Ibrahim Abdelrazek

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Profile

A hardworking ambitious medical geneticist, who is highly passionate about research in rare human genetics disorders. I am an established researcher with around 6 years' experience in clinical and molecular genetics. I am competent in various molecular laboratory techniques and Bioinformatics data analysis. Possessing excellent communication and teamwork skills, I demonstrate great perseverance and initiative. Not only I am always motivated to learn but also put great work into assimilating every piece of knowledge into practice. Furthermore, I have co-authored a few publications in high impact scientific journals.

Education and Training

MSc Human Genetics; Alexandria University, Nov 2023

Thesis Title: "Phenotypic and molecular characterization of Egyptian children with tuberous sclerosis in Alexandria, Egypt"

A one-year training program at a pediatric neurology center in Alexandria from 2021 to 2022, where I developed valuable skills in neurological and behavioral assessments for neurogenetic cases.

Bachelor's degree of Medicine and Surgery M.B. Bch. Alexandria University, 2015- Excellent with honours.

Professional & Academic experience

Assistant lecturer of Human Genetics: Department of Human Genetics, Alexandria University Dec 2023-current

As an assistant lecturer I have participated in the teaching of clinical and practical laboratory sessions for post graduate students. Besides, I am working in the genetic clinic and molecular genetic laboratory. Since Sept 2024, I have started a preparatory course for PhD.

Teaching Assistant of Human Genetics; Alexandria University March 2019- Nov 2023

I was trained in the Genetic clinic and Molecular genetic laboratory. I completed my MSc in Genetics by the end of 2023.

Skills

- **Clinical Skills:** Experienced clinical geneticist with deep understanding of inherited rare genetic disorders. Experience in studying genetics of rare disorders in different ethnic populations. Reporting research-based genetic results to clinicians. Interpreting and explaining genetic results.
- **Laboratory skills:** Basic laboratory safety procedures, record keeping and general lab skills. Experience in molecular biology and genetic testing procedures including PCR, QPCR, RT-

PCR, Sanger Sequencing, and cell culture.

- **Bioinformatics skills:** Experience in bioinformatics analysis of high throughput genetic data, using a range of pipelines, and dealing with large genetic datasets.
- **Teaching skills:** Experience in teaching and training colleagues on different laboratory techniques and bioinformatics analysis.

Publication

- Bobbi McGivern,…, **Ibrahim M. Abdelrazek,….,** Ingrid M. Wentzensen. Homozygous variants in EIF3K associated with neurodevelopmental delay, microcephaly, and growth retardation. HGG Advances 2025. DOI: 10.1016/j.xhgg.2025.100438
- Shaymaa Elsayed, Gehad A. Elmakkawy, **Ibrahim M. Abdelrazek, ….,** Ebtesam M. Abdalla. An Update on 3M Syndrome: Review of Clinical and Molecular Aspects and Report of Additional Families. American Journal of Medical Genetics Part A 2025 https://doi.org/10.1002/ajmg.a.64068
- Zeynep Yalcin, Zheng Gao, **Ibrahim M.Abdelrazek,…,** Rima Slim**.** Two Novel Protein-Truncating Variants in *NLRP2* and Their Functional Impacts on the Subcortical Maternal Complex. Clinical Genetics 2025. https://doi.org/10.1111/cge.14718
- Loisa Dana Bonde, **Ibrahim M.Abdelrazek,…,** Frederike Leonie Harms. Homozygous synonymous FAM111A variant underlies an autosomal recessive form of Kenny-Caffey syndrome. Journal of Human Genetics 2024. DOI: 10.1038/s10038-024-01301-1
- **Ibrahim M.Abdelrazek,….,** Sheetal Kumar. Acromesomelic Dysplasia With Homozygosity for a Likely Pathogenic BMPR1B Variant: Postaxial Polydactyly as a Novel Clinical Finding. Molecular Genetics & Genomic Medicine 2024. DOI: 10.1002/mgg3.70023
- Tess Holling, **Ibrahim M.Abdelrazek**,…., Kerstin Kutsche**.** A homozygous nonsense variant in the alternatively spliced VLDLR exon 4 causes a neurodevelopmental disorder without features of VLDLR cerebellar hypoplasia. Journal of Human Genetics 2024. DOI: 10.1038/s10038-024-01279-w
- Zeynep Yalcin, Manqi Liang, **Ibrahim M. Abdelrazek**…., Rima slim**.** A report of two homozygous TERB1 protein-truncating variants in two unrelated women with primary infertility. Journal of Assisted Reproduction and Genetics 2024. DOI: 10.1007/s10815-024-03031-x
- **Ibrahim M. Abdelrazek**, Tess Holling,…, Kerstin Kutsche. Craniofacial dysmorphism, skeletal anomalies, and impaired intellectual development syndrome-1 in two new patients with the same homozygous TMCO1 variant and review of the literature. European Journal of Medical Genetics Volume 66, Issue 3, March 2023, 104715
- H. Lesmann, S. Moosa, S…., **Ibrahim M. Abdelrazek**,…., T-C. Hsieh. A preprint under review in Nature genetics. GestaltMatcher Database - A global reference for facial phenotypic variability in rare human diseases. DOI: 10.1101/2023.06.06.23290887

Fellowships

Daniel Turnberg Travel Fellowship from Academy of Medical Sciences for a month internship at neuromuscular department, UCL. The research project was “Unraveling the molecular etiology of affected Egyptian families with congenital brain malformations” September 2023.

Presentations

- Oral presentation. ‘The Hidden Role of Genetics in Lymphatic Disease’ 1st lymphedema

Egyptian conference, Alexandria, Egypt June 2025.

- Oral presentation. 'From Mutation to Malformation: Understanding the Genetic Basis of Congenital Anomalies' Congenital Anomalies Integrated Care Congress May 2025 Alexandria, Egypt
- 13th Alexandria pediatric neurology conference. Oral presentation.' Targeted Transcript Analysis in Neurogenetic Disorders' Alexandria, Egypt April 2025
- Oral presentation 'Challenges in the Diagnosis of Rare Genetic Disorders Using Exome Sequencing: Experience from Egypt' Institute of Human Genetics, Hamburg-Eppendorf university, Hamburg, Germany- August 2024.
- 12th Alexandria pediatric neurology conference. Oral presentation. 'Implications of AI based facial analysis in diagnosing neurogenetic disorders', Alexandria Egypt, March 2023.
- 7th international conference, Medical Research Institute. Oral presentation. 'Implications of one hundred whole-exome sequences in an Egyptian cohort'. Alexandria, Egypt, Feb 2022.

Research projects

- **"Identification and characterization of the Molecular Basis of female infertility in Egypt".** An ongoing collaboration between Prof. Rima Slim team at McGill University and the departmentof Human Genetics at Alexandria University. March 2022-current.
- **"Use of High Throughput Sequencing for the Identification of Pathogenic Mutations responsible for Mendelian Disorders"** conducted through a twin agreement between theHuman Genetics Department, Alexandria University and 3billion company. Aug 2022
- **"A clinical and molecular genetic study of Egyptian patients with dysmorphic facial features"**An ongoing collaboration between Pedia study team at IGSB, Bonn University and the Human Genetic Department at Alexandria University. Oct 2022-current.
- **"Genetics of neurodevelopmental delay disorders in Paediatric Egyptian patients".** Research collaborations between Human Genetic Department, Alexandria University and Institute of Human Genetics in Hamburg-Eppendorf, Germany. 2022-current.

Workshops and courses

- **Interpreting Genomic Variation: Overcoming Challenges in Diverse Populations:** Wellcome Connecting Science online course from 29th July to 16th August 2024.
- **Ensembl REST API workshop**. Cambridge Bioinformatics training courses. Oct 2022
- **Tissue culture basics and techniques**. Medical Research Centre. Alexandria. May 2021.
- **Scientific writing**. Coursera online courses. May 2020

HIGHLIGHTS of MY EXPERIENCE

Clinical experience

As a medical doctor specialized in clinical genetics, I have been working in the genetic clinic foralmost 6 years in diagnosis and counselling of patients and families with different genetic disorders. Attending the Genetic clinic for more than 6 years with a weekly rate of

approximately20 cases, has provided me great clinical skills. My areas of expertise include dysmorphology, infertility,
skeletal disorders, multiple congenital anomalies, skin genetic disorders, ophthalmic genetics,intellectual disability and neuromuscular disorders. I developed and excelled in the skills of history taking, pedigree analysis, clinical genetic examination, case formulation, differential
diagnosis and deciding a management plan, requesting appropriate genetic testing and ultimately genetic counselling and recurrence risk estimation. I am familiar also with prenatal cases of genetic disorders and families with history of reproductive losses and perinatal deaths.

Laboratory experience

I have been working in the molecular genetic laboratory since September 2021. I am aware ofbasic laboratory safety procedures and general lab skills. I am confident in performing PCR, RT- PCR,
QPCR, Sanger Sequencing and Western Blot. I was trained on tissue culture.

My main interest was clinical and molecular characterization of genetic disorders. My aims in studyingrare disorders in the untested Egyptian population were to improve understanding of these highly
heterogeneous diseases and improve accuracy of genetic counselling to the affected families. In my early career, I was one of the members of research projects. Through these projects, not only did I improve mylaboratory skills but also upgraded my team-work abilities.

My research is focused on neurodevelopmental delay disorders with dysmorphic facies. The aim of the research, in collaboration with Prof. Peter Krawitz, the director of the Institute for Genomic Statistics andBioinformatics at Bonn University, Germany, is to improve the interpretation of genomic data through
Pedia approach that integrates multiple information from the molecular level and phenotypic level. Thiscollaboration was further emphasized by my visit to Prof. Peter Krawitz Lab in July 2022. It helped me
enrich my knowledge in molecular biology techniques. Moreover, I also helped on interpretation of results, variants prioritization and segregation analysis. This research resulted in two abstracts presented in international conferences and additional publications in
preparation.

I also had a collaboration with 3billion company to define the genetic etiology of 1300 patients with different rare disorders. Then now, Prof. Peter Krawitz team and me are doing the reanalysis of the rawdata of those patients for SNV and CNV to delineate any potential candidate novel genes.

Teaching

As a part of my duties as a previous teaching assistant and current assistant lecturer of Human

Genetics department I participated in the teaching of different courses to MSc students including teaching of Basic molecular genetics techniques,pharmacogenetics, Blood genetic disorders courses. I contributed to training courses and workshops to students in basic laboratory and molecular skills, basic clinical genetics and Reproductive genetics. I was one of the coordinators of department`s monthly scientific meetings.