. (Milani-Comparetti, 1992)”

Luisa Gianferrari’s research, and particularly the one connected to eugenics, has been analyzed by Cassata (2006a). Gianferrari was the founder and first director of the *Study Centre of Human Genetics*, that began its activities in 1940 under the presidency of Luigi Zoja (Gianferrari, 1960). This institution offered genetic counselling for married couples and built up a large archive of clinical data, peaking at some 20000 files. It is remarkable to note that she failed to attract much interest from other geneticists working in Milan at the same time. Neither Adriano Buzzati-Traverso, nor Luigi Luca Cavalli-Sforza – as we will see, two key figures in the development of molecular biology and genetics in Italy – seems to really acknowledge her presence in the field. For example, in both the biography and autobiography recently published, Cavalli-Sforza does not mention her once (Cavalli-Sforza & Cavalli-Sforza, 2005; Stone & Lurquin, 2005). Though Gianferrari did some research in the field of human population genetics, mainly devoted to the study of consanguinity in small isolates (her other major interest being the genetic factors of cancer), Cavalli-Sforza only refers to her in the bibliography of the recent publication of the results of the study of consanguinity and inbreeding in Italy (Cavalli-Sforza *et al.*, 2004), with two entries in the bibliography. It must be duly noted that in the 1940s Cavalli-Sforza was mainly interested in bacterial genetics, and when he turned in human genetics at the beginning of the 1950s he started working with A. Serra, Gianferrari’s former assistant (Milani-Comparetti, 1992). Similarly, Adriano Buzzati-Traverso was mainly dealing with *Drosophila* and radiogenetics. However, it is well
known that Gianferrari was connected to Luigi Gedda, a very controversial self-appointed geneticist in Rome, with strong religious and political bonds to the Vatican and the Democrazia Cristiana. These powerful connections allowed him not only to gain an appointment in medical genetics at the University of Rome, despite the lack of a solid scientific curriculum, but even to open the richly and privately endowed Mendel Institute for the Study of Twins. His views on genetics and eugenics, as well as the web of intrigue leading to his university appointment have been described elsewhere (Cassata, 2006a; 2006b): the main point to be made here is the sharp division that divided the geneticists. This division roughly corresponds to a particular generation gap: while Gianferrari was born in 1890 (and Gedda in 1902), the other geneticists working in Milan where much younger: Buzzati-Traverso born in 1913, Ceppellini in 1917, Cavalli-Sforza in 1922. This gap is strictly connected to the political viewpoints of the two parties – for clear and easily guessed historical reasons. With respect to what happened after WWII, it is well known that Gedda has been the president of the Azione Cattolica (an important catholic activist association) and organiser of the Comitati Civici, right wing associations that greatly helped Democrazia Cristiana (the Christian Democrat Party, that ruled Italy roughly for four decades) in winning the first general elections after the Fascist regime, in 1948. Cassata (2006a) also points out Gedda’s lifelong political involvement, always on the right wing of Democrazia Cristiana, even working in the 1960s for a governmental alliance with the neo-fascist party Movimento Sociale Italiano.

**Ruggero Ceppellini and Immunogenetics**

These three younger scientists, who are key figures in the history of human genetics, were much more connected to Claudio Barigozzi, who became professor of genetics at the University of Milan in 1948, the same year in which Buzzati-Traverso managed to get the chair in Pavia. Cavalli-Sforza, after obtaining his degree, spent some time at the ISM, before going to Cambridge (to Ronald A. Fisher’s lab), and when back in Italy worked again at the ISM, before moving to Pavia, where Buzzati-Traverso managed a well organised and equipped department (founded in 1939 by Carlo Jucci).

Ceppellini – as Buzzati-Traverso a graduate from the University of Milan – was at the ISM too. At the beginning, Ceppellini was mainly an haematologist (working on RH antigens), and turned to genetics only in the first few years of the Fifties, thanks to the acquaintance with Cavalli-Sforza, who introduced him to Barigozzi. He later moved to Turin, where he was appointed professor of medical genetics (Biographical informations about Ruggero Ceppellini are mainly due to personal communications by sir Walter Bodmer and the obituary (1989) he wrote for his good friend. Other sources are Bach (1988) and Van Rood and Amos (1988).

Dealing with Ceppellini we go back to our “guiding” paper. In 1954, working in Milan, he was moving into the field of immunogenetics, which combined his haematological expertise with genetics. Immunogenetics was in fact quite different from what is now understood by this term. It was (and maybe still is) “the art of studying genetically determined polymorphisms with immunological techniques” (van Rood and van Leeuwen, 1990), while now the term is used in a much different way, immunogenetics being the study of the “genetic control of components of the immune system” – as stated on the International Journal of Immunogenetics website (http://www.blackwellpublishing.com/aims.asp?ref=1744-3121&site=1, accessed on June 5th, 2006. Sir Walter Bodmer, in a personal communication, confirmed Van Rood’s view).

In 1954, when the paper was published, Ceppellini was about to leave Italy to go to the Institute for the Study of Human Variation at Columbia University in New York, where he worked with Leslie C. Dunn (founder and first director of the Institute). Together (and with Mario Turri, an extremely skilled technician at the ISM) Ceppellini and Dunn did a significant work on Rh alleles that was the outcome of researches Ceppellini started in Italy, summarised in a book published three years earlier (Ceppellini et al., 1952). As a result, they described one of the first known cases of allelic interaction in trans position (Ceppellini et al., 1955a), a complex issues not fully understood yet.
In the same years, Ceppellini started to develop an interest in population genetics: our exemplary 1954 paper is the first important paper that Ceppellini published in this field. It is important not only for the present historical research, but also because it expanded the concept of linkage studies in human genetics. At the time, linkage (the fact that two genes are carried on the same chromosome) was not thoroughly studied in man (Falk, 2003, 2004), and few blood antigens other than the ABO blood-group system where fully characterised (Bodmer, 1992; Gannett and Griesemer, 2004): the Lewis blood group system was discovered as late as 1946, and the search for linkage between blood group genes and other genes had had little success (Race and Sanger, 1954). For the 1954 paper, Ceppellini typed the blood samples (from 203 families) that were collected in 1952 mainly by Ezio Silvestroni and Ida Bianco in the Po valley around Ferrara, where it was well-known that the heterozygosity for the thalassaemic trait was present at very high frequency, in up to 18% of the population. The linkage between the gene for thalassemia and the ones for Lewis and Secretor resulted to be uncertain.

The linkage and the interactions of Lewis and Secretor characters were thereafter analyzed by Ceppellini himself (together with Marcello Siniscalco (1955)), and a few years later he showed these two genes to be closely interacting in the production of the Lewis b phenotype, by means of a genetic interpretation of the available data (Ceppellini, 1959; Ceppellini et al., 1959).

Linkage was to be a concept of the uttermost importance in Ceppellini’s subsequent career. In fact, his mastery of genetics made him a central character in the development of the genetics of histocompatibility complex in men. First discovered in 1958 – simultaneously by Jean Dausset in Paris, Rose Payne in Stanford and Jon J. van Rood in Leiden – research about human leukocyte antigens (HLA) soon became the ‘next big thing’ in biology and medicine. HLA determines the histocompatibility between two individuals, thus it regulates the acceptance of graft and organ transplantation. It should not surprise that the discovery and the clarification of the genetics of the histocompatibility complex as well as the methods for tissue typing aroused much interest in the scientific community. Ceppellini and Bodmer were the only two trained geneticists in the early community of HLA research; as such they had a pivotal role. The serological data rapidly increased after the 1958 discoveries, and only by using a strong genetic approach was it possible to make sense of them.

Ceppellini played a crucial role in the development of this field. Since 1964, the community of scientists in the HLA field gathered in the so-called International Histocompatibility Workshops (IHW). During these events, the few laboratories working in this area of research met and tested their own methods and techniques against a standard set of cells and antigens. The outcomes of these ‘collective experiments’ were later discussed in long and tiring meetings, in order to standardise lab practices, but also to find the genetic meaning of the data. The first two workshops, which took place in Durham (USA) and in Leiden (Netherlands), mostly dealt with techniques. But as laboratory methods progressed, for years after the first encounter with the leukocyte antigens, it was not clear whether the immune reactions that blood transfusions and skin grafts yielded were controlled by a single genetic system, or by multiple systems. This dilemma somewhat reflected the first acknowledgments of leukocyte antigens: while in Leiden Van Rood tried to frame his data in a system called 4a4b, in Paris Dausset called his system Hu, and Payne and Bodmer simply called their system LA. In 1965, at the second workshop, it was clear that Hu and LA were the same thing, yet it was not known if they were the same system as Van Rood’s 4a4b (Dausset, 1991). The influence of the studies on blood groups, where there are multiple independent systems, was a misleading clue and, as Van Rood himself put it, “the idea that there would be one major histocompatibility complex was non-existent” (van Rood & van Leeuwen, 1990).

In 1967, the third IHW took place in Turin, where Ceppellini was professor of human genetics since 1958. In this workshop, he introduced into international literature the term and the concept of haplotype, which himself coined the year before in a paper published in Italian (Ceppellini, 1967; Ceppellini et al., 1967). The term was introduced
to better specify the concept previously expressed by the word *phenogroup*, and was defined as the set of determinants that an offspring inherits from one parent. The Greek root of the term, *haplo-* , was meant to stress the fact that the haplotype was “not an observed phene and corresponds to the product of a single gene dose.” (Ceppellini et al., 1967) Thus, the haplotype was a real entity, though not observable through the serological tests available at the time. As a matter of fact, it was the result of genetic calculations made possible by the use of family data, a brilliant intuition by Ceppellini himself (Ceppellini et al., 1966). In the 1967 IHW, the panel of cells to be tested belonged to large families from the Turin area. The genetic interpretation of the experimental data lead to one of the most important theoretical breakthrough in the genetics of immune system. Thus, it has been described as “the triumph of Ceppellini” (Kissmeyer-Nielsen and Svejgaard, 1991) the demonstration that the antigens detected by serological reactions were controlled by a single highly polymorphic genetic system, the HLA.

“La grande aventure HLA”, as the subtitle of Dausset’s autobiography runs (1998), and the IHWs may have been the first example of biological big science, gathering scientists from different countries and gathering funds from different national and international institutions. Ceppellini and the Department of Medical of Genetics he directed at the University of Turin, were part of this international network. They got several grants by the Italian National Research Council (nr.11/282/171 and nr.14-216-2); they had a grant from EURATOM (The European Atomic Energy Comunity,an European institution that played an important role in the development of molecular life sciences); since its inception they were contractors for the project “A Collaborative Research study in Transplantation Immunology”, funded by the US National Institute of Allergy and Infectious Diseases (contract nr. PH 43-65-655); finally, Ceppellini was part of the Nomenclature Committee of the World Health Organisation, that was the body in charge of standardising terminology. Ceppellini’s international role allowed him to set up a collaborative network and to exchange students with laboratories around the world, especially with Payne and Bodmer’s lab in Stanford (and later with Bodmer research group in Oxford). Ceppellini was therefore able to found a tradition in immunogenetics and transplantation genetics in Turin, with a number of valuable students spread in Italy and abroad.

**Population genetics and thalassemia investigations**

Ceppellini’s skill in genetics, which proved to be extremely fruitful in the development of immunogenetics, dealing as it did with linkage and family studies, were part of the theoretical toolbox that he developed in the 1950s, working in populations genetics. In this field, Ceppellini developed an important collaboration with Marcello Siniscalco: the two scientists became good friends, and Siniscalco greatly helped Ceppellini in getting pioneering treatments in the USA when the latter was diagnosed with the lung cancer that would eventually kill him.

As we have seen, at the time of 1954 paper we took as a start, Siniscalco was attached to the University of Naples: this immediately lead us to the first Italian chair in Genetics, established in 1940 and occupied by Giuseppe Montalenti, one of the most important life scientists in Italy after WWII, and “the most authoritative Italian geneticist” (Canali & Corbellini, 2006). His interest in human population genetics lead him to a long collaboration with Ezio Silvestroni and Ida Bianco, whose studies on microcythaemia had drawn Montalenti’s attention since the end of the 1940s. Silvestroni and Bianco were conducting extensive population studies on the frequency of the microcythaemic trait in Italian populations, and since their work failed to attract interest in the medical community, they turned to Montalenti looking for help for a genetic interpretation of their data. The history of these studies has been extensively analyzed elsewhere (Bianco Silvestroni, 2002), as well as the history of the “malaria hypothesis” (also known as “Haldane hypothesis”, due to a suggestion made by J.B.S.Haldane at a congress in 1948) – that is, the fact that malaria may have been a major factor in human evolution, shaping gene frequencies according to the selective value of certain genotypes (Canali & Corbellini,
What is more important here is to underline the fact that a collaborative research effort in Italy lead to outstanding results, not only on the strictly scientific side, but also with regard to public health policy. On the scientific side, the genetics and physiology of microcythaemia were explained. Montalenti, his student Siniscalco, and Ceppellini strongly contributed to the understanding of the genetic distributions in different places (Silvestroni et al., 1950), developing new methods for assessing gene frequencies (Ceppellini et al., 1955b) and clearly establishing the relationship between malaria and thalassemia upon experimental studies in Sardinian populations. The latter result was mostly due to the groundbreaking work made by Carcassi, Ceppellini and Pitzus in 1954 and 1955 in four villages: two in a highly malarial zone and two at an altitude of about 1000 metres, where the only malarial cases were imported. The figures they obtained by the study were striking: in the two lowland villages, around 20% of the population carried the microcythaemic trait; in the other two, the frequency dropped to 5% (Carcassi et al., 1957). They also excluded the hypothesis of an ethnic difference between the two villages, by comparing the frequencies of blood groups, that eventually resulted similar.

This line of research was advanced especially by Siniscalco, who tested the “malaria hypothesis” (with positive results) to understand the population genetics of other diseases, particularly G6PD (glucose-6-phosphate dehydrogenase deficiency, commonly known as favism), and for haemoglobin variants (Siniscalco et al., 1961; Siniscalco et al., 1966). It is important to underline that.

“the research carried out by Carcassi, Ceppellini and Pitzus defined the experimental framework within which the Montalenti group was to work during the late Fifties” (Canali & Corbellini, 2006).

On the side of public health policy, the outcome of these intensive years of research was the setting up of a strong effort – mostly due to Silvestroni and Bianco – to develop prevention campaigns in several areas. Together with those later implemented by Antonio Cao in the mid-Seventies, these efforts – by means extensive population screening, genetic counselling for heterozygote couples, and finally prenatal diagnosis – ultimately led to important successes in the struggle to eradicate thalassemias (Cao et al., 1990; Bianco Silvestroni, 2002).

As a matter of fact, the study of haemoglobin proved to be a very fruitful research line in Italy. The young students that entered the field of human genetics in the Sixties made important contributions, and for reason of space we only quote the studies conducted since the end of the Sixties on the adaptations to high altitude in human populations from Peru and Nepal (Morpurgo et al., 1970; Modiano et al., 1972; Morpurgo et al., 1976; Santachiara-Benerecetti et al., 1976). Among the authors of these studies, we find the names of a number of students that attended the proficiency courses in molecular biology held in Pavia at the end of the 1950s (see below): for example, Guido Modiano and Silvana Augusta Santachiara-Benerecetti, that also made interesting investigations on the genetics of Italian populations.

**Pavia’s Connection**

It should be now clear that human population genetics in the 1950s was a fast growing field, with excellent research groups that obtained outstanding results. However, we have hardly mentioned the Pavia group and Cavalli-Sforza’s works. As said, Adriano Buzzati-Traverso was directing the Department of Genetics there since 1948, and was actively involved in the development of genetics and radiogenetics. Though a good scientist, he also exhibited skills as a scientific manager (the detailed history of Buzzati-Traverso’s scientific career has been told in Capocci (2005)). He had a major role in the innovation of Italian life sciences, in two different ways. On one hand he imported the new techniques and approaches of molecular biology. On the other, he tried to set up a new kind of scientific institution, something that was unheard of in Italy. His efforts can be summarized in two important initiatives that took place between 1957 and 1969.

The first one is the two graduate courses on “Biological effects of Hionizing...... radiations”, biennial schools that were meant to be equivalent to an American PhD course. The first one started...
in 1957, the second one in 1959 (while the first official Ph.D. in Italy was established in 1984). The course granted a monthly stipend for students, and had an International faculty. Thus, prospective students had to pass an exam to assess their command of English, and were strongly encouraged to spend a period of time abroad. Buzzati-Traverso was very well connected with American laboratories, such that it was not difficult to find a place for young students. Looking at the names of those who attended the courses, one can find names of the best of the Italian genetics, that is the generation that entered science after WWII.

The other great enterprise undertaken by Buzzati-Traverso, closely linked to the courses – most of the funding came from the same sources: the National Research Council (CNR) and the EURATOM – was the foundation of the International Laboratory of Genetics and Biophysics (ILGB) in Naples. The ILGB opened in the early months of 1962 and was characterised by extreme administrative flexibility – largely unheard in the bureaucratic Italian academy – due to the peculiar agreement that EURATOM and CNR signed. This flexibility allowed a rapid turnover of scientists and staff as well a different resource management, similar to the American model that Buzzati-Traverso was trying to implement. Having been the first director of the Genetics Division of the Scripps Institution of Oceanography in La Jolla, he knew exactly the needs of a modern and successful laboratory in genetics and molecular biology. Buzzati-Traverso served as director until 1969, when he resigned as consequence of a political turmoil. The protest eventually resulted also in the collapse of the Institute itself, whose scientific productivity and international relevance quickly sank (Capocci & Corbellini, 2002).

At the ILGB in Naples, human genetics was not the main focus, though there was a group led by Corrado Baglioni researching protein synthesis. Baglioni, a researcher at the Massachusetts Institute of Technology before moving to the ILGB, was especially known for his research on the genetics and biochemistry of haemoglobins. In particular, he solved the riddle posed by the particular haemoglobin variant called Lepore, which he correctly interpreted as the fusion of two peptide chains d e b on the structural genes coding for the non a-chain of the protein (Baglioni, 1962). Later, at the ILGB a research group for the molecular aspects of G6PD was constituted. The core of human genetics research at the ILGB was actually located in Pavia, where there were two research groups: in vitro mammalian cytogenetics (the group was lead by Luigi De Carli) and human genetics. The latter one was headed by Luigi Luca Cavalli-Sforza. According to the research activities report, the group did not move to Naples because of the large space needed. They had in fact a computer, on which they stored the data for a substantial project that Cavalli-Sforza began in 1951, with different sponsorship (CNR and Rockefeller Foundation). The project was the study of evolution of human populations, which was to become Cavalli-Sforza's major effort for many years. The group in the Fifties (well before the ILGB was founded) started the study of consanguinity and inbreeding in Italy, originally using the Catholic Church's record for consanguineous marriages, particularly for the Parma valley and Sardinia. The research – whose results have been finally published in Cavalli-Sforza et al. (2004) – was meant to be an extensive study on human evolution, and lead Cavalli-Sforza and his group to develop a whole new set of concepts and tools that proved to be extremely useful and soon became widely used. The story of Cavalli-Sforza's researches has been extensively told, by himself as well as by other students (Stone & Lurquin, 2005). However, for our present study it is important to underline how at some point, his group attracted major interest at international level, bringing to Pavia young and brilliant students, such as M. Kimura and A.W.F. Edwards. Cavalli-Sforza was soon to leave Italy, moving to the USA, where his fame has considerably, and deservedly, grown. Yet, he – along with Buzzati-Traverso and his courses – successfully managed to train a group of scientists that still may be considered the backbone of Italian human genetics.

Apart from the study conducted in Italy, in the second half of the Sixties Cavalli-Sforza laid the foundations for another long line of research about the African populations. In 1966 he mounted the first of ten expeditions in twenty years to Central Africa, in order to investigate the group of populations that usually go under the
name of Pygmies. The genetic analysis that Cavalli-Sforza undertook, amidst big difficulties in field work, revealed since its inception interesting evolutionary features of these populations. For example, Cavalli-Sforza observed a fine tuned adaptation to the forest environment where these groups of hunter-gatherers lived. Furthermore, he correlated genetic features of these populations to their characteristic life-style (Cavalli-Sforza, 1971). His studies on the African Pygmies ultimately resulted in a book published in 1986, as well as a series of papers in which he identified common genetic and phenotypic features among the different groups (Cavalli-Sforza, 1986). Later results of this line of research, which has been explored by several Italian students, showed important differences between Pygmies and other sub-Saharan populations. The Pygmies study was possibly the first bit of the Human Genome Diversity Project (HGDP), a massive research programme aiming at the collection of DNA samples from every known population on the planet. HGDP sparked much controversy: it was accused of reviving a sort of scientific racial investigation, as well as of endorsing a scientific colonialism with no benefit for the ethnic groups involved. Eventually, the harsh debate, as well as the competition with the Human Genome Project, considerably slowed down Cavalli-Sforza's initiative.

In the 1970s, Cavalli-Sforza (who in 1971 officially moved to Stanford University) also pursued another line of research – possibly his most famous contribution: the conjugation of studies in cultural and genetic evolution. This eventually lead him to a fruitful collaboration with Marcus W. Feldman in order to assess a quantitative analysis of cultural evolution (Cavalli-Sforza & Feldman, 1981), and with the archaeologist Albert Ammerman. They elaborated the so-called “demic” model of cultural diffusion similar to the spreading of novel genetic traits, and successfully mapped genetic and archaeological evidences for the Neolithic revolution (Ammerman & Cavalli-Sforza, 1984). Cavalli-Sforza further explored this field, correlating genetics to linguistics and to other cultural features, such as family names. He therefore managed to join many different disciplines, overcoming the boundaries that anthropological sciences showed for a long time: a fragmentation that Italian community suffered for long time. As a matter of fact, probably due to the peculiar history of the anthropologists in Italy (most of them were committed to racist theories), human genetics and anthropology did not communicate much, and also physical anthropology went through a long period of relative difficulty – both theoretical and institutional.

Conclusions

Though necessarily brief, this overview of the early history of human and medical genetics raises a considerable amount of historiographical issues. For example, the relative lack of interaction between human genetics and medical genetics, shown for example by the difficulties that Silvestroni and Bianco encountered in their work. This issue also brings to face another question, that is now under the scrutiny of historians, i.e. the large gap that divided the two big factions: Gedda’s side vs. the “new” geneticists – Montalenti, Ceppellini, Buzzati-Traverso. These questions also address another and possibly more fundamental issue: the progressive decline of Italian life sciences that resulted in the loss of international relevance as well as of cultural importance in Italian society. Of course, there were still some very good laboratories, and research did not stop producing good results. Yet, there is the widespread feeling that Italy in the Seventies moved away from the centre to the periphery of the relevant scientific networks. Furthermore, it is widely acknowledged that Italy failed to develop an efficient R&D system despite a series of favourable conditions, such as the emergence of a small group of entrepreneurs, politicians and scientists keen to foster the diffusion and the enhancement of science and technology. This paper is not intended to address this question. Still, what we may infer from the story told here is the existence of large and lively scientific community that nonetheless failed to acquire a “critical mass” in terms of academic power and political influence, as shown by the systematic and constant mistreatment that life sciences in Italy have suffered for many decades. This is also true for the whole Italian scientific establishment, though
life sciences (and particularly those connected to evolutionary thinking) lacked the long tradition of other disciplines (especially physics). More over, Italian biology did not manage to get involved with an active participation and large financial commitment by the government in international projects (the Human Genome Project being probably the last example).

Future historical research, in order to face these complex issues, has to consider at least two different directions: some have already been partly explored. On the one side, there are the private and personal aspects of the people involved. Also as a consequence of a peculiar academic system that did not encourage cooperation and hampered the efforts to create efficient institutions, many important scientists ultimately emigrated (Cavalli-Sforza, Buzzati-Traverso, Ceppellini, Siniscalco among others), and even they were in Italy they failed to act as a whole: non-communicating vessels in terms of academic management and political influence.

On the other side, the political aspects lie. The polarization and harsh struggle between Democrazia Cristiana and the Communist Party somewhat involved science and culture not only in term of ideas and styles of thought, but also regarding academic power, institutional management and low-level political manoeuvring in scientific institutions. As a result, very few people thought and acted for the good of science and the diffusion of scientific culture. Furthermore, most of Italian politicians were trained in humanities, thinking science to be extraneous to the cultural high table, and a luxury that the country could not afford. Italian science still suffers the results of this apparently chronic inability of politicians to grasp the importance of science.

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Info on the web

http://www.igb.cnr.it/abt/
A website devoted to Adriano Buzzati-Traverso.

http://lgxserver.uniba.it/lei/storiasc/diffusione/genetica_italia/SpecGenIt.htm
A special issue of the “Quaderno Swif di Storia della Scienza” dedicated to the “Paths of Italian Genetics”.

http://library.cshl.edu/
Cold Spring Harbor Laboratory Library maintain a huge archive in the History of Genetics and Molecular Biology, including James D. Watson Collection and the Cold Spring Harbor Symposia.

http://agi.unipr.it/
The website of the Italian Genetic Society (Associazione Genetica Italiana).

http://hpgd.stanford.edu/
It provides information on the Human Genome Diversity Project.

References


Ceppellini R., Tecilazich F. & Nasso S. 1952. La
malattia emolitica del neonato. Istituto Sieroterapico Milanese Serafino Belfanti, Milano.


