

SYLVIA SAFWAT FAHMY

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Professional Summary

Lecturer of Molecular Genetics Medical Research Institute, Alexandria University, Egypt	Jan 2024 - Present
PhD visiting student at Manzini Lab Child Health Institute of New Jersey, Rutgers Robert Wood Johnson Medical School, NJ, USA	Jan 2022 - Jan 2023
Assistant lecturer of Molecular Genetics Medical Research Institute, Alexandria University, Egypt	Mar. 2018 - Jan 2024
Demonstrator of Human Genetics Medical Research Institute, Alexandria University, Egypt	Oct. 2012 - Mar. 2018
General practitioner Ministry of Health, Health unit of Sidi Gaber, Ministry of Health, Alexandria, Egypt	Mar. 2012 - Oct 2012

Clinical

Medical Research Institute clinic of Human Genetics Medical Research Institute, Alexandria University	November 13 - Present Alexandria Egypt
Nile of Hope Hospital	July 23- Present Alexandria Egypt

Education

Ph.D. Human Genetics Medical Research Institute, Alexandria University PhD thesis : Genetic Blueprint of Congenital Muscular Dystrophies in Egyptian Patients 1-year joint supervision scholarship at Rutgers University from Central Department of Missions Ministry of Higher Education	Dec 2023
Master of Human Genetics Medical Research Institute, Alexandria University Master thesis: A Study of the effect of GLUTATHIONE STRANSFERSE M1 and T1 polymorphic gene deletions on the susceptibility and response to BCG in Grade: Excellent.	2018 Alexandria, Egypt
Bachelor of Medicine and Surgery, MBBCh (MD) Faculty of Medicine, Alexandria University Grade: Excellent with Honor.	2010 Alexandria, Egypt

Publications

- Safwat, S., Flannery, K. P., El Beheiry, A. A., Mokhtar, M. M., Abdalla, E., & Manzini, M. C. Genetic blueprint of congenital muscular dystrophies with brain malformations in Egypt: A report of 11 families. *Neurogenetics*
- Flannery KP, Safwat S, Matsell E, et al. A novel missense variant in the ATPase domain of ATP8A2 and review of phenotypic variability of ATP8A2-related disorders caused by missense changes. *Neurogenetics*. 2024;25(4):425-433. doi:10.1007/s10048-024-00773-9
- S. G., Abdel-Hamid, M. S., AlAbdi, L., Safwat, S., Flannery, K. P., Dardas, Z., Fatih, J. M., Murali, C., Kannan, V., Lotze, T. E., Herman, I., Ammouri, F., Rezich, B., . . . Nguyen, L. N. Biallelic variation in the choline and ethanolamine transporter FLVCR1 underlies a severe developmental disorder spectrum. *Genetics in Medicine*, Volume 27, Issue 1.
- J. T., Hagen, M. T., Thong, M.-K., Mazlan, R. A. B., Tae, S. K., Kamphans, T., Meiswinkel, W., Li, J.-M., Javanmardi, B., Knaus, A., Safwat, S., Uwineza, A., Knopp, C., . . . Hsieh, T.-C. GestaltMatcher Database - A global reference for facial phenotypic variability in rare human diseases.
- Li, M., Fischer, J., Safwat, S., Shoman, W., Chazli, Y. E., Alter, S., Has, C., & Abdalla, E. (2023). Lipoid proteinosis: Novel ECM1 pathogenic variants and intrafamilial variability in four unrelated Arab families. *Pediatr Dermatol*, 40(1), 113-119.
- Broojeni JV, Safwat S, Bareke E, Majewski J, Abdalla E, Slim R. PLCZ1 mutation and failed IUI, ICSI, and spontaneous pregnancy loss: Insights from a case study. *Placenta*. 2024;154:e55
- Broojeni, J. V., Elmahdy, M., Mitchell, S., Rezaei, M., Saharan, A., Elhady, G., Safwat, S., Bareke, E., Abdelrazek & Slim, R. (2025). Lessons learned from the exome sequencing of nine cases of infertility and the way forward. *Reproductive BioMedicine Online*, 105359.

Conferences and Workshops

Genomic Variant Interpretation Workshop (Speaker)	2025
Alexandria University	Alexandria , Egypt
Pharmacogenetics overview and goals (Speaker)	2025
Alexandria University	Alexandria , Egypt
Drug-Induced Congenital Anomalies: A Call to Prophylaxis, Diagnosis, and Management (Speaker)	2025
Alexandria University	Alexandria , Egypt
Milestone in cancer research from genomics to practice (Speaker)	2023
Alexandria University	Alexandria , Egypt
MENA Metabolic and Genetic Conference	2023
	Virtual
American society of human genetics conference	2022
	Los Anglos, USA
The Eighth Annual Brain health institute Symposium	2022
Rutgers university	NJ , USA
Applications of genomics in health and social development (Speaker)	2021
Alexandria University	Alexandria , Egypt
Introduction to Bioinformatics Training 2019 - H3ABioNet	2019
The course 140 hours, combines theoretical and practical sessions.	University of Cape Town
What face and teeth can tell you about syndromes	2018
Prof Nik Kantaputra, professor of medical genetics, faculty of Dentistry, Thailan - Alexandria Regional centre	Alexandria , Egypt
2nd annual conference of laboratory department of MRI	2017
The Medical Research Institute	Alexandria , Egypt
DNA sequencing	2016
Microbiology department, Medical Research Institute	Alexandria , Egypt
Diagnostic cytogenetics workshop	2016
Cytogenetics lab , Human genetics department , Medical Research Institute	Alexandria, Egypt
An introduction to fluorescene in situ hybridization - Hands-on workshop	2015
Clinical Pathology department , faculty of Medicine	Alexandria, Egypt
Essential of clinical genetics workshop (Speaker)	2015
National research centre	Alexandria, Egypt
13th Asian European workshop on inborn errors of Metabolism	2015
National research centre	Cairo, Egypt

Languages

English	Excellent Command
French	Excellent Command
Arabic	Mother Tongue

Skill Highlights

Genetics	
PCR, qPCR, gel electrophoresis	Cytogenetics (FISH, karyotyping)
Genomics & Bioinformatics	
Whole-exome sequencing (WES), NGS analysis	BLAST, GATK, Exomiser
Variant annotation (ClinVar, gnomAD, HGMD, OMIM)	
Data Analysis	
R, SPSS, GraphPad Prism	Basic Python
Software & Tools	
Microsoft Office (Word, Excel, PowerPoint)	Linux
Reference management (EndNote, Mendeley)	
Research & Academic	
Scientific writing & publications	Conference presentations
Project management, teaching & mentoring	

Training courses

Orientation to Animal Care and Use

Rutgers University

Online Laboratory Safety

Rutgers University

Introduction to Bioinformatics Training - H3ABioNet

Online- University of Cape Town

An introduction to fluorescence in situ hybridization

Alexandria University***Activities & Affiliations*****Gene Curator****Mar 2022 - Present**

Member of Brain Malformations Gene Curation Expert Panel

ClinGen - Clinical Genome Resource

References**Prof. Dr. Chiara Manzini**

Associate Professor in Neuroscience and Cell Biology

Child Health Institute of New Jersey, Rutgers Robert Wood Johnson Medical School

Professor Ebtessam Abdalla

Professor of Human Genetics

Medical Research Institute, Alexandria University

Professor Amira Nabil Khalaf

Assistant professor of genetics

medical research institute, Alexandria university