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Human Genetics with Global Aspirations: Inventing Community Genetics within and beyond the World Health Organization (1960s–2000s)

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SUMMARY: The lack of investment in noninfectious diseases by international health organizations after World War II is an understudied topic. By examining the global trajectory of hereditary and congenital disorders within and beyond the WHO, the authors provide insight into the reasons for this failure to invest in noncommunicable diseases management. In the 1970s, a network of geneticists, physicians, and WHO officials aimed to address the most frequent hereditary disorders, notably thalassemia, by putting them on the organization's agenda. However, despite significant epidemiological stakes, community genetics did not expand globally. The paper examines how Global South instantiations have reshaped aspirations for Southern alternatives to medical genetics as it had developed in the Global North. It also emphasizes the importance of analyzing new discursive activities in the field of global health and the characteristics and practical implications of these global aspirations, such as program funding, design, and operation.

KEYWORDS: community genetics, World Health Organization, noncommunicable diseases, global health, international health, thalassemia

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In 2003, *Gulf News* reported the visit in the Sultanate of Oman of a "world authority on genetic disease control," Bernadette Modell, a British clinical geneticist and World Health Organization (WHO) consultant.[1] Invited by the Ministry of Health to assess the needs for genetic services, Modell acknowledged the burden of congenital and genetic disorders in Oman and recommended integrating their management into primary health care. This expert mission exemplifies the activities of a WHO-funded network of clinical geneticists. Throughout over the final decades of the twentieth century, they advocated for genetic disorders not to be siloed by WHO's typical vertical programs, since genetics "has a contribution in everything."[2]

The historiography of the WHO has granted a central role to the differentiation between vertical and horizontal interventions.[3] This narrative presents the vertical programs that dominated international public health in the 1950s and 1960s as symbol and problematic instantiations of a fascination for technical fixes, which would permit to eliminate or control specific diseases. However, most of these programs were overly simplistic and reductionist, resulting in mixed outcomes, with few exceptions. In addition to this flawed understanding of the relations between health and technologies, most of the vertical programs implemented in

[1] Bernadette Modell and Matthew Darlison, interview by Jean-Paul Gaudillière and Claire Beaudevin, London, November 2016; Sunil K. Vaidya, "Congenital Anomalies High in Oman," *Gulf News*, February 18, 2003.
[2] Bernadette Modell and Matthew Darlison, interview by Lucile Ruault, London, June 2019.
[3] Anne-Emanuelle Birn, "The Stages of International (Global) Health: Histories of Success or Successes of History?," *Global Pub. Health* 4, no. 1 (2009): 50–68; Marcos Cueto, Theodore M. Brown, and Elizabeth Fee, *The World Health Organization: A History* (Cambridge: Cambridge University Press, 2019); Randall M. Packard, *A History of Global Health: Interventions into the Lives of Other Peoples* (Baltimore: Johns Hopkins University Press, 2016); Jean-Paul Gaudillière, Claire Beaudevin, Christoph Gradmann, Anne M. Lovell, Laurent Pordié, eds., *Global Health and the New World Order* (Manchester: Manchester University Press, 2020).

the Global South were designed and funded in the North, leading to significant criticism in the context of decolonization and the end of European empires.

These views resulted in a change of policy for the WHO in the late 1970s. The organization committed to the primary health care strategy—considered horizontal in several respects. First, it aimed to link health policies with development and planning, advocating for instance for coordination with agricultural or educational investments. Second, the strategy highlighted the role of rural communities as the most vulnerable and those whose needs should prevail when defining policy targets and the local organization of services. Finally, the strategy provided an alternative to hospital-centered biomedical interventions based on costly and fragile technologies by instead advocating for the creation of integrative local centers, the training of nonmedical personnel, and the use of "socially appropriate" techniques, including traditional healing practices of recognized efficacy.

Historians working on international health after World War II agree that investments and interests in noninfectious diseases have been weak from all parties involved—WHO, providers of health-related aid, or nation-states of the Global South. Despite predictions that Latin America, Asia, and Africa would go through a similar demographic transition as European and North American countries, noncommunicable diseases (NCDs) in these regions became a matter of international concern only in the 1990s. At that time, the Global Burden of Diseases, a new metric developed by the WHO and the World Bank, began to make their rising incidence and impact visible. Historians are aware of the problem, but they have yet to fully investigate the mechanisms of NCDs invisibility to entrepreneurs of health with global aspirations.[4]

[4] E.g., Cueto, Brown, and Fee, *World Health Organization* (n. 3); and Packard, *History of Global Health* (n. 3).

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This paper contributes to filling these gaps and adds to the scholarship on genetics in biomedicine, with a history of community genetics within and beyond the WHO.[5] We trace the winding road taken by a network of geneticists, physicians, and WHO officials, among whom Bernadette Modell played a leading role. This network began working in the 1970s to put the most frequent hereditary disorders on the scientific and political agenda of international public health. In doing so, they aimed to enable the design and implementation of national programs targeting major congenital disorders in the Global South, notably thalassemia, but also sickle cell anemia and spina bifida.[6] We approach community genetics (and its strong focus on thalassemia) as an unusual register of international public health practices that stopped dealing with diseases in silos, by designing the control of a specific group of NCDs as part of the primary health care strategy.

The trajectory of community genetics on an international scale can indeed be traced back to the 1980s. A small group of physicians (mainly British at first) were involved in the control of thalassemia (the most prevalent group of hereditary anemias) in Cyprus. They were concerned about the incidence of inherited blood disorders. The clinical geneticists built up a global program based on their successful experience with population-based services for thalassemia. This model was deemed relatively simple and transferrable for integrating the control of genetic diseases into community health care. In the 1990s, they lobbied the WHO

[5] Our article complements research on the history of globalized genetic research, the importance of population approaches, and the role of WHO in the science of heredity from the Cold War era onward, such as Joanna Radin, *Life on Ice: A History of New Uses for Cold Blood* (Chicago: University of Chicago Press, 2017); Jenny Bangham, *Blood Relations: Transfusion and the Making of Human Genetics* (Chicago: University of Chicago Press, 2020); Soraya de Chadarevian, *Heredity under the Microscope: Chromosomes and the Study of the Human Genome* (Chicago: University of Chicago Press, 2020); Elise K. Burton, *Genetic Crossroads: The Middle East and the Science of Human Heredity* (Stanford, Calif.: Stanford University Press, 2021).

[6] Congenital disorders are diseases present at birth including genetic diseases (caused by DNA abnormalities), which may or may not be inherited.

to develop an approach they called "community genetics,"[7] which they considered a new mode of intervention in social and preventive medicine. Contiguous with medical genetics and public health, community genetics, as framed in collaboration with WHO officials, was meant to be part of primary health care worldwide. The physicians involved in this international group of experts were supported for years by the WHO's headquarters in Geneva and by several of its regional offices. The WHO coordinated and funded the expert network and scoped the activities they carried out.

In this paper, we consider community genetics a hybrid initiative: a seemingly vertical program, it was deeply rooted in technology (including routine genetic testing) and apparently defined by biomedicine (genetic medicine). It was nonetheless conceptualized as a part of primary health care and therefore as a form of social medicine. Hence, our exploration of community genetics as a specific territory shared by the worlds of biomedicine and primary health care. As a boundary object to these two worlds, its coherence (and thus its use by the network of experts) rested on a lack of consensus about the respective role of technology and social intervention in its constitution.[8]

[7] Bernadette Modell and Anver Kuliev, "The History of Community Genetics: The Contribution of the Haemoglobin Disorders," *Commun. Genet.* 1 (1998): 3–11. Two additional aspects about the term are noteworthy: first, it was in use at least since the 1970s, for smaller scale interventions, beginning with Tulsa's Children's Medical Center (J. G. Coldwell, B. Say, and K. Jones, "Community Genetics 1," *J. Okla. State Med. Assoc.* 68, no. 8 [1975]: 299–302). After Modell's team, several physicians at the center of genetic counseling development have come up with another definition of community genetics (L. P. Ten Kate, L. Al-Gazali, S. Anand, A. Bittles, J. J. Cassiman, A. Christianson, M. C. Cornel, H. Hamamy, H. Kääriäinen, U. Kristoffersson, D. Marais, V. B. Penchaszadeh, P. Rahman, and J. Schmidtke, "Community Genetics: Its Definition 2010," *J. Commun. Genet.* 1, no. 1 [2010]: 19–22). With a clear distancing from public health, their definition focuses on the challenges of genetic counseling and the individual autonomy of patients and families.

[8] Susan Leigh Star and James Griesemer, "Institutional Ecology, 'Translations' and Boundary Objects: Amateurs and Professionals in Berkeley's Museum of Vertebrate Zoology, 1907–39," *Soc. Stud. Sci*. 19, no. 3 (1989): 387–420.

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Despite the number of lives at stake, community genetics has not been globally expanded, nor has it become the target of WHO-coordinated programs funded by philanthropic global health actors (wealthy states, individuals, or nongovernmental organizations). Instead, it has taken shape in numerous local, and a few national, initiatives aimed at improving access to genetic medicine, with a clear regional focus on areas affected by hemoglobinopathies. Accordingly, we document the development of community genetics as a programmatic discourse with global aspirations in Geneva. Additionally, we explore the London and Cyprus origins of community genetics and the shape it took in the Middle East and South Africa by following the activities of Modell and her closest associates. This paper shows how local initiatives reshaped global aspirations for Southern alternatives to medical genetics and genetic counseling as they had developed in Europe and North America. The text follows two distinct but interconnected lines of inquiry: the diverse set of activities related to human genetics initiated by the WHO, and the medical and political work carried out by participants in the community genetics network. Accordingly, our aim is to connect two bodies of academic work: the study of the WHO's development and the rise of global health in the late twentieth century; and research on genetics and its unique place in the history of biomedicine.

The paper draws on a wider collective project on the emergence of global health, and the forms of knowledge, medical practices, and policies it has fostered since the 1990s.[9] Our community genetics inquiry is a historical and anthropological endeavor. We base our analyses on the following corpus: collective research in the archives of WHO's Geneva headquarters, including an extensive analysis of the WHO's programmatic reports and regulatory documents about genetics, during the period 1970 to 2010; oral history interviews

[9] Globhealth, https://globhealth.cnrs.fr.

we conducted together with a dozen professionals who were involved in the community genetics network (including three interviews with Modell); and ethnographic fieldwork in Oman (2005–18) focusing on the clinical handling of hereditary blood disorders and the implementation of genetic services.

Bridging several sites and political contexts from the 1960s to the 2000s, we analyze the implementation of genetic interventions (individual screening, preconception counseling, medical abortion, and care for children born with congenital diseases) by various institutional and professional actors, both within and outside WHO interventions.

The first section takes us to the 1960s in Cyprus and the United Kingdom, following Modell and her colleagues as they design care protocols and improve diagnostic techniques for the community of Cypriot patients affected by thalassemia in London. We then trace how during the 1970s and until the mid-1980s a WHO network of practitioners grew out of the design of the Cypriot national strategy for thalassemia control, partly made possible by these improved diagnosis techniques. The resulting drop in the incidence of thalassemia in Cyprus made the country a pilot site (and later an ideal site) for the approach that was to become community genetics.

The second section covers the period from 1978 to 1991 and the development of a global network of genetics experts under the WHO's Human Genetics Programme. Originally tasked with the "community control of hereditary anemias," the network expanded its scope to hereditary diseases as a general category. It gradually defined its approach and named it "community genetics" in 1990. The section also shows the network seeking support from the WHO's Middle Eastern office, when facing reluctance in Europe and WHO's central offices regarding the integration of genetics with public health goals and primary health care.

The third section of the paper discusses the development of community genetics outside of the WHO's purview. It begins by examining the case of the Sultanate of Oman.

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During the mid-1990s to the early 2000s, the country's oil and gas revenue and international connections enabled the development of genetic services and an attempt at nationally tackling hereditary anemias. We then move to another country example, South Africa in the 1990s, where the main emphasis was on care (before prevention). This resulted in the integration of community genetics with maternal and child health. The paper concludes by showing how this "clinical" turn of community genetics allowed, in the late 1990s and early 2000s, for the involvement of the March of Dimes, which worked along the WHO network of experts to transform "birth defects" into a target category of global health.

Modell, the Thalassemia Network, and Cyprus as a Reference Case

The high prevalence of thalassemia in Cyprus was described after World War II,[10] and Cypriot authorities declared it a major sociomedical issue in the early 1960s. About one out of seven Cypriots was a carrier of beta-thalassemia, and one out of a thousand was homozygous—a condition leading to chronic anemia, slow infant growth, bone deformities, and early death in its severe forms. In 1971, Cyprus sought WHO support.

A Clinical Approach to the Emerging Thalassemia Community in London (1960–80)

In the early 1960s, Modell, a recently qualified physician (born in 1935) and pediatric house officer at University College London Hospital, realized the epidemiological burden affecting ethnic minorities in the city. At this time, the emergence of beta-thalassemia in North London coincided with a period of large-scale migration from Cyprus: after World War II, up to 1966, about 100,000 Cypriots settled in Britain.[11]

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[10] Alan Fawdry, "Erythroblastic Anaemia of Childhood (Cooley's Anaemia) in Cyprus," *Lancet* 243, no. 6284 (1944): 171–76.

[11] Verity Saifullah Khan, ed., *Minority Families in Britain: Support and Stress* (London: Macmillan, 1979). The two communities were segregated in London (Greek Cypriots being predominantly in Camden, which could explain the strong link with Modell's team); cf. Robert Winder, *Bloody*

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Modell received a research grant around 1964 and focused on thalassemia, partly because of her proximity to the Cypriot community. She notes that it was "a disorder of the control of [hemoglobin] protein synthesis rather than a structural disorder. And that was the mystery at the time" in molecular biology, thereby providing a stimulating research topic.[12] Moreover, she felt that "this sample [of a common disease from the (sub)tropics] brought into our country, gave [Northern medicine] the opportunity and the privilege to use our scientific resources to develop treatment and prevention" for those regions.[13]

She began her involvement in the clinic where she worked, focusing on designing timely and appropriate care. Her initial strategy involved transfusing multiple units of red blood cells to increase hemoglobin levels, similar to Irving Wolman's approach at the Children's Hospital of Philadelphia. This treatment, combined with advancements in the use of iron-chelating agents,[14] led to a significant reduction in thalassemia complications in the late 1970s. As these advances in treatments became more widely known, Modell's team had more contacts with the Cypriot community. By 1965–67, she "got used to the fact that parents of the children would ring [her] . . . , terribly anxious, asking to arrange an early termination of pregnancy,"[15] or to perform a tubal ligation.

Modell's second move was to measure the disease's burden. Surveying "all the hospitals in London," her team recorded the thalassemia patients while gaining access to the registry of births, deaths, and marriages. "Pick[ing] out all the Cypriot names" and comparing the number of births and of affected children admitted to hospitals, they showed the incidence

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Foreigners: The Story of Immigration to Britain (London: Abacus, 2004). However, we lack information about the proportions of Greek-Cypriots and Turkish-Cypriots in Modell's patients.

[12] Modell and Darlison interview, June 2019 (n. 2).

[13] Bernadette Modell, interview by Peter Harper, December 2007.

[14] To maintain the hemoglobin level, thalassemia treatment includes (bi)weekly blood transfusions and the daily use of infusion pumps to excrete excess iron resulting from these transfusions.

[15] Modell and Darlison interview, June 2019 (n. 2).

of beta-thalassemia was 15 to 18 percent within the Cypriot community. This important shift from affected families to the community entailed the development of heterozygote detection and prospective counseling for thalassemia among Cypriots in the United Kingdom. This approach aimed to make patients of reproductive age aware of their heterozygote status, rather than discovering it incidentally through their child's diagnosis.

Faced with the anxiety of pregnant couples, Modell and her colleagues soon joined the ongoing efforts in the United States and Italy (especially Sardinia) for the development of prenatal screening for thalassemia.[16] Between 1975 and 1979, this led to a new phase of coordinated research with the Boston Children's Hospital / Harvard team of Blanche Alter, Andrée-Marie Dozy, and Yuet-W. Kan, to whom Modell shipped some "at-risk" embryonic material from three London obstetric units for cell analysis. Under pressure from a pregnant patient who refused to give birth to another affected child, Modell eventually began collaborating with obstetrician Denys Fairweather to conduct prenatal diagnosis: he performed "blind amniocenteses" to sample fetal blood. Thus, prenatal diagnosis of thalassemia started in Britain in 1975 (abortion was legal since 1967), long before Modell's team published their results in 1980.

Shortly after, several researchers, notably Kan (United States) and Antonio Cao (Italy), developed a DNA technology for diagnosing thalassemia in the early 1980s. They used what came to be known as chorionic villus sampling (testing for genetic abnormalities a sample of tissue removed from the placenta). Employing these innovations, as well as her own "Portex cannula" technique, Modell worked with research teams (such as the one led by

[16] Cf. Y. W. Kan, M. S. Golbus, R. Trecartin, M. Furbetta, and A. Cao, "Prenatal Diagnosis of Homozygous β-Thalassæmia," *Lancet* 306, no. 7939 (1975): 790–91; H. H. Kazazian Jr., M. M. Kaback, A. P. Woodhead, C. O. Leonard, and W. S. Nersesian, "Further Studies on the Antenatal Detection of Sickle Cell Anemia and Other Hemoglobinopathies," *Adv. Exp. Med. Biol*. 28 (1972): 337–46.

molecular biologist Robert Williamson) in investigating the genetic cause of thalassemia. In the early 1980s, her team developed a first-trimester diagnosis, enabling earlier therapeutic abortions of affected fetuses. From 1981 onward, the University College London Hospital Department of Obstetrics received continued support from the WHO hereditary diseases program for research on early fetal diagnosis and hemoglobinopathy control.

Through these collaborative efforts, Modell built a research and clinical network focused on hemoglobin disorders. Her approach to prevention gradually embraced the notion of community, making the network even more influential. As soon as 1972, Modell's team had concluded that "thalassaemia trait occurs with the same high frequency among Cypriots in London as it does in Cyprus. Because of the 14% incidence of beta-thalassemia trait the risk to all Cypriots of having children with thalassemia major is considerable."[17] Her awareness of targeting a *community issue* grew steadily,[18] as her counseling activity expanded among the Cypriots:

> Once we had the prenatal, . . . the news spread in Cyprus, . . . they started getting on planes and coming over here. I remember walking in the street, . . . crossing the road to the obstetric hospital, this Mediterranean-looking woman stopped me and said, "Where's the thalassemia service?" I said, "It's me." She had just got off the plane."

[17] B. Modell, A. Benson, and C. R. Payling-Wright, "Incidence of Beta-Thalassemia Trait among Cypriots in London," *BMJ* 3 (1972): 737. This situation follows the course of epidemiological events Rajtar and Knoll frame as "the 'endemic' versus 'rare' disease distinction [being] challenged by global migration" (Małgorzata Rajtar and Eva-Maria Knoll, "Anthropology, Global Health and Rare Diseases," in *Routledge Handbook of Anthropology and Global Health*, ed. Tsitsi B. Masvawure and Ellen E. Folley [New York: Routledge, 2024], 353–65).

[18] As shown also in a clinical publication of 1977, where she states, "In all communities, prevention by population screening, antenatal diagnosis, and selective abortion seems the long-term solution, and this will probably become a possibility in the near future" (B. Modell, "Total Management of Thalassaemia Major," *Arch. Dis. Child.* 52 [1977]: 489–500).

The association of prenatal diagnosis with the option of termination of pregnancies thus created a demand for fetal monitoring among the Cypriot community—all the more so with the creation of the U.K. Thalassaemia Society in 1976.

Cyprus and the Globalization of Thalassemia Control (1971–84)

Population screening already existed in Cyprus. Based on the demonstration by a WHO consultant that necessary care for the increasing number of children surviving with thalassemia would jeopardize national health resources, WHO recommendations led to the approval of a five-year plan in 1971.[19] In addition to extensive efforts to improve curative services,[20] the Cypriot government implemented a control program to prevent further homozygote births, which involved public education, genetic counseling, and population screening.[21] Its first line of attack consisted of reminders circulated through the mass media, introduction of the topic into school curricula, ad hoc talks in rural community centers, circulation of booklets in most endemic areas, and voluntary blood donation campaigns. From 1972 to 1976, other prongs of the program sought to discourage marriages between carriers.[22] Population screening and counseling were thus geared to homozygotes' relatives, young single people, and volunteers.[23] This latter option, however, proved partially unsuccessful

[19] T. Ashiotis, Z. Zachariadis, K. Sofroniadou, D. Loukopoulos, and G. Stamatoyannopoulos, "Thalassaemia in Cyprus," *BMJ* 2 (1973): 38–41.

[20] The efforts included the creation of thalassemia outpatient clinics, specialization of some pediatricians, improved blood transfusion, supply of iron chelator, and portable infusion pumps.

[21] Ruth Schwartz Cowan, *Heredity and Hope: The Case for Genetic Screening* (Cambridge, Mass.: Harvard University Press, 2008).

[22] Michael A. Angastiniotis, Sophia Kyriakidou, and Minas G. Hadjiminas, "How Thalassaemia Was Controlled in Cyprus," *World Health Forum* 7 (1986): 291–97. Our corpus does not mention the partition. Modell and her team collaborated with the Greek side, through national institutions in Nicosia. The fact that the *Pan*Cyprian AntiAnemia Association was among the first contacts of Modell points to the inclusion of Turkish Cypriots. However, there is no explicit account of this, and further investigation is needed.

[23] Michael A. Angastiniotis, "Factors Limiting the Effective Delivery of Fetal Diagnosis for Thalassaemia, May 2–4, 1984" (WHO/SERONO Meeting on Perspectives in Fetal Diagnosis of

since people often concealed their heterozygote status for fear of social stigma and rejection from the marriage market.

The Pancyprian Anti-Anemia Association, a parent-support association founded in the mid-1960s,[24] established close bonds with Modell around 1973—when the military involvement in clinical management of thalassemia for both Greek and Turk inhabitants was threatened by the Turkish invasion[25]—and invited her to present recent advances in cure and prevention. In 1976, the BBC documentary *Sea in the Blood* (literal translation of "thalassemia") was aired, serving as favored information media in the United Kingdom and in Cyprus.[26] In addition, the association pushed Modell to meet with the Cypriot minister of health, with whom she negotiated better standards of treatment, including imported portable lightweight pumps for iron-chelating infusion therapy. Starting in 1977, the general approach to prevention shifted significantly under the influence of technical changes taking place in Great Britain. Families themselves, starting with pregnant women, connected with London medical teams by using the newly available fetal diagnosis. A few Cypriot clinicians also sent patients to London, Athens, and to a lesser extent Jerusalem.[27] At the same time, the circulation of information within the Cypriot community at large aroused Modell's interest in the management and control of the disease in Cyprus. With a grant, she visited Cyprus and the pediatrician Michael Angastiniotis, with whom she met patients who explained "the situation

Hereditary Diseases, Archbishop Makarios, Thalassemia Centre, Nicosia, Cyprus, Geneva, WHO Archives, G3-181-129, 1984).

[24] Theodoros Kyriakides, "Tactics as Empirical and Conceptual Objects: Patient Activism and the Politics of Thalassaemia in Cyprus," *ESTS* 2 (2016): 13–32.

[25] Our information about events in London before the partition does not reveal tensions, e.g., the meeting aimed at encouraging the creation of a patients' association in the late 1960s was introduced successively in Greek and Turkish.

[26] *My Children, My Children: Sea in the Blood* (BBC, 1976).

[27] Michael A. Angastiniotis and Minas G. Hadjiminas, "Prevention of Thalassaemia in Cyprus," *Lancet* 14 (1981): 369–71.

about how the parents spent their life on their knees begging people to give blood for their child."[28]

The first team trained in care delivery, epidemiology, and research for thalassemia in the London Galton laboratory comprised an obstetrician and Angastiniotis, who was to become a key actor in developing the thalassemia program in Cyprus in the 1980s and 1990s, particularly by introducing prenatal diagnosis. Other researchers completed their doctorates under Modell's supervision and specialized in hemoglobinopathies in the late 1980s.

This new knowledge and technology influenced the architecture of the Cypriot prevention policy. In the late 1970s, the advent of prenatal diagnosis shifted the target group of screening from young single people to pregnant women and couples planning to conceive. On the one hand, the new policy requested "antenatal clinics . . . to demand screening from the first visit"; on the other hand, it implied that "other married and engaged couples were encouraged to come while singles were discouraged."[29] The availability and general acceptance of fetal diagnosis in Cyprus encouraged the shift in screening toward people with immediate risk of transmitting thalassemia. However, in 1983, the strategy "[returned] to the screening of single people" in addition to prenatal screening.[30] Indeed, the Cypriot Christian-Orthodox Church introduced a premarital certificate requesting couples, who sought religious blessing for their engagement, to undergo testing for thalassemia trait.[31] Hopes of reducing intermarriage between carriers have proven illusory, but this compulsory policy meant that reproductive-age people would not neglect the test, so that carrier couples would be counseled early and decide whether to conceive and use prenatal screening. The changing architecture of

[28] Modell and Darlison interview, June 2019 (n. 2).
[29] Modell and Darlison interview, November 2016 (n. 1).
[30] Angastiniotis, Kyriakidou, and Hadjiminas, "How Thalassaemia Was Controlled in Cyprus" (n. 22).
[31] Modell and Darlison interview, June 2019 (n. 2).

the Cypriot control program thus shows how the British and Cypriot clinical and research teams endorsed a combination of screening, genetic counseling, prenatal diagnosis, and termination of pregnancies.

Between the early 1970s and 1982 in Cyprus, thalassemia prevalence at birth fell by 90 percent, resulting in what we call the Cypriot archetype of eradication of thalassemia.[32] International experts soon considered it a success story and concluded that thalassemia was the first genetic disease to be controlled, making it a prime example for global projections regarding the control of congenital diseases.

The first framing of community genetics by Modell and her colleagues had thus been achieved within the context of British and European debates about medical genetics, its practices and its future. It was dominated by the idea of advancing a public-health-oriented form of clinical genetics. She also rapidly assumed that "the pattern of management recommended here for Britain is impracticable in many countries in the Mediterranean area, the Middle and the Far East, where thalassaemia is a major public health problem. . . . However, in all communities, prevention by population screening, antenatal diagnosis, and selective abortion seems the long-term solution."[33] Therefore, Modell's understanding of the Cyprus experience was critical: it considered the community to be the entire population of Cyprus because of the high incidence of thalassemia; it framed a preventive intervention as a vertical initiative implemented by physicians within clinical genetics and fetal medicine

[32] WHO Hereditary Diseases Programme, Division of Non-communicable Diseases, Report of the 2nd Annual Meeting of the WHO Working Group on the Community Control of Hereditary Anaemias, Archbishop Makarios Thalassaemia Centre, Cyprus, November 29–31, 1983, WHO Archives, G3-181-129.

[33] Modell, "Total Management of Thalassaemia Major" (n. 18), 498.

units;[34] even if it included social technologies like counseling and certificates, it relied upon the progress in obstetrical ultrasound, the access to protocols including biochemical tests, fetal sampling methods, and abortion procedures, all technologies that were central to the disciplinary rise of medical genetics in Europe. It prioritized prevention over care, not the least because progress in the latter implied fewer premature deaths and increasing costs for the health system.[35]

The Emergence of Community Genetics as a WHO Target (1978–91)

The WHO's interest in genetics started in the mid-1950s when assessment of the effects of atomic radiation on human heredity began to incorporate study of genes. Apart from works on "vanishing" groups, the WHO supported collaborative research on the "frequency of diseases with a genetic component" in order to establish a complete hemoglobin nomenclature,[36] but a significant shift happened in the late 1970s, when community genetics became a key category for the network arguing for a major WHO initiative.

Birth of a WHO Network of Experts

In 1978, Robert Williamson organized a workshop on thalassemia molecular genetics in Crete, including "young people from the whole Mediterranean area [so as to] open their eyes

[34] "Prenatal Diagnosis and Genetic Screening, Community and Service Implications, Summary and Recommendations of a Report of the Royal College of Physicians," *J. Roy. Coll. Physicians London* 23, no. 4 (1989): 215–20.

[35] The 1983 HGP meeting memorandum included a cost-benefit analysis of thalassemia programs in Italy and Cyprus. The memorandum explained that within twenty years about 40 percent of blood donors in Cyprus might have to donate blood annually for thalassemia treatment alone and the total medical budget would double. In 1977, the government of Cyprus arranged for training of an antenatal diagnosis team due to a similar projection by a WHO consultant. (A. Boyo et al., "Community Control of Hereditary Anaemias: Memorandum from a WHO Meeting," *Bull. World Health Organ.* 62 [1983]: 63–80, quotation on 66).

[36] Chadarevian, *Heredity under the Microscope* (n. 5).

to these future possibilities,"[37] notably prenatal testing and the transition from fetal blood test to chorionic villus sampling. In June 1981 in Sardinia, pediatrician Antonio Cao organized another conference,[38] where some of these experts in thalassemia "[discussed] the desirability of a WHO program for the control of hemoglobinopathies."[39] These discussions provided Anver Kuliev with a basis for the "substantial reorientation" of WHO activities about genetics,[40] since prevention on a population scale "by heterozygote detection, prospective genetic counseling, antenatal diagnosis and selective abortion of affected foetuses" had proved its reliability (thalassemia control programs had indeed started in the 1970s in Cyprus and Sardinia, but without WHO involvement).[41] Kuliev, a young Soviet geneticist recently appointed head of the WHO Human Genetics Programme (HGP), had started to work at WHO as a consultant in 1979, when the preexisting Genetics Unit at WHO was coming to a crisis point due to internal and external criticism. Kuliev remembers that the director-general of the WHO "challenged [him], as a scientist in genetics, to demonstrate whether genetics was really important for public health and WHO."[42]

To rehabilitate medical genetics into an institution whose official strategy was by then primary health care, Kuliev aimed at extracting genetics from its aura of "pure" research.

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[37] Angastiniotis, "Factors Limiting the Effective Delivery of Fetal Diagnosis for Thalassaemia" (n. 23).

[38] Anver Kuliev, interview by Lucile Ruault, Chicago, December 2018. The cofounder of the Athens Centre for the Prevention of Mediterranean Anaemia, Dimitris Loukopoulos, Thai hematologist Prawase Wasi, and U.S. hematologist Arthur Nienhuis joined Williamson and Modell.

[39] Robert Williamson, interview by Lucile Ruault and Jean-Paul Gaudillière, Paris, January 2019.

[40] Memorandum from Kuliev to Director-General, May 15, 1981, "Proposal for Task Group Meeting on HMG Programme," WHO Archives, G3-87-5.

[41] "Minutes of a Meeting Held in Cagliari, Sardinia on 9.6.1981 in Association with the International Congress on Recent Advances in Thalassaemia," 2 (Williamson's archives). "In 1977–79 the birth-rate of affected children has fallen by 60% in Southern Sardinia, 70% in Southern (Greek) Cyprus, 80% in London and nearly 100% in Ferrara (Italy) and this downward trend in births is expected to continue." Ibid., 2.

[42] Kuliev interview, December 2018 (n. 38).

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Thus, in the name of expanding preventive programs to "the whole Mediterranean area,"[43] he called for a WHO working group on the community control of hereditary anemias to convene in November 1981. Kuliev had indeed managed to task temporary advisers (assigned in Geneva as the HGP activities were under review) with prioritizing hereditary anemias—using a paper Williamson prepared on WHO request in 1981.[44]

At first, the reorientation of the WHO program rested on pilot studies in the Mediterranean basin, where national policies for thalassemia had already been implemented. The Cypriot program was in itself a "strong argument for providing population-based services"[45] and exporting them elsewhere, such as the Middle East and Southeast Asia.[46] The new HGP set up Cyprus as an ideal model of control using *preventive* policies to target genetic reproductive risks.[47]

The first report of the "task group on HGP," preparing its contribution to "Health for All by the Year 2000," promoted the incorporation of human genetics "into primary healthcare in communities" through the establishment of genetic centers in so-called

[43] "Community Control of Hereditary Anaemias: Memorandum from a WHO Meeting," *Bull. World Health Organ.* 61 (1983): 63.
[44] "A WHO Programme to Eliminate Human Genetic Disease: The Haemoglobinopathies," sent by Williamson to Kuliev, May 1, 1981, Williamson's archives.
[45] Modell and Kuliev, "History of Community Genetics" (n. 7).
[46] Memorandum from Kuliev to the Director of the Department of Non-communicable Diseases, March 13, 1984, WHO Archives, G3-181-129.
[47] Andrew McDowell, Lucile Ruault, Olivia Fiorilli, and Laurent Pordié, "Localization in the Global," in *Global Health for All: Knowledge, Politics, and Practices*, ed. Jean-Paul Gaudillière, Andrew McDowell, Claudia Lang, and Claire Beaudevin (New Brunswick, N.J.: Rutgers University Press, 2022), 29–55. The Advisory Group on Hereditary Diseases only marginally mentioned the Sardinian thalassemia control program and did not hold it up as an example, despite its striking results in decreasing incidence. Antonio Cao, cocreator of the program, was present at several meetings of the group.

developing countries and "appropriate training" of "primary care physician[s]," enabling them to provide basic genetic counseling.[48]

The HGP's "community-based" approach used hemoglobinopathies as a starting point to formulate, under the auspices of the WHO, a framework for "other genetic disorders on a global basis."[49] This enthusiastic group first mapped the distribution and prevalence of hemoglobinopathies. Investigating the possibilities for community control of selected genetic disorders, they reported the progress of programs in countries coming under the European and Eastern Mediterranean WHO offices.[50] This led the WHO Advisory Group on Hereditary Diseases, created in November 1982, to develop an action plan for the prevention and control of "genetic morbidity in the communities" and "a feasibility study of the genetic approaches for the control of common diseases."[51] The group deemed "community approaches to the control of hereditary disorders"[52] essential to address the growing and often hidden burden of NCDs in developing countries, for whom the creation of dedicated genetic services and the delegation of simple diagnosis and counseling tasks to primary health care centers were definitely not a luxury.

Moreover, the HGP ensured dissemination of knowledge, including junior researchers and clinicians in annual meetings in places affected by hereditary anemias (Sicily, Nicosia, Milan, Bangkok). For instance, Modell recalls that the Crete meeting motivated Italian, Greek, Cypriot, Portuguese, as well as Indian and Iranian physicians (obstetricians,

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[48] Report of the Task Group on WHO Human Genetics Programme, "Health for All by the Year 2000: The Contribution of Human Genetics" (Geneva, November 1981).
[49] "Community Control of Hereditary Anaemias" (n. 43), 63.
[50] "Progress in the Community Control of Thalassaemia" (1983), Report of the Second Annual Meeting of the WHO Working Group for the Community Control of Hereditary Anaemias, Nicosia, November 29–31, 1982, HMG/WG/83.9.
[51] Kuliev's Memorandum, July 9, 1984, on Advisory Group on Hereditary Diseases, Geneva, October 7–9, 1985, WHO Archives, G3-87-6.
[52] Report of a WHO Advisory Group, Geneva, October 3–4, 1985, HDP/WG/85.10.

pediatricians, clinicians, biochemists), to apply for training grants in the United Kingdom—sometimes accompanied by patients. Modell and her colleagues ended up nicknaming their London-based laboratory "the Club Méditerranée." Such training meant urging a global perspective for thalassemia eradication, so that young physicians could then locally "run a preventive program appropriate for the needs of their own community."[53] The gradual construction of an international interest for the control of genetic disorders rested on the circulation of scientists, but also on the strong links with newly created patients' associations (e.g., the Thalassaemia International Federation was established in 1986).

The very concept of "community genetics," albeit framed in 1990,[54] largely emerged from the working group's activities on the community control of hereditary anemias throughout the 1980s. Kuliev carried out the internal lobbying for the HGP. Insisting that time was right for the WHO to lead the standardization of genetic services in the then-called developing countries,[55] he encouraged WHO-sponsored workshops at a country level, including advanced WHO training courses in Cyprus (1983) and Thailand (1985). Similarly, Williamson remembers teaching in the 1980s and 1990s, along with British and U.S. counterparts, "for Africa, for Asia, for Greece and Italy," using "a bit of money" from the WHO.[56]

Kuliev endorsed the application of professional associations such as the African Genetics Association in 1983,[57] and actively bolstered the development of a network of

[53] "Minutes of a Meeting Held in Cagliari" (n. 41).
[54] Modell and Kuliev, "History of Community Genetics" (n. 7).
[55] One decade later, international organizations labeled the same group of countries low- and middle-income countries (LMICs).
[56] Williamson interview, January 2019 (n. 39).
[57] Correspondence between Kuliev and Aromose (Biological Sciences Dept., University of Benin, Nigeria, president of the African Genetics Association), April–May 1983, WHO Archives, G3-348-1 J2.

collaborating centers in research and training for achieving regional targets. Following a series of correspondences and Kuliev's official visit to the Greek Ministry of Health in November 1983,[58] one such collaboration with the National Unit for Prevention of Thalassaemia in Athens was agreed upon.[59] Other collaborating centers also emerged—the Siriraj Hospital in Bangkok, the Human Genetics Program of Cuba (as a result of close ties between HGP members and geneticist Luis Heredero in Havana). Even if the WHO-collaborating center label is mainly honorary, the HGP managed to provide modest financial support and training. More importantly, these collaborations participated in the HGP attempt to standardize genetic services by embarking WHO regional offices into prioritizing genetics. Thus, regional directors were regularly asked to attend HGP meetings. The following section deals with the specific role played by the WHO's Regional Office for the Eastern Mediterranean (EMRO, originally based in Alexandria, later in Cairo).

EMRO: The Middle Eastern WHO Hub for Community Genetics

From Morocco to Iran and Pakistan, the WHO's Regional Office for the Eastern Mediterranean deals with twenty-one states and the occupied Palestinian territories. In Kuliev's words, EMRO became the "home" of the genetics program at the end of the 1990s, when Geneva's interest seemed to fade away.[60] For him, the motivation for shifting the center of operations to Alexandria was the need for more concrete action: "In Eastern Mediterranean

[58] One of the first letters sent by Fessas (professor of Medicine, University of Athens) underlined the "expertise and willingness" of the unit but also its "geographic position of Greece near to countries of the Middle East, which face the same problems." Letter sent to Kuliev, October 7, 1983, WHO Archives, G3-286-2 J1.

[59] WHO travel report of Kuliev in Greece and Cyprus, November 22–December 3, 1983, WHO Archives, G3-286-2 J1.

[60] Our corpus allows only speculation about the motives of this move, for example, the declining commitment of the WHO leadership of HGP after Kuliev's departure.

you cannot do much laboratory things. . . . But organizational things! For example, on WHO behalf, we developed the prevention of thalassemia in Iran. It is primary, community approach prevention. This is a place where all marriages are arranged. . . . And you may be surprised that they cut the incidence by more than half in Iran."[61] Global guidelines were drafted in general debates in Geneva, and then more precisely defined at WHO's European office before being passed to EMRO.

EMRO's role in the history of community genetics is tightly related to Ala Alwan, a physician from Iraq, who served, during the 1990s, as regional adviser for noncommunicable diseases, then director of the office before becoming WHO country representative to Oman and later director of the Division of Health Systems Development in Geneva. In him, Modell found an ally, and the 1990s were a decade of sustained activity in the region in relation to genetics. In 1993, EMRO's annual report included a section about genetics, consanguinity, and the importance of hereditary disorders, the first time an EMRO publication specifically mentioned community genetics. Up to the early 2000s, EMRO gradually increased its focus on genetic disorders, leading the office to expand its original Mediterranean focus. Throughout the 1990s, Iran and Saudi Arabia were frequently advised on inherited blood disorders and hosted WHO-collaborating centers. Iran emerged as the regional success story as introducing a national program in 1997 with mandatory premarital screening and genetic counseling. That same year, medical termination became legal before sixteen weeks of gestation for fetuses affected by severe disorders.[62]

[61] Kuliev interview, December 2018 (n. 38).

[62] Hossein Najmabadi, Alireza Ghamari, Farhad Sahebjam, Roxana Kariminejad, Valeh Hadavi, Talayeh Khatibi, Ashraf Samavat, Elaheh Mehdipour, Bernadette Modell, and Mohammad Hassan Kariminejad, "Fourteen-Year Experience of Prenatal Diagnosis of Thalassemia in Iran," *Commun. Genet.* 9 (2006): 93–97.

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In 2001, EMRO launched the Eastern Mediterranean Network to Non-Communicable Diseases. The aim was to create guidelines suitable for each national context as well as specific regional guidelines for the management of inherited blood disorders. Planners inevitably used Cyprus's experience, referring to it as "a model of a successful control programme."[63] In the same year, the regional office planned a multicountry meeting to address genetic disorders. The goal was to develop a specific plan for each country. However, during those years, EMRO's publications showed a decline in the importance of medical and community genetics within the WHO. Starting in 2004, inherited blood disorders, genetics, and thalassemia were no longer included in the NCDs strategy or reports.

Community Genetics for Primary Health Care?

Despite these initiatives, the HGP's influence was limited and its role within the WHO was shaped by various elements. First, the combination of *genetics*, which is often seen as a cutting-edge field, and *community*, which falls under public health and primary health care, caused hesitation in the human genetics landscape. U.S. and European clinicians dominated this field and emphasized its incommensurability with eugenics. They considered genetic diagnosis an individual or, at best, a family issue.[64]

Second, the HGP struggled with the critical social and political issue of termination of affected pregnancies. Aware of what they called "religious or cultural objections" in various

[63] WHO Regional Office for the Eastern Mediterranean, "The Work of WHO in the Eastern Mediterranean Region: Annual Report of the Regional Director, 1 January–31 December 2003" (WHO EMRO, Cairo, 2004).

[64] Replying to Herman Vanden Berghe's letter (November 23, 1982), who had felt "some uneasiness" among the renowned experts invited in Geneva to renew the WHO Human Genetics activities in November 1982, Kuliev approved (letter, January 31, 1983): "Even in some common diseases where the genetic component seems to be obvious, geneticists have, until now, been very reluctant to try to use relevant data in general health measures." WHO Archives, 83-87-6. See the alternative definition of community genetics proposed in 2010 for these reasons in Ten Kate et al., "Community Genetics" (n. 7).

countries and communities, the experts relied on first-trimester fetal diagnosis to build legitimacy for prevention (early termination being lawful in several countries of the Global South). In early discussions, the HGP acknowledged that depending on "the cultural attitudes of the community concerned," measures of diagnosis and treatment of homozygotes could in some settings be "preferable" to advocating for medical termination.[65]

Third, the HGP experts had to challenge the preconceived idea that in low- and middle-income countries (LMICs) genetic services were complex technological solutions to secondary issues that took resources away from more pressing concerns. They reviewed and published epidemiological data to demonstrate the heavy burden of genetic diseases on health resources and carried out political advocacy to assert the feasibility of genetic services in the Global South.[66] Kuliev was careful to argue for the integration of genetic disorders management into primary health care—without specifying how this should be implemented.

Modell and the HGP experts envisioned community genetics as a population-based approach that favored screening and the conjunction of early diagnosis and abortion. However, Modell and her fellow clinicians discussed with WHO officials, who certainly endorsed a different understanding of community in health.

Community was a key concept in the debates framing the primary health care strategy and later the initiatives to implement it. Within this framework, communities are defined as social groups that inhabit a specific territory and have their own social and cultural organization and priorities.[67] The strategy reflected the visibility of communities as both targets and actors in four dimensions: (1) prioritizing neglected and vulnerable populations, such as rural communities; (2) emphasizing the use of "appropriate" technologies that are

[65] "Community Control of Hereditary Anaemias" (n. 43).
[66] Ibid.
[67] Gaudillière et al., *Global Health for All* (n. 47).

simple, affordable, and socially/culturally acceptable; (3) approaching health as one of several dimensions of development, including food production, water management, and education; (4) recognizing that communities have their own health needs and visions. This recognition should lead to advocating for community involvement, which may create tension between providing logistical support to local services and participating in the definition of their aims and the allocation of resources.

The inscription of community genetics within WHO debates resulted in neither a direct endorsement of these priorities by the Modell network nor a formal acceptance of the population genetics approach by Kuliev and other WHO officials. The creation of the HGP expert group, its debates, and the production of its many reports led to mutual adjustments, resulting in a juxtaposition of meanings typical of boundary objects. Community genetics thus remained a polysemic term with two interacting layers of meaning. The first layer was simultaneously shared, encompassing, and fuzzy, as it focused on the importance of hereditary disorders in developing countries without any specific reference to primary health care. It involved reducing the burden of inherited diseases through population-oriented measures (premarital counseling, prenatal diagnosis, neonatal screening, etc.) in parallel with the care of affected individuals. Community involvement was viewed as the best way for ensuring legitimacy for these interventions. The second layer was more precise and adapted to the needs of specific actors. For the European clinicians in the Modell network, community genetics relied on a politically supported combination of biochemical and cellular techniques of diagnosis, prenatal screening, legal incentives for testing, therapeutic abortion, and care for affected individuals. Within this context, education about the disease and its prevention was high on the agenda. In contrast, WHO staff involved in genetic initiatives during the 1980s and 1990s had a broader range of interventions in mind, but their view of community genetics was politically more precise. It revolved around the post-Alma-Ata WHO's official strategy,

an assemblage of prioritizing primary health care centers as sites of intervention, favoring task shifting from medical to paramedical personnel, and framing community as both a local site and a group of people whose participation (and not only education) was necessary. These tensions played a central role in the following decade when the question of translating the global aspirations of community genetics into policies and practices came to the fore.

Beyond WHO: National Developments and Alternatives in the Global South

Nationalizing the Strategy: The Omani Example

Oman is one of the countries where national programs on hereditary disorders have emerged in the wake of the work of the WHO Human Genetics Unit. The Omani example shows how national programs could be implemented without strong support from the WHO regional office. In addition, Oman's three-tiered health system is characterized by a strong network of primary health care facilities, a context described by international experts as conducive to the implementation of a community genetics program.[68]

Hereditary diseases appeared on the agenda of Omani health policy in the 1990s: in 1995, the Ministry of Health published the National Genetic Blood Disorders Survey, which estimated the birth prevalence of congenital disorders in Oman at 73/1,000 (compared to 44/1,000 in Europe). Hemoglobin disorders accounted for 3.5–4.7/1,000 of these cases, with beta-thalassemia affecting 2/1,000 children and sickle-cell disease 0.7/1,000.[69] High fertility

[68] Thalassaemia International Federation, "Visit Report, Sultanate of Oman—8–10 October 2017." (https://thalassaemia.org.cy/wp-content/uploads/2017/10/Report-TIF-visit-to-the-Sultanate-of-Oman-2017-TIF-finalised-3.5.2018.docx)

[69] Asya Al-Riyami, "National Genetic Blood Disorders Survey (Part of National Family Health Survey)" (Muscat: Ministry of Health, 1995). This relatively early review of the overall congenital disorders situation did not result in major governmental action.

rates at that time made genetic counseling after the birth of one affected child still relevant to decreasing the incidence of inherited blood disorders.

Similar to the collaboration between Cypriot and London doctors in Cyprus, the development of community genetics in Oman has been strongly influenced by international collaborations involving a handful of people. Among the central actors involved from the late 1990s to the late 2000s is Anna Rajab, a pediatrician from Ukraine who has lived in Oman since 1972 and trained in genetics in the 1990s in London. Upon completion of her Ph.D. in 1998, she went back to working in the Royal Hospital in Muscat as the only physician with a degree in genetics in the country—her doctoral committee included a certain Bernadette Modell. Despite Rajab's proximity to the Omani government (her husband was a minister), and the reference to Cyprus as a model of success, she depicts the establishment of genetics in the country as an arduous process. She also emphasizes that "genetic service at that time looked expensive, esoteric, not applicable in curative medical services, and irrelevant in Islamic culture."[70]

The development of genetic services in Oman draws two parallel and complementary paths, both made possible by the massive oil and gas rents that make most of the country's income. The first path involves attempts to implement a national community genetics program for the control of the most common genetic diseases, namely inherited blood disorders. Close to a vertical program, it requires national guidelines and a dense network of screening and counseling facilities. The second path deals with the diversification of tertiary health care services in clinical genetics. This involves building and equipping specialized laboratories in

[70] Middle-East Molecular Biology Society, "Professional and Personal CV" (2017, 2019), https://membs.org/membs//uploads/user/1499694847_Professional%20and%20personal.docx. Rajab provides a public account of her professional trajectory in this document, as confirmed in personal communications.

the capital as well as training Omani specialists in genetics, genetic counseling, and genomics. The multidisciplinary expertise necessary to diagnose and manage genetic disorders as well as the multiplicity of diagnostic tools involved make medical genetics a mainly hospital-based specialty, in Oman as elsewhere.[71]

Despite their quasi-absence from the Annual Health Reports of the Ministry of Health, important milestones for both paths took place in the late 1990s and early 2000s: the period 1997 to 1999 saw the design of the National Program for the Control of Genetic Blood Disorders. The central cytogenetic service and molecular genetic laboratory of the Ministry of Health opened in 2000 and was later in the 2000s equipped with molecular biology technologies—all of these developments being paid for by donations from private companies.[72] Despite these important steps, the mandatory use of premarital screening that was planned in the national program did not occur.

During the early 2000s, several other important events helped put national hemoglobinopathy prevention on the official agenda. In 2003, after suggestions from Alwan (WHO representative to Oman at that time), the government invited Modell.[73] Her mission was to report on the "possibilities for the control of congenital and genetic disorders" in Oman, similar to what she had done in Iran.[74] In 2006, the Ministry of Health organized a workshop about birth defects, with the aim of "lead[ing] to a five-year plan to develop programs to prevent birth defects, and [strengthening] the care of babies affected by them."[75]

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[71] Claire Beaudevin, Fanny Chabrol, and Claudia Lang, "Persistent Hospitals," in Gaudillière et al., *Global Health for All* (n. 47), 147–69.
[72] A. Rajab, I. Al Rashdi, and Q. Al Salmi, "Genetic Services and Testing in the Sultanate of Oman: Sultanate of Oman Steps into Modern Genetics," *J. Commun. Genet.* 4, no. 3 (2013): 391–97.
[73] Modell and Darlison interview, November 2016 (n. 1); Vaidya, "Congenital Anomalies High in Oman" (n. 1).
[74] Ibid.; cf. also the opening paragraph of this paper.
[75] March of Dimes Foundation, "Giving Every Baby a Healthy Start: Annual Report 2006" (White Plains, N.Y., 2007).

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In the absence of a regulatory framework and consensual protocol (particularly concerning the age of screening),[76] the screening for hemoglobinopathies developed slowly. Until the early 2010s, genetic counselors were indeed not available in the country, and hematologists and pediatricians performed the counseling for inherited blood disorders.

It is noteworthy that despite time passing, the returns of Omani geneticists from studies abroad, and continuous institutional and professional debates, the divide between genetic medicine and community genetics remained in the following decade. Public and private investments allowed for the creation of two cutting-edge tertiary medical genetics centers, but countrywide basic screening remained elusive (despite the availability of the necessary hemoglobin analysis machines for several years).[77] This situation is partly rooted in a persistent disagreement among physicians dealing with hemoglobinopathies regarding the best timing for screening: Prenatal? Neonatal? Premarital? Preconceptional?[78] In 2021, eighteen years after Modell's visit, figures of prevalence reported in the press have not changed.[79] What started as an exemplary integration of community genetics and primary care is not really working as community genetics.

"Care Is an Absolute, Prevention Is the Ideal": South Africa as a New Model

In the 1990s, the WHO community-based control strategy for the most common inherited diseases was losing momentum. Despite advocating for a reoriented program, Kuliev and Modell's network of experts faced challenges in persuading headquarters to support global

[76] Rajab retiring from her position as a physician in the Royal Hospital was also a contributing factor.
[77] Thalassaemia International Federation, "Visit Report" (n. 68).
[78] Claire Beaudevin, "Faqr al-dam, l'indigence du sang, comme héritage" (Ph.D. diss., Aix-Marseille University, 2010); Thalassaemia International Federation, "Visit Report" (n. 68).
[79] Times News Service, "Majority of Oman's Population Carry Genetic Blood Disease Gene: Experts," *Times of Oman*, June 12, 2021.

genetics policy, due to a lack of resources and attention. The HGP network published numerous reports during the 1980s and 1990s. However, these reports did not lead to any significant programs endorsed and coordinated by the WHO, beyond its support for national initiatives such as in Oman. In 1987, genetic and hereditary disorders were explicitly put on the agenda of the WHO's Programme of Work related to NCDs for 1990–95. It was meant to support "the testing and implementation of community-based programmes for the control of the commonest hereditary diseases, such as the haemoglobinopathies, cystic fibrosis and birth defects, as well as other common chronic diseases with genetic predisposition."[80] A modest budget of $1.6 million was allocated and remained similar in the early 1990s, , while most activities remained prospective.

In this uncertain context, the network developed a new framework focusing on primary health care. The framework was boosted by local experiments in the Global South, particularly those of South African pediatrician Arnold Christianson. Christianson broke new ground by bringing genetics into rural areas through a comprehensive approach implemented by nonmedical staff. This change would focus on "birth defects" as a target category and strengthen the link between community genetics and the established effort to promote maternal and child health.

Christianson began working in academic hospitals in Zimbabwe in 1974. Later, he worked in primary health care hospitals in rural areas, where he was responsible for surgery and obstetrics. In 1990, he became a pediatrician specializing in developmental pathologies at the University of Witwatersrand (Johannesburg), before joining the department of Medical Genetics at the University of Pretoria in 1992. Even as a senior lecturer in medical genetics,

[80] World Health Organization. (1987). Eighth general programme of work covering the period 1990-1995. World Health Organization. Health for all series ; no. 10. https://iris.who.int/handle/10665/39340

he continuously sought opportunities to "find out what the situation [was] in deep South Africa," whereas his neurodevelopmental colleagues refused to discuss childcare in rural settings.[81]

Despite what Christianson considered a "very [narrow] vision of human genetics," in 1990 the South African government supported a genetic outreach program in seven rural hospitals with the assistance of academic departments in medical genetics.[82] Going into rural areas to teach, Christianson met Philip Venter, a cytogeneticist who was working at a Black university. Together, they trained nursing staff in a rural primary care hospital in the northern region in basic genetics, enabling them to recognize infants who suffered dysmorphias. From 1989 to 1992, an important study was conducted based on the work of these nurses. The study documented high levels of neural tube defects and Down syndrome in Black communities.[83] Considering that defects detected at birth were only part of a "congenital anomalies iceberg," Christianson evidenced that the cumulative incidence of severe congenital disorders in Black neonates was significantly high in rural, socioeconomically deprived South Africa. Christianson and Venter argued that this major issue remained unrecognized due to a lack of research and medical facilities for diagnosis, in addition to the absence of statistics about affected children. For instance, the corresponding deaths were typically included in the general count of infectious diseases and malnutrition. They concluded that rural areas required the introduction of "prenatal, genetic, family planning and pediatrics facilities into the

[81] Arnold L. Christianson, interview by Lucile Ruault, videoconference, May 2019.
[82] This occurred three years before the end of apartheid and the arrival to power of the ANC, which reorganized the health system in resonance with WHO's primary health care principles.
[83] P. A. Venter, A. L. Christianson, C. M. Hutamo, M. P. Makhura, and G. S. Gericke, "Congenital Anomalies in Rural Black South African Neonates—A Silent Epidemic?," *S. Afr. Med. J.* 85 (1995): 15–20.

primary healthcare delivery system[s],"[84] which was then not a priority in recently postapartheid South Africa.

A medical genetics education program was gradually taking shape, including lectures and practical interactions with patients. Every four months, Christianson visited nursing staff in primary health care hospitals with at least one geneticist teaching on birth defects diagnosis and counseling. The guiding principle was to examine and treat patients as close to home as possible. Christianson refers to his group as the "clinic gang." The group consisted of genetics-trained nursing sisters (GTNSs) who were respected in the area and could communicate with patients in their own languages. The group also included local actors, such as the wife of a rural chief, who had influence and could locate children with birth defects in the villages.[85]

Advocating for community genetics as a primary-health-care-based genetic system in "developing countries," Christianson's team converged with the 1991 report by the WHO's European office about the need for "political will and financing" from health administration to ensure national/provincial implementation of such programs in the public sector and the central role of nonmedical personnel. However, the South Africans' vision differed from that of the HGP experts, since the former prioritized care over screening and preventive measures. The "best possible patient care" encompassed prenatal, genetic, family planning, and pediatrics facilities, aiming at an early diagnosis of genetic problems for the sake of optimal management (treatment, genetic counseling and psychosocial support, including social welfare and education).[86] Neither did these primary-care-oriented approaches separate

[84] Ibid.
[85] Christianson interview, May 2019 (n. 82).
[86] A. L. Christianson, P. A. Venter, J. H. Modiba, and M. M. Nelson, "Development of a Primary Health Care Clinical Genetic Service in Rural South Africa—The Northern Province Experience 1990–1996," *Commun. Genet.* 3 (2000): 77–84.

prenatal and postnatal health; they called for integrating genetics into existing maternal and child care packages, notably growth monitoring and birth spacing. In the name of these approaches, also promoted in Latin America,[87] Christianson and his colleagues argued that the ideal role of genetics in primary health care in LMICs was "entirely different from that envisaged for developed countries."[88]

As these experiences gradually constructed a new model for primary health care genetic services in rural areas, Christianson's interests took an international turn. In 1997 at the twenty-fourth meeting of the International Clearinghouse for Birth Defects Monitoring Systems in Cape Town, he met Michael Katz from the March of Dimes (a U.S. philanthropic organization funded in 1938 to eradicate polio) and Ysbrand Poortman from VSOP, the Dutch Alliance of Parent and Patient Organizations. The Cape Town Declaration, which resulted from the conference, called on governments, NGOs, and health care providers to acknowledge the burden of birth defects in "developing countries" and the need to promote both fundamental research and the establishment of comprehensive genetic services. Following this statement, Christianson, Katz, and Poortman organized an expert meeting to evaluate the status of genetic services in developing countries and develop plans for managing and preventing birth defects. This joint meeting between WHO and WAOPD (World Alliance of Organizations for the Prevention of Birth Defects), held in The Hague in January 1999, highlighted two key issues in developing countries: the lack of dependable empirical epidemiological data and the importance of clinical care in addressing the burden of congenital diseases. During the meeting, the latter turned out to be inseparable from the

[87] Victor B. Penchaszadeh, "Genetics and Public Health," *Bull. Pan Amer. Health Organ.* 28 (1994): 62–72.
[88] A. L. Christianson, G. S. Gericke, P. A. Venter, and J. L. Du Toit, "Opinion: Genetics, Primary Health Care and the Third World," *S. Afr. Med. J.* 85 (1995): 6–7.

extremely contentious issue of abortion, which raised heated debates among participants. Christianson thus formulated "the overarching principle that came out" of his experience in South Africa: "care is an absolute, prevention is the ideal."[89]

This emphasis on care reversed the strategy that had guided the model of community genetics up until then. At the same time, Christianson joined the WHO advisors on genetics. In December 1999, he chaired one of the meetings in Cairo that formed part of a general advocacy for a WHO epidemiological study of genetic disorders so as to design a general approach to their control "in any country."[90]

In Comes the March of Dimes

This turn toward a more clinical vision of community genetics received full support in the following years with two mounting commitments of the March of Dimes: to fostering a greater global understanding of its activities and to making "birth defects" more visible in the health policies of LMICs and thus more widely treated. At this time, the community genetics network was demonstrating growing frustration with the diminishing power of the WHO (from the early 1990s onward) and its lack of initiative about hereditary disorders. Engaging with this U.S. organization focusing on congenital disorders at large (i.e., including genetic disorders) thus appeared at the time a welcomed opportunity.

In 1997, Christianson had begun working with Modell, who saw in him a prime example of community genetics. She recalls, "We met and . . . I said to him, 'Do you know another geneticist who's actually got out in the community and become engaged with the population?' He said, 'No,' and I said, 'Neither do we. You're the only one.'"[91] The

[89] Christianson et al., "Development of a Primary Health Care Clinical Genetic Service" (n. 87).
[90] "Primary Health Care Approaches for Prevention and Control of Congenital and Genetic Disorders, 2000" (report of a WHO meeting, Cairo, Egypt, December 6–8, 1999), WHO/HGN/WG/00;1.
[91] Modell and Darlison interview, June 2019 (n. 2).

collaboration between Modell, Christianson, Howson, and Katz resulted in a report and set of recommendations that the March of Dimes endorsed in 2006.[92] The initiative was considered the "culmination of B. Modell's work on hemoglobin disorders for WHO."[93] The first recommended target was to create a database for assessing the quantitative toll of all diseases affecting newborn babies, whether caused by a genetic mutation or not. The second one was the analysis of existing genetic services and the discussion of policy priorities for the Global South. The main obstacle to implementing this agenda was identified as misconceptions held by donors and policy makers, resulting in a lack of investment in NCDs in LMICs. Regarding congenital disorders, these misconceptions were of two sorts: (1) they were not important epidemiological problems (a misrepresentation allegedly reinforced by the lack of relevant data); (2) care as well as the prevention of birth defects were costly interventions LMICs could not afford since such programs would cut budgets for more important targets, such as maternal and child health programs. In other words, congenital disorders were victims of the prevailing modus operandi in international public health, which remained an approach linking public health, epidemiology, and infectious diseases.

To counter this configuration, the new alliance proposed a two-step strategy, which might—along with a wealth of data—convince health policy makers. Based on the premise that certain interventions were not costly in relation to their benefits and in line with existing practices in primary health care (such as diet supplementation, family planning, and the controlling of infections in pregnant women), the report's authors proposed a first phase of genetic services. This phase would include basic prevention, education of primary care

[92] A. L. Christianson, C. P. Howson, and B. Modell, "March of Dimes: Global Report on Birth Defects, the Hidden Toll of Dying and Disabled Children" (New York: March of Dimes Birth Defects Foundation, 2006).
[93] Ibid., 6.

professionals, and expansion of maternal child health. The second phase would involve more direct investments in medical genetics, creating integrated services for prenatal diagnosis of hemoglobin disorders, Down syndrome, and congenital malformations as well as newborn screening for the most common metabolic disorders.[94] The persistent opposition between prevention and care was thus simply dismissed: "The credibility of medical genetics services depends on commitment to care both within the service and among the public. If care and prevention are provided simultaneously, the success of prevention can help counterbalance the costs of care, and make it more available."[95]

In parallel with these programmatic developments, the promoters of "congenital" as an overarching category attempted to create a new, and more political, network involving physicians and geneticists from developing countries. However, by the second of these now called International Conferences on Birth Defects and Disabilities in the Developing World, which took place in Beijing in 2005, the relationship had deteriorated again due to the WHO's increasing commitment to global genomics, a technology- and research-centered approach, which the congenital disorders network viewed as direct competition to the support of community genetics.

Tensions between the community genetics network, the March of Dimes, and the WHO indeed increased after a 2002 meeting in Toronto, which turned genomics into a first priority for the WHO in spite of an open dissent from the two developing-world geneticists present, Christianson and Heredero.[96] The United States and most European countries actively supported genomics, which was soon endorsed by the World Health Assembly. Despite these discordances, Katz and the March of Dimes chose to avoid an open conflict with the WHO

[94] Ibid., 13.
[95] Ibid., 39.
[96] Christianson interview, May 2019 (n. 82).

and continued to seek their endorsement of the strategy for controlling congenital disorders in developing countries. As a result, all participants, including the Chinese deputy minister of health, signed a petition at the 2005 meeting, asking the WHO to engage more decisively with birth defects. China's program, which included folic acid supplementation, the use of iodized salt, and rubella vaccination, has allegedly reduced the burden of congenital disorders by 50 percent. China was then ready to push the World Health Assembly to adopt a document on the importance of reducing the burden of birth defects. By 2008, India was also ready to join this effort after the third meeting of the network in Brazil.[97]

Thus, Ala Alwan, then–assistant director general for NCDs at the WHO and participant in most meetings since 2001, launched the drafting of a resolution to be examined by the WHO executive board. He approached Christianson, who produced a first version of a resolution, focusing on data and the public health ramifications of the problem. Alwan then rewrote this draft in order to turn it into a WHO policy document urging member states "to set priorities, commit resources, and develop plans and activities for integrating effective interventions to prevent and care for children with birth defects into existing maternal, reproductive and child health services."[98]

As the collaboration with the March of Dimes and endorsement of South Africa's experience in caring for children with congenital malformations in remote areas stabilized, community genetics was thus redefined. The main ambition became the integration of screening for priority diseases into local and comprehensive health centers, implementation by nonmedical health staff, education/counseling of target populations, and fostering participation. Although approved by the World Health Assembly in 2010, this plea did not

[97] Ibid.
[98] World Health Assembly, "Resolution 63.17. Birth Defects" (Geneva: WHO, 2010).

provide many changes in the status of genetic and congenital disorders in the global governance of health: as a field *global health* remains focused on communicable diseases, which gather more than 95 percent of all operational investments.

Conclusion

Bringing together Cyprus, London, Oman, and South Africa, this paper has traced the four-decades-long work of a group of biomedical scientists, physicians, WHO personnel, and health policy makers interested in a particular set of NCDs—that they alternatively labeled hereditary or congenital disorders. The group aimed to make these diseases a global public health issue deserving significant investments in targeted control programs, focusing on prevention but including care. Similar to many processes of health globalization, their attempts were multilayered, including framing disease categories, creating expert groups, organizing reference experiences for diagnosis, care, and counseling, and seeking institutional niches and political support. Throughout this process, the concept of community genetics emerged as a shared category to distinguish their globalized practices from medical genetics. The experts we followed believed that the latter was too focused on hospital care, clinical specialties that vary widely, and an individualized approach to care and counseling. Instead, they recommended a population-centered and preventive approach.

In their seminal 1989 paper, Star and Griesemer insist on the role of the multiple meanings of boundary objects in mediating the views and interests of heterogeneous actors.[99] A similar pattern of interactions took place around community genetics. In the 1990s, the core set of geneticists who had mobilized around the Cyprus program and sought WHO support for similar initiatives in the Global South used community genetics to create a target of

[99] Star and Griesemer, "Institutional Ecology" (n. 8).

intervention in global health and engaged in dialogues that taught them about WHO's priorities and what they could expect. By the end of the decade, they had acculturated to the organization's vocabulary and approach. They also situated community genetics within the political context of primary health care. As a result, community genetics took on two distinct meanings: (1) a medical genetics approach that is more focused on the population and less clinical than the approach used in the Global North; (2) vertical screening initiatives related to maternal and child health, a mounting priority for WHO and other international organizations in developing countries.

The globalization of community genetics may seem to have been achieved, albeit slowly and ultimately at arm's length from the WHO. However, we argue that one should take into account not only the new layer of discursive activity in global health but also the practical characteristics and implications of these global aspirations. This includes the funding of programs, their design and operations, beyond the national initiatives used as references. In this perspective, "global community genetics" is similar to most NCDs that global health has addressed. Like "global mental health," hereditary and congenital disorders have gained significant attention in global epidemiology data and, to some extent, in global health's list of urgent issues. Yet they have not attracted any significant international funding, any more than all other NCDs. Overall, NCD programs account for only 3 to 5 percent of global health expenses.[100]

The global trajectory of hereditary/congenital disorders can shed light on the reasons for this general failure to invest in NCD management. In our previous analysis, we identified three characteristics of global health that discourage investments in the fight against NCDs: (1) the economization of health accompanied by a managerial culture of performance, based

[100] IHME website, https://vizhub.healthdata.org/fgh/.

on global metrics and cost-benefit analysis, which does not place NCDs among high-priority targets; (2) the pharmaceuticalization of care, focused on access to medicines and vaccines and therefore favoring areas where these technical responses are readily available and easy to implement; (3) the marginalization of state-based programs for health system strengthening, in favor of the promotion of single-disease-oriented private-public alliances.[101] Discussions of costs and performance among community genetics experts were rare. Our corpus does not indicate that the pharmaceuticalization or generalization of public-private partnerships were matters of interest to them. Even though collaboration with the March of Dimes revived momentum in the late 2000s, their prevailing political strategy remained WHO-centered, focused on documenting local experiences and collecting data on the "burden" of hereditary/congenital disorders. Within a global health field mostly ruled by *medico-economic* performance, such arguments proved not authoritative enough.

Tracing these attempts to turn hereditary/congenital disorders into a target of worldwide interventions also reveals an unexpected periodization in the history of international public health. The trajectory of community genetics challenges the now classical three-era divide between the 1950s and 1960s period of technology-oriented vertical programs focusing on the control or the eradication of one disease; the 1970s and 1980s dominance of public health system building and horizontal initiatives in the name of primary health care; and finally the 1990s and 2000s rise of global health with its renewed vertical private-public partnerships. Community genetics provides for a different narrative, starting with a quasi-vertical experience focused on a single disorder, a well-targeted population, and the implementation of a "technopack" mostly defined in the United Kingdom.[102] Interestingly,

[101] Gaudillière et al., *Global Health for All* (n. 47).

[102] We coined the term "technopack" to describe a set of technologies and techniques that make health technical, transportable, and transferable without significant infrastructural requirements. See Andrew

primary health care characteristics were integrated into community genetics as a practical aspiration only in the 2000s, coinciding with the successful dismantling of the global primary health care strategy. Unsurprisingly, this inclusion was permitted by a clear distancing from the WHO and its (paradoxical?) new commitment to genomic technologies.[103]

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McDowell, Claudia Lang, Mandy Geise, Sameea Ahmed Hassim, and Vegard Traavik Sture, "Tech for All," in Gaudillière et al., *Global Health for All* (n. 47), 124–46.

[103] For an analysis of the consequences of the WHO's genomic turn, see Sangeeta Chattoo, "Inherited Blood Disorders, Genetic Risk and Global Public Health: Framing 'Birth Defects' as Preventable in India," *Anthrop. Med.* 25, no. 1 (2018): 30–49.