CURRICULUM VITAE

Dr. Ebtesam Mohamed Abdalla

Ph.D. Human Genetics

PERSONAL INFORMATION

First name / Surname: Ebtesam Nasr

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EDUCATION AND QUALIFICATIONS

- **Medical Research Institute, Alexandria University** **2003**

  Ph.D. Human Genetics

- **Medical Research Institute, Alexandria University** M.Sc. **1996**
  Human Genetics

- **Faculty of Medicine, Alexandria University** **1988**

  M.B.B.Ch. degree

EMPLOYMENT HISTORY

- **AU, Medical Research Institute, Human Genetics Department** **Current**
  Professor of Human Genetics
- **KAU, Faculty of Medicine, Department of Genetic Medicine** **2016-2017**
  Professor of Medical Genetics
- **AU, Medical Research Institute, Human Genetics Department** **2015**
  Professor of Human Genetics
- **AU, Medical Research Institute, Human Genetics Department** **2009-2014**
  Assistant professor
- **AU, Medical Research Institute, Human Genetics Department** **2003-2009**
  Lecturer
- **AU, Medical Research Institute, Human Genetics Department** **1996-2003**
  Assistant lecturer
- **AU, Medical Research Institute, Human Genetics Department** **1990-1996**
  Demonstrator
- **AU, Faculty of Medicine, University Hospitals** **1989-1990**
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RESEARCH AND PUBLICATIONS

Thesis:

- "Early Detection and Management of Certain Treatable Inborn Errors of Metabolism" An assay submitted for partial fulfillment of Master degree in Human Genetics, Alexandria University, 1996

- "Genetic Screening for Sickle Cell Disease and G6PD Deficiency in newborn Infants" An assay submitted for partial fulfillment of Ph.D. degree in Human Genetics, Alexandria University, 2003

Research projects:

- "National Project for Prevention of Genetic Diseases & Congenital Anomalies in Newborns" National Academy of Sciences 1994-1995

- "National Project for Early Detection and Primary Intervention of Inherited Neuro-metabolic Diseases" NCCM & European Community 2006-2008

Publications:

1. Hellen Lesmann, Gholson Lyon, Pilar Caro, Ibrahim Abdelrazek, Shahida Moosa, Jean Tori Pantel, ……, **Ebtesam Abdalla**, Markus Nöthen, Peter Krawitz, Tzung-Chien Hsieh. Gestalt Matcher Database-a FAIR database for medical imaging data of rare disorders. ***Medrxiv : the Preprint Server for Health Sciences. 2024.***

2. Asier Iturrate, Frédéric Tran-Mau Them, Alain Verloes, ……., **Ebtesam Abdalla**, Christel Thauvin-Robinet, Victor L. Ruiz-Perez, Ange-Line Bruel. Expanding the phenotype associated with biallelic SCNM1 variants. ***Human Genomics, 2025.***

3. Jalal Vallian Broojeni, Mohamed Elmahdy, …….., **Ebtesam Abdalla**, Rima Slim. Lessons learned from the exome sequencing of nine cases of infertility and the way forward. ***Reproductive BioMedicine Online, 2025.***

4. Ghada Elhady, Asmaa K Amin, Asier Iturrate, ……, **Ebtesam Abdalla.** Genetic and Clinical Spectrum of Osteogenesis Imperfecta in an Egyptian Cohort with a High Rate of Lethal Phenotypes**.** ***Clinical Genetics, 2025***

5. Natascha Rosen, Tess Holling, Inken Junod, Malik Alawi, Heba Ossama, Rabab K. ElGhandour, **Ebtesam Abdalla**, Kerstin Kutsche. KREMEN1 variants associated with ectodermal dysplasia impair complex formation of KREMEN1 with DKK1 and LRP6 and attenuate WNT3A response***. Journal of Investigative Dermatology, 2025.***

6. Asmaa Amin, Sara El-Dessouky, Marwa Abd Elmaksoud, ……, **Ebtesam Abdalla.** Analysis of Copy Number Variants is an Important Consideration in Exome Sequencing. ***Clinical Genetics, 2025; 197(8):e64068.***

7. Shaymaa Elsayed, Gehad Elmakkawy, Ibrahim Abdelrazek, ……, **Ebtesam 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; 197(8):e64068.***

8. Zeynep Yalcin, Zheng Gao, Ibrahim M. Abdelrazek, Eric Bareke, Jacek Majewski, **Ebtesam Abdalla**, Seang-Lin Tan, Lei Li, Rima Slim. Two Novel Protein-Truncating Variants in NLRP2 and Their Functional Impacts on the Subcortical Maternal Complex'. ***Clinical Genetics 2025;108(2):179-183.***

9. Sara Eldessouky, Wessam Sharafeldin, Mona Aboulghar, ……, **Ebtesam Abdalla.** Integrating Prenatal Exome Sequencing and Ultrasonographic Fetal Phenotyping for Assessment of Congenital Malformations: High Molecular Diagnostic Yield and Novel Phenotypic Expansions in a Consanguineous Cohort. ***Clinical Genetics 2025; 108:33-48.***

10. Nagwa Meguid, Susan R. Ismail, Mona Anwar, Adel Hashish, Yuliya Semenova, **Ebtesam Abdalla**, Mohamed Taha, Amal Elsaeid, Geir Bjørklund. Gamma-aminobutyric acid and glutamate system dysregulation in a small population of Egyptian children with autism spectrum disorder. ***Metabolic Brain Disease, 2025; 40:146.***

11. Sara Eldessouky, Wessam Sharafeldin, Mona Aboulghar, ……, **Ebtesam Abdalla.** Fetal Phenotyping and Whole Exome Sequencing for Twelve Egyptian Families with Serine Biosynthesis Defect: Novel Clinical and Allelic Findings with a Founder Effect. ***Prenatal Diagnosis 2025: 45:204–217.***

12. Daniel G Calame, Jovi Huixin Wong, Puravi Panda, et al. Biallelic variation in the choline and ethanolamine transporter FLVCR1 underlies a severe developmental disorder spectrum. ***Genetics in Medicine, 2025: 27(1):101273.***

13. Loisa Dana Bonde, Ibrahim Abdelrazek, Lara Seif, Malik Alawi, Khaled Matrawy, Karim Nabil, **Ebtesam Abdalla**, Kerstin Kutsche, Frederike Harms. Homozygous

synonymous *FAM111A* variant underlies an autosomal recessive form of Kenny-Caffey syndrome. ***Journal of Human Genetics, 2025; 70, 87–97***

14. Ibrahim Abdelrazek, Alexej Knaus, Behnam Javanmardi, Peter Krawitz, Horn Denise, **Ebtesam Abdalla**, Sheetal Kumar. Acromesomelic dysplasia with homozygosity for a likely pathogenic BMPR1B variant: postaxial polydactyly as a novel clinical finding. ***Molecular Genetics & Genomic Medicine 2024: 12: e70023.***

15. Tess Holling, Ibrahim Abdelrazek, Ghada Elhady, Marwa Abd Elmaksoud, Seung Woo Ryu, **Ebtesam Abdalla**, 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: 69:623–628.***

16. Maryam Rezaei, Manqi Liang, Zeynep Yalcin, et al. Defects in Meiosis I contribute to the genesis of androgenetic hydatidiform moles. ***The Journal of Clinical Investigation 2024;134(22):e170669.***

17. Altunoglu U, Palencia-Campos A, Güneş N, et al. Altunoglu U, Palencia-Campos A, Güneş N, Turgut GT, Nevado J, Lapunzina P, Valencia M, Iturrate A, Otaify G, Elhossini R, Ashour A, K Amin A, Elnahas RF, Fernandez-Nuñez E, Flores CL, Arias P, Tenorio J, Chamorro Fernández CI, Güven Y, Özsu E, Eklioğlu BS, Ibarra-Ramirez M, Diness BR, Burnyte B, Ajmi H, Yüksel Z, Yıldırım R, Ünal E, **Abdalla E**, Aglan M, Kayserili H, Tuysuz B, Ruiz-Pérez V. Variant characterization and clinical profile in a large cohort of patients with Ellis-van Creveld syndrome and a family with Weyers acrofacial dysostosis. ***Journal of Medical Genetics, 2024: 61(7):633-644.***

18. Sylvia Safwat, Kyle Flannery, Ahmed El Beheiry, Mohamed Mokhtar, **Ebtesam Abdalla**, Chiara Manzini. Genetic blueprint of congenital muscular dystrophies with brain malformations in Egypt: A report of 11 families. ***Neurogenetics, 2024: 25, 93-102.***

19. Zeynep Yalcin, Manqi Liang, Ibrahim M Abdelrazek, Corinna Friedrich, Eric Bareke, Amira Nabil, Frank Tüttelmann, Jacek Majewski, **Ebtesam Abdalla**, Seang-Lin Tan, 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: 41:751-756.***

20. Sara Hisham Eldessouky, Rana Mohamed Abdella, Hassan Mostafa Gaafar, Mona Fouad, Sherin Mohamed Sobh, Maha M Eid, **Ebtesam Abdalla**, Dalia Samir Zolfokar. Prenatal Diagnosis of Fetal Ventral Wall Defects: Associated Anomalies and Chromosomal Aberrations. ***Evidence Based Women's Health Journal, 2023, 13: 395-404.***

21. Catarina Gonçalves, Josianne Carriço, Omneya Magdy Omar, **Ebtesam Abdalla**, Manuel Lemos. Hypoparathyroidism, Deafness and Renal dysplasia (HDR) syndrome caused by a GATA3 splice site mutation leading to the activation of a cryptic splice site. ***Frontiers in Endocrinology, 2023, 14: 1207425.***

22. Ibrahim Abdelrazek, Tess Holling, Frederike Harms, Malik Alawi, **Ebtesam Abdalla**, 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, 2023, 66(3):104715.***

23. Mingfeng Li, Kenneth Lay, Andreas Zimmer, Kristin Technau-Hafsi, Jasmine Wong, Antonia Reimer-Taschenbrecker, Jan Rohr, **Ebtesam Abdalla**, Judith Fischer, Bruno Reversade, Cristina Has. A homozygous p. Leu813Pro gain-of-function NLRP1 variant causes phenotypes of different severity in two siblings. ***British Journal of Dermatology, 2023, 188:259–267.***

24. Mingfeng Li, Judith Fischer, Sylvia Safwat, Walaa Shoman, Yasmine El-Chazli, Svenja Alter, Cristina Has, **Ebtesam Abdalla**. Lipoid proteinosis: Novel ECM1 pathogenic variants and intrafamilial variability in four unrelated Arab families. ***Pediatric Dermatology 2023, 40:113–119.***

25. Asier Iturrate, Ana Rivera-Barahona, Carmen-Lisset Flores, Ghada A Otaify, Rasha Elhossini, Marina L Perez-Sanz, Julián Nevado, Jair Tenorio, Juan Carlos Triviño, Francesc R Garcia-Gonzalo, Francesca Piceci-Sparascio, Alessandro De Luca, Leopoldo Martínez, Tugba Kalaycı, Pablo Lapunzina, Umut Altunoglu, Mona Aglan, **Ebtesam Abdalla**, Victor L Ruiz-Perez. Mutations in SCNM1 Cause Orofaciodigital Due to Minor Intron Splicing Defects Affecting Primary Cilia. ***American Journal of Human Genetics 2022, 109: 1–22.***

26. Christiane Bauer, Tess Holling, Denise Horn, Mário Laço, Ebtesam Abdalla, Omneya Magdy Omar, Malik Alawi, Kerstin Kutsche. Clinically Relevant KCNQ1 Variants Causing KCNQ1-KCNE2 Gain-of-Function Affect the Ca2+ Sensitivity of the Channel. ***International Journal of Molecular Sciences 2022, 23: 9690.***

27.Ebtesam Abdalla, Malik Alawi, Peter Meinecke, Kerstin Kutsche, Frederike L. Harms. Cardiofacioneurodevelopmental syndrome: Report of a novel patient and expansion of the phenotype. ***American Journal of Medical Genetics Part A 2022, 188A:2448–2453.***

28. Isabelle Jéru, Amira Nabil, Gehad El-Makkawy, Olivier Lascols, Corinne Vigouroux, **Ebtesam Abdalla.** Two Decades after Mandibuloacral Dysplasia Discovery: Additional Cases and Comprehensive View of Disease Characteristics. ***Genes 2021, 12 (10): 1508.***

29. Luigi Donato, **Ebtesam Mohamed Abdalla**, Concetta Scimone, Simona Alibrandi, Carmela Rinaldi, Karim Mahmoud Nabil, Rosalia D'Angelo, Antonina Sidoti. Impairments of Photoreceptor Outer Segments Renewal and Phototransduction Due to a Peripherin Rare Haplotype Variant: Insights from Molecular Modeling. ***International Journal of Molecular Sciences; 2021, 22(7): 3484.***

30. Luigi Donato, Concetta Scimone, Simona Alibrandi, **Ebtesam Mohamed Abdalla**, Karim Mahmoud Nabil, Rosalia D'Angelo, Antonina Sidoti. New Omics—Derived Perspectives on Retinal Dystrophies: Could Ion Channels-Encoding or Related Genes Act as Modifier of Pathological Phenotype? ***International Journal of Molecular Sciences; 2021, 22(1): 70.***

31. Hanan E. Shamseldin, Ranad Shaheen, Nour Ewida, Dalal K. Bubshait, Hisham Alkuraya, Elham Almardawi, Ali Howaidi, Yasser Sabr, **Ebtesam Abdalla**, et al. The morbid genome of ciliopathies: an update. ***Genetics in Medicine; 2021; 22(6): 1051-1060***.

32. Amr Shujaa-Addin, Mervat Hashish, Nahla Nazmy, Amany Srour, **Ebtesam Abdalla.** Detection of SHOX gene deletions in Egyptian children with idiopathic short stature using FISH. ***Meta Gene; 2020; 24: 100697.***

33. Eliza Thompson, **Ebtesam Abdalla**, Andrea Superti-Furga, William McAlisted, Lisa Kratz, Sheila Unger, Beryl Royer-Bertrand, Belinda Campos-Xavier, Laureane Mittaz-Crettol, Asmaa K. Amin, Cori DeSanto, David B. Wilson, Ganka Douglas, Beth Kozel, Marwan Shinaw. Lamin B receptor-related disorder is associated with a spectrum of skeletal dysplasia phenotypes. ***Bone; 2019; 120: 354-363.***

34. Ivan Ivanovski, Olivera Djuric, Stefano Giuseppe Caraffi, Daniela Santodirocco, Marzia Pollazzon, Simonetta Rosato, Duccio Maria Cordelli, **Ebtesam Abdalla**, et al. Phenotype and genotype of 87 patients with Mowat–Wilson syndrome and recommendations for care. ***Genetics in Medicine; 2018; 20(9): 965-975***.

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36. Cecilia Giunta, Matthias Baumann, Christine Fauth, Uschi Lindert, **Ebtesam M. Abdalla**, Angela F. Brady, et al. A cohort of 17 patients with kyphoscoliotic Ehlers–Danlos syndrome caused by biallelic mutations in FKBP14: expansion of the clinical

and mutational spectrum and description of the natural history. ***Genetics in Medicine 2018; 20(1): 42-54.***

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38. Ebtesam Abdalla, Gianina Ravenscroft, Louay Zayed, Sarah J. Beecroft, Nigel G. Laing. Lethal multiple pterygium syndrome: A severe phenotype associated with a novel mutation in the nebulin gene. ***Neuromuscular Disorders 2017; 27(6):537-541.***

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40. Ebtesam Abdalla, Oliver Bartsch, Danuta Galetzka, Ulrich Zechner. Novel Clinical Findings in the First Egyptian Case of Sotos Syndrome Caused by Complete Deletion of the NSD1 Gene. ***American Journal of Medical Genetics A 2017; 173A(4):1090–1093.***

41. Miriam Reuter, Hasan Tawamie, Rebecca Buchert, Ola Hosny Gebril, Tawfiq Froukh, Christian Thiel, ….., **Ebtessam Abdallah**, Heinrich Sticht, Dagmar Wieczorek, André Reis, Rami Abou Jamra. Diagnostic yield and novel candidate genes by exome sequencing in 152 consanguineous families with neurodevelopmental disorders. ***JAMA Psychiatry 2017; 74(3):293-299.***

42. Ragaa A. Ramadan, Lubna M. Desouky, Mai Moaaz, Mostafa A. Elnaggar, Mohamed Selima, Mohamed Samir, **Ebtesam Abdalla**. Association of vitamin D receptor and toll like receptor variants with colon cancer risk: A case control study in Egypt. ***Meta Gene 2017; 11:209–216.***

43. Morteza Seifi, Tim Footz, Sherryl Taylor, Ghada El-Hady, **Ebtesam M. Abdalla**, Michael A Walter. Novel *PITX2* gene mutations in patients with Axenfeld-Rieger syndrome. ***Acta Opththalmologica 2016***; ***94(7):e571-e579.***

44. Reddy R, Nguyen NM, Sarrabay G, Rezaei M, Rivas MC, Kavasoglu A, Berkil H, Elshafey A, **Abdalla E**, Nunez KP, Dreyfus H, Philippe M, Hadipour Z, Durmaz A, Eaton EE, Schubert B, Ulker V, Hadipour F, Touitou I, Fardaei M, Slim R. The genomic architecture of NLRP7 is Alu rich and predisposes to disease-associated large deletions. ***European Journal of Human Genetics 2016; 24:1445-1452.***

45. Agnieszka Gaczkowska, **Ebtesam M. Abdalla**, Karin M.L. Dowidar, Ghada M. Elhady, Pawel P. Jagodzinski, Adrianna Mostowska. De novo EDA mutations: Variable expression in two Egyptian families. ***Archive of Oral Biology 2016; 68:21-28.***

46. Karim M. Nabil, Lubna M. El-Desouky, **Ebtesam M. Abdalla**. Association of PPAR γ Pro12Ala and C1431T polymorphisms with type 2 diabetes and diabetic retinopathy in a sample of Egyptian patients***. Journal of Ophthalmic Clinical Research 2016; 3: 019.***

47. Ebtesam Abdalla & Israa Alaa-Eddin. Bilateral Fibular Dimelia with Mirror Foot: An Additional Case Report. ***Journal of Genetics Syndromes & Gene Therapy 2016; 7:2.***

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49. Ebtesam M. Abdalla, Louay H. Zayed, Noha M. Issa, Asmaa K. Amin. Fraser syndrome: Phenotypic variability and unusual findings in four Egyptian families. ***Egyptian Journal of Medical Human Genetics 2016; 17:233–238.***

50. Ebtesam Abdalla, Karim nabil, Ghada El-Hady. It's not Mccune-Albright Syndrome, It's Neurofibromatosis-1. ***Journal of Genetics Syndromes & Gene Therapy 2016; 7:1.***

51. **Ebtesam M. Abdalla**, Lubna M. El-Desouky, Nargues M. Hassanein. Post-mortem clinical examination by experienced clinical geneticists as an alternative to conventional autopsy for assessment of fetal and perinatal deaths in countries with limited resources. ***Turkish Journal of Pediatrics 2015;57:146-53.***

52. **Ebtesam M. Abdalla**, Marianne Rorbach, Céline Bürer, Marius Kraenzlin, Hazem Eltayeby, Mervat El-belbesy, Amira Nabil, Cecilia Giunta. Kyphoscoliotic type of Ehlers Danlos syndrome (EDS VIA) in six Egyptian patients presenting with a homogeneous clinical phenotype. ***European Journal of Pediatrics 2015;174:105–112.***

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55. Mahmoud R Fassad, Lubna M El-Desouky, Samir Asal, **Ebtesam M Abdalla**. Screening for the mitochondrial A1555G mutation among Egyptian patients with non-syndromic, sensorineural hearing loss. ***International Journal of Molecular Epidemiology & Genetics 2014;5:200-204***

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" The 10th European Pediatric Neurology Society Congress EPNS 2013, 25–28 September 2013, Brussels, Belgium "

60. Abdalla EM, Morsy HA. Mental retardation, short stature and synpoly-dactyly in a manifesting heterozygote of Bartsocas–Papas syndrome. ***Clinical Genetics 2013; 84: 300–301.***

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the autosomal-recessive form of popliteal pterygium syndrome. ***American Journal Human Genetics*** **2012; 90(1):76–85.**

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