Professor Samia Temtamy: the founder of human genetics at the National Research Centre, Egypt

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Received 21 November 2021
Revised 28 November 2021
Accepted 01 December 2021
Published 08 June 2022

Middle East Journal of Medical Genetics
2022,2:51–57

Professor Samia Temtamy is a great Egyptian icon of human genetics and one of the first three most eminent international human genetics scientists who obtained a PhD degree in the aforementioned field. Prof. Temtamy had successfully laid the foundation for the science of human genetics and genomics at the National Research Centre shortly after the discovery of the correct number of human chromosomes and all the way to the human genome era. The story began more than 50 years ago with the establishment of a small unit at the National Research Centre in Cairo, Egypt. The unit started as a clinic that accepted patients with various inherited abnormalities. Later, that small unit became the Human Genetics Department in 1977. Prof. Temtamy had made a great effort to continue improving the services provided to patients with genetic disorders and the research facilities by converting the Human Genetics Department into the Division of Human Genetics and Genome Research that included several specialized departments in 2003. The Center of Excellence for Human Genetics was then established in 2014, which is considered the largest institute of human genetics in Egypt. This review summarizes the long journey taken and the great efforts put in by Prof. Temtamy and her team at the National Research Centre in the field of human genetics in Egypt, to keep pace with the continuous rapid advancement of human genetics and genomics and contribute to the international scientific community.

Keywords:
awards and prizes, hand malformations, human genetics, the National Research Centre, the National Society of Human Genetics, Samia Temtamy

Middle East J Med Genet 2:51–57

2090-8571

Biography

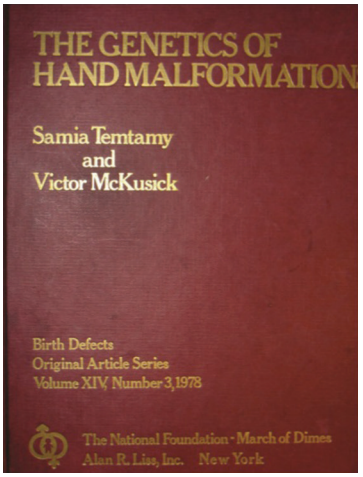
Professor Temtamy graduated from the Faculty of Medicine, Cairo University, in 1957. As an eminent student, she received the certificate of graduation from the president of Egypt at that time, Gamal Abdel-Nasser, at the occasion of the Festival of Science in 1961. Prof. Temtamy specialized in pediatrics, where she had a 2-year residency at the Children's Hospital, Cairo University, and obtained a diploma in child health (Fig. 1).

Being very much interested in tackling complicated cases and studying inherited abnormalities, Prof. Temtamy attempted to advance her career by exploring the research field. At that time, Professor El Nabawy El Mohandes, prof. of pediatrics, had established a medical research unit at the National Research Center in Cairo and accepted her as a fellow on his team owing to her high research potential and great ambitions. Prof. Temtamy accompanied her husband (Dr Moustafa Raafat, currently Professor Emeritus of Pathology, Cancer Institute, Egypt) in his scientific fellowship to the United States at Johns Hopkins University in Baltimore, Maryland. At Johns Hopkins University, she met Dr Victor McKusick, the father of genetics, who was impressed by her scientific mindset and ambitions. He accepted her as a candidate in the Division of Medical Genetics, where she started her PhD in human genetics. She had chosen the genetics of hand malformations as the subject of her PhD thesis (Temtamy, 2019). Based on comprehensive literature reviewing and studying patients from Dr McKusick's files, with tracing of families and completing clinical evaluation, Prof. Temtamy proposed a new classification for hand anomalies and was able to report new syndromes. She obtained her PhD in human genetics in 1966 and her thesis was described by Dr McKusick as among the notable nosologic outputs from the Moore Clinic (Temtamy, 1966). She was one of the first three most eminent international human genetics scientists who obtained PhD degree in genetics, including Alan H. Emery and David Rimoin. She then published her thesis as a monograph coauthored by her and Dr McKusick in 1978, describing in detail the genetics of hand anomalies and including new classifications. The book included other updated information obtained through a second postdoctoral fellowship at

Figure 1

Prof. Temtamy wearing the 'Pendant of The Nile' prize.

Figure 2



The Genetics of Hand Malformation: the famous monograph by Temtamy and McKusick.

the Moore Clinic (1973–1974) and new patients from Egypt were added. Her book is considered by many geneticists as one of the most important references in this field (Fig. 2) (Temtamy and McKusick, 1978; Temtamy, 1985).

Identification of new syndromes

'Treasure your exceptions,' this advice from Dr McKusick was the goldstone of Prof. Temtamy's subsequent scientific work. Through her exceptional observational power and the capability of recalling specific reported features and analysis of similarities and differences, she had identified more than 30 new syndromes throughout her scientific career (Temtamy, 2019). She named those newly identified syndromes after the authors who first reported them in the literature or after the main syndrome anomalies. Two brachydactyly syndromes were named after Prof. Temtamy by Dr McKusick in his renowned OMIM site (Mendelian Inheritance in Man) (Temtamy *et al.*, 1996, 1998a, 1998b, 1998c) – Temtamy-type brachydactyly (A4), OMIM #112800, and Temtamy preaxial brachydactyly syndrome OMIM # 605282.

She aimed at characterizing accurate molecular bases and proper genetic counseling. Subsequent identification of molecular bases underlying these syndromes had been carried out by comprehensive international efforts that resulted in various publications, including Carpenter syndrome (Meguid and Temtamy, 1988), Greig syndrome (Marafie *et al.*, 1996), Cenani–Lenz syndrome (Temtamy *et al.*, 2003b; Li *et al.*, 2010a, 2010b), Baller–Gerold syndrome (Temtamy *et al.*, 2003a), Temtamy preaxial brachydactyly syndrome (Temtamy, 2005; Li *et al.*, 2010a, 2010b; Temtamy *et al.*, 2012; Temtamy, 2013; Aglan *et al.*, 2014), Roberts syndrome (Temtamy *et al.*, 2006a, 2006b; Ismail *et al.*, 2016), 3-M syndrome (Temtamy *et al.*, 2006a, 2006b; Hanson *et al.*, 2009, 2012), the autosomal recessive variant of Adams–Oliver syndrome (Shaheen *et al.*, 2013), and Nager acrofacial dysostosis (Ismail *et al.*, 2017).

Establishing human genetics in Egypt

After spending 5 years in the United States, Prof. Temtamy decided to return to her country so that she could apply all the science and practice she had gained to facilitate early and accurate diagnosis, prevention, and management of genetic diseases among the Egyptian population. However, the task of introducing human genetics at the National Research Centre was not an easy one, as this science was in its very primitive stages in Egypt at the time (Temtamy and Hussen, 2017). She started by establishing a genetics unit in the Basic Medical Sciences Department at the National Research Centre in 1966. At the time, very few human genetics units had been founded in the pediatric departments of Ain Shams University by Prof. Nemat Hashem and in Cairo University by Prof. Ekram Abdel-Salam. Later, another unit was founded in the Medical Research Institute of University of Alexandria by Prof. Suzan Roushdy Ismail. Prof. Temtamy initiated a specialized clinic that accepted patients with congenital anomalies and genetic diseases with a trained staff. She also established a cytogenetics laboratory with a small tissue culture room, to which patients were referred from various areas in Egypt. Through the following years, she succeeded in increasing the funding facilities of the unit, as well as the offered services, which made the establishment of the Human Genetics Department a reality in 1977. Prof. Temtamy was able to form a team specialized in human genetics through teaching and transferring her expertise and practice of diagnosis of

challenging cases with open discussions and review of the latest research studies in the field. She also started accepting candidates of MS, MD, and PhD, and she had supervised more than 100 theses in genetics and other specialties in collaboration with Egyptian universities (Temtamy, 2019).

As the genetics field was rapidly expanding with multiple subspecialties and as a result of the increase in the number of researcher studies and PhD and MD students, the Human Genetics and Genome Research Division was established in 2003. It included eight integrative departments: clinical genetics, human gytogenetics, medical molecular genetics, molecular genetics and enzymology, biochemical genetics, immunogenetics, orodental genetics, and prenatal diagnosis and fetal medicine. These departments include about 229 well-qualified researchers covering various subspecialties (https://www.nrc.sci.eg/). The division became the referral center for all genetic conditions from all over the country. In 2021, the division has become the Institute of Human Genetics and Genome Research, NRC (https://www.nrc.sci.eg/*human-genetic-and-genome-research-institute/*) (Fig. 3).

As a continuation of her great efforts, the Center of Excellence for Human Genetics was established in 2014 acquired by a capacity building project through a Science and Technology Development Fund (STDF project no. 5253) supported by the Academy of Scientific Research and Technology, Egyptian Ministry of Higher Education and Research. Through this capacity building project, the equipment facilities of the division were upgraded facilitating better diagnosis and management of genetic diseases, making it the largest institute of human genetics in Egypt. Through her meticulous evaluation of referred patients with several genetic disorders, reporting rare observations, and sharing in the international publications and conferences, Prof. Temtamy and her team provided the field with a wealth of valuable research studies

Figure 3

Prof. Temtamy with a group of her team and younger colleagues during the basic genetics course organized by the division at the NRC.

in syndrome identification, limb malformations, and skeletal dysplasias. She provided a chapter of genetic disorders in Egypt as part of a book on genetic disorders in the Arab World (Temtamy *et al.*, 2010).

The center of excellence for human genetics, aim and scope

The aim behind the decision and monumental work of Prof. Temtamy to establish the Center of Excellence for Human Genetics was the advancement of the diagnostic strategies of human genetic diseases through introducing the latest technologies including next-generation sequencing, chromosomal microarray, laser dissection microscope, expression studies, and proteomics. She also aimed to identify prevalent gene mutations among Egyptian patients and early diagnostic and management strategies for genetic diseases among Egyptians. A comprehensive research program was put forward for diagnosis of rare genetic disorders with functional studies to provide insights to new genes and genetic pathways, thus leading to setting up novel therapeutic or preventive approaches. In addition, the strategy of the Center of Excellence aims at facilitating national and international collaborations and allowing the exchange of expertise worldwide.

National genetic health programs

A considerable effort has been made by the Human Genetics and Genome Research Division, NRC, toward national genetic health programs. In 2003, Prof. Temtamy and her team were granted a neonatal screening project through STDF fund, in collaboration with the Pediatric Genetics Units of Ain Shams and Cairo Universities, Mansoura University, and the Medical Research Institute at Alexandria University. A total of 15 000 neonates were screened for hypothyroidism, PKU, galactosemia, and biotinidase deficiency and for any other congenital anomalies. As a result, high frequencies of hypothyroidism and PKU were observed among Egyptian neonates (Temtamy, 1998), which paved the way for a national neonatal screening program for hypothyroidism in all Egyptian governorates in 2003, followed by a screening program for PKU. Several epidemiological studies were also carried out by Prof. Temtamy and her team.

Studies of consanguinity among Egyptians reported a rate from 29 to 39% (Temtamy and Loutfy, 1970; Temtamy *et al.*, 1994a, 1994b, 1998a, 1998b, 1998c; Khayat and Saxena, 2000; El-Nekhely *et al.*, 2008). The rate of parental consanguinity was found to be significantly high among groups of Egyptian patients with various birth defects and anomalies affecting various organs compared with that of the general population (Temtamy, 2001, 2004), with a significant

increase in autosomal recessive conditions (Temtamy and Aglan, 2012). Other studies reported the frequency of neonatal congenital anomalies (Temtamy *et al.*, 1998a, 1998b, 1998c) and of mental subnormality in Assiut Governorate, Egypt (Temtamy *et al.*, 1994a, 1994b), with a higher frequency of autosomal recessive disorders.

National society of human genetics

(https://silo.tips/download/the-national-*society-of-human-genetics-egypt*).

Prof. Temtamy was keen to help her community and to increase social awareness about genetic diseases through media and open workshops. Furthermore, she published a simplified book about genetic diseases in the Cairo International Book Fair in 2009. This effort toward her community culminated in the establishment of the National Society of Human Genetics, Egypt in 2005. At the meeting of the executive committee of the International Federation of Human Genetics Societies (IFHGS), in November, 2010, in Washington DC, USA, the National Society of Human Genetics was accepted as a corresponding member in the IFHGS, thus becoming the first member in the Federation from Egypt and from the Arab world to be honored with such a membership. The main objectives of this nonprofit society are to increase public awareness regarding the prevention and management of genetic diseases through the mass media such as newspapers, television, and radio programs; increasing public knowledge about care and rehabilitation of genetic disabilities through leaflets disseminated to the public; encouragement of recent graduates to join social activities with enhancement of national, regional, and international networking; and co-operation for early detection, prevention, and management of genetic diseases.

International conferences were arranged by the society in conjunction with the African Society of Human Genetics in Cairo (2007 and 2017). International speakers were invited for lectures and workshops to exchange experiences.

The projects conducted through the society included medical treatment of patients with osteogenesis imperfecta, constructing a database for genetic disorders in Egyptians, and increasing public awareness regarding genetic diseases by publishing a series of simplified booklets.

The Middle East Journal of Medical Genetics is the official Journal of the National Society of Human Genetics, Egypt, published since January 2012 by Wolters-Kluwer (https://review.jow.medknow.*com/mxe*).

International collaboration

It is rather hard to list all of the international collaborations that Prof. Temtamy had throughout her long journey. She actively participated as an invited speaker mostly in plenary sessions at more than 50 conferences worldwide. Several scientific research collaborations and joint projects had taken place with eminent scientists from all over the world – as an example but not limited to, a project for identification of mutations in the beta-globin gene among Egyptian patients with beta-thalassemia using recent technologies in collaboration with Prof. Giovanni Romeo, University of Genoa, Italy in 1993–1997 (Hussein *et al.*, 1997; Rady *et al.*, 1997). Another project with Prof. Giovanni Romeo was conducted between 2006 and 2008, through which, hybrid courses were streamed from Bertinoro, Italy, during the project period and continued afterward supported by the European Society of Human Genetics, the European School of Genetic Medicine, the National Society of Human Genetics, Egypt, and the National Research Centre.

A research project on the biochemical diagnosis of mucopolysaccharidosis was carried out with Prof. Ed Wraith, Manchester University, United Kingdom, from the year 2000–2002. This project facilitated exchange visits between Manchester University and our biochemical genetics team, enabling them to establish the foundation of the Biochemical Genetics Department, the main referral center in Egypt for diagnosis and enzyme replacement therapy for patients with mucopolysaccharidosis.

A 3-year project for identifying the genomic causes of rare genetic disorders among consanguineous families was conducted in 2012 with Prof. Stylianos Antonarakis and Prof. Hanan Hamamy, University of Geneva, Switzerland, and resulted in important publications (Hamamy *et al.*, 2011; Makrythanasis *et al.*, 2014a, 2014b). A 10-year collaboration with Prof. Victor L. R. Pérez and his team in Instituto de Investigaciones Biomédicas, Madrid, Spain, was continued through multiple projects on clinical and molecular aspects of Ellis–van Creveld syndrome and bone fragility disorders, through which, new genes were identified in autosomal recessive osteogenesis imperfect (Valencia *et al.*, 2009; 2014; Lapunzina *et al.*, 2010; Martínez-Glez *et al.*, 2012; Puig-Hervás *et al.*, 2012; Keupp *et al.*, 2013; Caparrós-Martin *et al.*, 2013, 2015, 2016; Tenorio *et al.*, 2017; Doyard *et al.*, 2018).

Other important international collaborations were carried out with Professors Bernd Wollnik, Fowzan Alkuraya, Han Brunner, Stanislav Kmoch, A. Megarbane, M.J. Marafie, Maximilian Muenke and Professors James Lupski from Baylor College of Medicine, and David Valle from Johns Hopkins (Temtamy, 2019).

These collaborations had resulted in novel research studies in the field of genetics, including limb anomalies, skeletal dysplasias, and novel genes responsible for some syndromic manifestations. These research studies could be viewed at the website of the National Society of Human Genetics, Egypt, where the staff publication list is updated, and all international publications could be searched for in Scopus and PubMed.

Multiple international speakers in various specialties of genetics were invited through the National Society of Human Genetics, including Prof. Joseph Gleason Head of Neurogenetics Laboratory, University of California, in 2006, 2007, and 2008, who also conducted a lot of joint research projects with the Clinical Genetics Department; Prof. Saddaf Farooqi, consultant technologist, Welcome Clinical Scientist, Cambridge University, UK, in 2006; Prof. Olaf Hiort, Head of Pediatric Endocrinology, Lubeck University, Germany, in 2006; Weem Dorlin, Representative of Agilent Company for European Technology for RNA/DNA microarray analysis in 2006; Prof. Elliott H. Sherr, Department of Neurology, the University of California, San Francisco, in 2006; and Prof. Albert Schinzel, the University of Zunich, in 2003 (Fig. 4).

Prof. Samia was an important member of the African Society of Human Genetics, which constitutes another forum for human genetics scientists in Africa. Regular meetings were organized to facilitate

Figure 4

Prof. Temtamy with Prof. Ghazi Tadmouri from the Faculty of Public Health, Jinan University, Tripoli, Lebanon, at an international conference.

Figure 5

Prof. Temtamy and a group from her team with Prof. Maximilian Muenke from the National Human Genome Research Institute, National Institutes of Health, USA, at the 10th conference of the African Society.

Figure 6

Prof. Temtamy receiving HUGO African Prize in 2017 by the International Human Genome Organization.

Figure 7

Prof. Temtamy with her husband Prof. Moustafa Raafat and Dr. Tarek Kabil who nominated her, according to a request by Al-Ahram Journal, to be chosen as the best scientific figure in Egypt. According to her biography and consultation of Al-Ahram readers and her colleagues, there was a consensus for that nomination.

collaborations and interactions of the African scientific community. These meetings aimed at exchanging recent information and applied knowledge and to attract the international community to the efforts of African scientists to keep pace with the rapidly advancing field of human genetics. The Fifth and the Tenth Conferences of the African Society of Human Genetics in conjunction with the National Society of Human Genetics, Egypt were hosted in Egypt in November 2007 and 2017. Renowned international speakers were invited including Prof. Charles Rotimi (Founding President of the African Society of Human Genetics) in 2007, Professor Michèle Ramsay (Current President of the African Society of Human Genetics) in 2017, Dr. Francis Collins (the US National Institutes of Health director), and Dr. Maximilian Muenke (National Human Genome Research Institute, Maryland, USA) (Fig. 5).

The Asian–European Workshops on Inborn Errors of Metabolism were also held in Egypt in 1995, 2005, and 2015.

In the meeting of the Human Genome Organization (HUGO) headed by Prof. Stylianos Antonarakis, Prof. Temtamy presented a plenary award lecture and was honored with the HUGO African Prize for lifetime contributions in human genetics. The meeting was held in Barcelona, Spain, in 2017 (Fig. 6).

Prizes

Prof. Samia Temtamy has been honored with multiple prizes, both nationally and internationally. The most prominent prizes were the Nile Prize in 2012, which is the most prestigious science award in Egypt, and the State Prize of Merit in Sciences and Arts in 2013. She was also awarded the HUGO African Prize in 2017 by the International Human Genome Organization. She was recently nominated by Al-Ahram Journal to be the best scientific figure in Egypt through a consensus voting (Fig. 7).

Until the very last days of her rich prolific life, Prof. Temtamy was regularly working in the clinic of Skeletal Dysplasias and Limb Malformations, the first specialized clinic initiated by her 20 years ago. She kept helping patients, disseminating hope, and contributing to various workshops, scientific meetings, and international conferences and publications. She was providing her younger colleagues, students, and her community with support, guidance, and scientific and life advices.

Professor Samia Temtamy will never be forgotten. Her guidance, enthusiasm, and passion for science along with her beautiful eyes and lovely smile will continue to inspire us, give us hope, and lead us on the way to continue the legacy she left behind, providing hope to our society.

Acknowledgements

All members of the Human Genetics and Genome Research Institute at the NRC greatly appreciate and acknowledge the sincere efforts of Prof. Samia Temtamy toward establishing a strong, state-of-the-art institute in our country. Her legacy will never be forgotten. May Allah bless her soul.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

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