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EDUCATION

School/College/University/Other	Degree obtained	Dates (from-to)
Faculty of medicine, Ain Shams University	Doctorate in medical genetics	1998-2001
Faculty of medicine, Ain Shams University	Master degree of medical genetics	1994-1997

EMPLOYMENT HISTORY

Faculty positions

Employer	Position	Dates (from - to)
Medical Genetics Department, Faculty of medicine, Ain shams university	Head of the department	October 2021- today
Medical Genetics Department, Faculty of medicine, Ain shams university	Professor of medical genetics	Feb. 2019- today
Pediatrics Department, Faculty of medicine, Ain shams university	Professor of medical genetics	Feb. 2012- 2019
Pediatrics Department, Faculty of medicine, Ain shams university	Assistant professor of medical genetics	2007- 2012
Pediatrics Department, Faculty of medicine, Ain shams university	Lecturer of medical genetics	2001-2007
Pediatrics Department, Faculty of medicine, Ain shams university	Assistant lecturer of medical genetics	1997-2001
Pediatrics Department, Faculty of medicine, Ain shams university	Resident of medical genetics	1994-1997

Others:

- Editor- in- Chief of Egyptian journal of Medical Human genetics. https://jmhg.springeropen.com/about/editorial-board

- President of the Egyptian society of Human Genetics

- A member of the editorial board of the Egyptian liver journal https://eglj.springeropen.com/about/editorial-board

MEMBERSHIP OF PROFESSIONAL ASSOCIATIONS

- Senior member of the Egyptian Expert committee e of Gaucher disease
- Member of the society of study of inborn errors of metabolism
- Member of the Egyptian society of Friends and Families of Genetics diseases
- Member of the Egyptian society of human teratology
- Member of Egyptian society of liver disease and research

FIELDS OF EXPERIENCE

- Clinical genetics
- Metabolic liver disease and genetic diseases involving the liver
- Inborn errors of metabolism (IEM)
- Cytogenetics

PUBLICATIONS:

International:

1. Cardiac and ocular manifestations in Egyptian patients with mucopolysaccharidoses *Eastern Mediterranean Health Journal.* Volume 7, No. 6, November 2001, 981- 991
2. Dorfman-Chanarin Syndrome in Egypt. American Journal of Medical Genetics. 2003; 212A: 75-78.
3. Evidence for Single origin of 35delG and delE120 mutations in the GJB2 in Antolia. Clin Genet. 2004:67:31-37.
4. Microdeletion of chromosome 22 in a series of patients with non- selective congenital heart defects. Proceedings of the 12th world congress on heart disease- New trends in Research, Diagnosis and Treatment. Vancouver, BC, Canada, July 16-19, 2005. Eds: Asher Kimchi. Mediamond international proceedings
5. Frequency of five thrombophilic polymorphisms in the Egyptian population. Turk J Hematol 2006; 23:100-103
6. A novel mutation in BAP/SIL1 gene causes Marinesco–Sjogren syndrome in an extended pedigree Clin Genet 2006: 70: 420–423

7. Molecular Genetic Analysis in Mild Hyperhomocystenemia: A common mutation in the Methylenetetrahydrofolate Reductase gene Associated with recurrent Cerebrovascular strokes *J. Med. Sci. 2004; 4 (2): 95-101*
8. Double homozygosity for mutations of AGL and SCN9A mimicking neurohepatopathy syndrome. *Neurology. 2008 Jun 10;70(24):2343-4.*
9. Mutational analysis of ATP7B gene in Egyptian children with Wilson disease: 12 novel mutations. *J Hum Genet. 2008;53(8):681-7.*
10. Mutational Analysis of the MEFV Gene in Egyptian Patients with Familial Mediterranean Fever. *Turk J Med Sci.* 2009; 39 (1):
11. Phenotype of apoptotic lymphocytes in children with Down syndrome. *Immunity and Aging Journal* 2009, 6:2
12. Adipsic hypernatremia and bilateral renal stones in a child with ectrodactyly-ectodermal dysplasia- cleft palate (EEC) syndrome. Genetic counseling 2010; 21 (2): 215-220
13. Ribosomal protein S19 - 631 insertion is an African-originated mutation] Turkish Journal of Hematology 27 (2) , pp. 123-124
14. Clinical and genetic characterization of Chanarin-Dorfman syndrome patients: first report of large deletions in the ABHD5 gene.. Orphanet J Rare Dis. 2010 Dec 1; 5:33
15. Cholestasis in patients with Cockayne syndrome and suggested modidied criteria for clinical diagnosis. Orphanet Journal of Rare Diseases 2011, 6:13
16. Phenotypic and genetic characterization of a cohort of pediatric Wilson disease patients. BMC Pediatr. 2011 Jun 17; 11:56
17. Mutations in KIF7 link Joubert syndrome with Sonic Hedgehog signaling and microtubule dynamics. J Clin Invest. 2011 Jul 1;121(7). pii: 43639. doi: 10.1172/JCI43639
18. Factor V G1691A (Leiden) is a major etiological factor in Egyptian Budd-Chiari syndrome patients] Turkish Journal of Hematology 28 (4) , pp. 299-305
19. Inherited thrombophilia in pediatric ischemic stroke: An Egyptian study. Pediatric Neurology 47 (2), pp. 114-118
20. TNF-α-308 G/A polymorphism | [TNF-α-308 G/A polymorphism in Egyptian budd-chiari syndrome patients] Turkish Journal of Hematology 29 (4) , pp. 420-421.
21. Prominent extensor truncal dystonia in Egyptian patients with Wilson's disease. Movement Disorders 2014, 29 (1): 151-153.
22. WNT1 mutations in families affected by moderately severe and progressive recessive osteogenesis imperfect. American Journal of Human Genetics 2014, 92: 590-597.
23. Autosomal dominant SCA5 and autosomal recessive SCA are allelic conditions resulting from SPTBN2 mutations. 2014; 22 (2): 286-288
24. Role of plasma amino acids and urinary organic acids in diagnosis of mitochondrial diseases in children. Pediatr Neurol. 2014 Dec;51(6):820-5
25. Non-manifesting AHI1 truncations indicate localized loss-of-function tolerance in a severe Mendelian disease gene. Hum Mol Genet. Hum Mol Genet. 2015 May 1;24(9):2594-603
26. Structural chromosomal abnormalities in couples with recurrent abortion in Egypt. Turk J Med Sci. 2015;45(1):208-13.
27. The Cockayne Syndrome Natural History (CoSyNH) study: clinical findings in 102 individuals and recommendations for care. Genet Med. 2016 May;18(5):483-93
28. A Novel Frameshift Mutation in SLC2A1 Associated with a Mild form of Glucose Transporter Type 1-Related Movement Disorder Journal of pediatric neurology: JPN 08/2015; 13(02):088-091.

29. Orthopedic manifestations of Proteus syndrome in a child with literature update Bone Reports, 2015: 3: 104-108
30. A case of infantile osteopetrosis: The radio clinical features with literature update. Bone reports 2016; 4: 11–16
31. Mutations in CDC45, Encoding an Essential Component of the Pre-initiation Complex, Cause Meier-Gorlin Syndrome and Craniosynostosis. Am J Hum Genet. 2016 Jul 7;99(1):125-38
32. Maladaptive Autophagy Impairs Adipose Function in Congenital Generalized Lipodystrophy due to Cavin-1 Deficiency. J Clin Endocrinol Metab. 2016 Jul;101(7):2892-90
33. Next-generation sequencing reveals the mutational landscape of clinically diagnosed Usher syndrome: copy number variations, phenocopies, a predominant target for translational read-through, and PEX26 mutated in Heimer syndrome. Molecular genetics & genomic medicine, 2017, 5: 531-52.
34. Frequency of inborn errors of metabolism screening for children with unexplained acute encephalopathy at an emergency department. Neuropsychiatric disease and treatment 2018; 14, 1715
35. Phenotypic characteristics of a tyrosinemia type I: an Egyptian cohort. QJM: An International Journal of Medicine. 2018. Volume 111, Issue suppl_1.
36. Relation of Cardia Dysfunction to Thyroid Stimulating Hormone in Children with Down Syndrome. European Journal of Human Genetics 26, 262-262, 2018
37. A rare type of skeletal dysplasia resembling rheumatoid arthritis. QJM: An International Journal of Medicine, 2020
38. Phenotypic characteristics of a tyrosinemia type I: an Egyptian cohort. QJM: An International Journal of Medicine 111 (suppl_1), hcy200. 165
39. MPI-CDG from a hepatic perspective: Report of two Egyptian cases and review of literature Journal of inherited metabolic disease report September 21
40. Disentangling molecular stratification patterns in GM1 gangliosides and mucopolysaccharidosis type IVB. Journal of medical genetics March 21
41. Skeletal radiology following hematopoietic stem cell transplantation in infantile osteopetrosis: an overlooked assessment tool. Bol. Med. Hosp. Infant. Mex. vol.78 no.5 México sep./oct. 2021 Epub 04-Oct-2021
42. Multicentric Osteolysis, Nodulosis, and Arthropathy in two unrelated children with matrix metalloproteinase 2 variants: Genetic-skeletal correlations. Bone reports, 2021; volume 15

Local publications:

1- Juvenile Hyaline Fibromatosis: case report. Egyptian Journal of Medical Human Genetics. 2002; 3 (1): 85-93
2- Emergency Management of Inherited Metabolic Diseases. Egyptian Journal of Medical Human Genetics. 2003; 4 (2): 95-103.
3- Fibrodysplasia Ossificans Progressive: A case report. Egyptian Journal of Medical Human Genetics. 2003; 4 (2): 43-47.
4- Dorfman-Chanarin Syndrome (DCS): Another Egyptian Patient. The Afro-arab Liver Journal. 2004; 3 (1): 61-65.
5- Extended Metabolic Screen in Sick Neonates and Children. Egyptian Journal of Medical Human Genetics. 2004; 5 (2): 1-7.
6- Bone Mineral Density in Children with Chronic Liver Disease the Egyptian Journal of Pediatrics2004; 21 (2): 271-279,

7- Down syndrome in Egypt. Egyptian Journal of Medical Human Genetics. 2004; 5 (2): 67-78.
8- Gaucher Disease with Pulmonary, Cardiac and Renal Involvement. Improvement on Fractionation of Cerezyme Dose. Egyptian Journal of Medical Human Genetics. 2004; 5 (2): 55-59.
9- Molecular Genetic Analysis in Mild Hyperhomocystenemia: A common mutation in the Methylenetetrahydrofolate Reductase gene Associated with recurrent Cerebrovascular strokes. J. Med. Sci. 2004; 4 (2): 95-101
10- Dyggve-Melchior_Clausen Syndrome: Case Report. Egyptian Journal of Medical Human Genetics. 2005; 6 (1): 67-72.
11- Hypomelanosis of Ito Associated with Neuroblastoma. Egyptian Journal of Medical Human Genetics. 2005; 6 (1): 73-79.
12- Prenatal onset infantile cortical hyperostosis (Caffey disease): an Egyptian report. The Egyptian Journal of Neonatology 2005; 6 (1):47-49
13- Cohen Syndrome. Egyptian Journal of Medical Human Genetics. 2005; 6 (2): 207-212.
14- Methylation Status of the KCNQ1OT and H19 genes in Beckwith-Wiedemann syndrome. Egyptian Journal of Medical Human Genetics 2005; 6 (1): 55-61.
15- Inherited Thrombotic Risk Factors in non-selected group of Egyptian population. Egyptian Journal of Medical Human Genetics. 2005; 6 (1): 55-61.
16- Gallbladder disease in children with Down syndrome Egyptian Journal of Medical Human Genetics. 2005.
17- Updated listing of mutation map at the Human phenylalanine locus among Egyptian population Egyptian Journal of Medical Human Genetics. 2006; 7 (1):15-22
18- Connexin 26 Mutations in Non-selected group of Egyptian Population The Egyptian Journal of Pediatrics 2005; 22 (2&3): 349-359
19- Femoral hypoplasia-unusual facies syndrome: femoral facial syndrome (FFS): A recessive severe skeletal form?? Egyptian Journal of Medical Human Genetics. 2006; 7 (2):251-254.
20- Moonlighting proteins: old proteins learning new tricks. Egyptian Journal of Medical Human Genetics. 2006; 7 (2):107-113.
21- Clinical evaluation of non-selected group of craniofacial dysmorphism Egyptian Journal of Medical Human Genetics. 2007; 8 (1)
22- Acrocallosal Syndrome (ACS) Egyptian Journal of Medical Human Genetics. 2007; 8 (1)
23- MURCS association: a case report. *Egypt. J. Med. Hum. Genet. Vol. 8, No.(2):219-224, Nov. 2007*
24- Triple A syndrome presenting with myopathy: an Egyptian patient: The Egyptian Journal of Medical Human Genetics: 2009, 10 (1): 105- 109
25- Therapeutic approaches to genetic disorders. Egyptian Journal of Medical Human Genetics: 2009, 10 (2): 118- 128
26- Maternal risk factors in young Egyptian mothers of Down syndrome Egyptian Journal of Medical Human Genetics: 2009, 10: (2): 144- 152
27- Progressive osseus heteroplasia: An Egyptian patient: Egyptian Journal of Medical Human Genetics: 2010, 11: (1):
28- Screening for subtle chromosomal rearrangements in an Egyptian sample of children with unexplained mental retardation: Egyptian Journal of Medical Human Genetics: 2011
29- Colchicine resistant FMF is not always true resistance Egyptian Journal of Medical Human Genetics: 2011
30- Connexin 26 (GJB2) Mutation in KID Syndrome: An Egyptian Patient Egyptian Journal of Medical Human Genetics: 2011

31- Outcome of enzyme replacement therapy in children with Gaucher disease: The Egyptian experience. Egyptian Journal of Medical Human Genetics: 2011
32- Multiple pterygium syndrome with marked pterygia of the fingers and MRI changes in the spine. Egyptian Journal of Medical Human Genetics. 2012, 13 (1): 107–113
33- Screening for subtle chromosomal rearrangements in an Egyptian sample of children with unexplained mental retardation. Egyptian Journal of Medical Human Genetics 2011, 12 (1): 63-68
34- Consanguinity and its relevance to clinical genetics Egyptian Journal of Medical Human Genetics:. 2013
35- Hypothyroidism could be the only manifestation of mitochondrial T8993C mutation in Leigh syndrome. Egyptian Journal of Medical Human Genetics: 2013
36- Familial Peters Plus syndrome with absent anal canal, sacral agenesis and sensorineural hearing loss: Expanding the clinical spectrum Egyptian Journal of Medical Human Genetics 2013; 14 : 421-428
37- Oral-facial-digital syndrome type II: Transitional type between Mohr and Varadi . Egyptian Journal of Medical Human Genetics 2013, 14 : 311-315
38- A severe form of cholestasis lymphoedema syndrome (Aagenaes syndrome) with progressive arthritis. Egyptian Liver Journal. 2014; 4(1):25-27
39- Maternal MTHFR C677T genotype and septal defects in offspring with Down syndrome: A pilot study . Egyptian Journal of Medical Human Genetics 2014; 15 : 39-44.
40- The blessing effect of an extra copy of chromosome 21. Egyptian Journal of Medical Human Genetics 2014; 15 : 209-210
41- Non-deletion mutations in Egyptian patients with Duchenne muscular dystrophy Egyptian Journal of Medical Human Genetics 2014; 15 (3)
42- Subclinical hypothyroidism in children with Down syndrome: To treat or not to treat??? Egyptian Journal of Medical Human Genetics 2014; 15(4)
43- Autosomal recessive ichthyosis with limb reduction defect: A simple association and not CHILD syndrome. Egyptian Journal of Medical Human Genetics 2016: 17 (3): 255-258
44- Wolman disease in patients with familial hemophagocytic lymphohistiocytosis (FHL) negative mutations Egyptian Journal of Medical Human Genetics2016 ; 17 (3): 277–280
45- Challenges in diagnosis and counseling of a family with two recessive neurometabolic disorders Egyptian Journal of Medical Human genetics 2016: 17 (3): 247-250
46- BH4 deficiency with unusual presentations: Challenges and lessons Egyptian Journal of Medical Human Genetics2016: 17 (3): 241-242
47- Treatment options for patients with Gaucher disease Egyptian Journal of Medical Human Genetics 2016 ; 17 (3): 281–285
48- Treatment-induced copper deficiency in two patients with Wilson's disease Egyptian Liver Journal 2016: 6 (1), 16-20
49- Neurofibromatosis type 1 and multiple sclerosis: Genetically related diseases Egyptian Journal of Medical Human Genetics 2016 In press

50- Pseudo achondroplasia in a child: The role of anthropometric measurements and skeletal imaging in differential diagnosis The Egyptian Journal of Radiology and Nuclear Medicine 2016 : In press
51- An Egyptian patient with Schwartz-Jampel syndrome type I and new ocular findings. Egyptian Journal of Medical Human Genetics 18 (4), 393-396
52- Reversal of skeletal radiographic pathology in a case of malignant infantile osteopetrosis following hematopoietic stem cell transplantation The Egyptian Journal of Radiology and Nuclear Medicine 2017; 48 (1), 237-243
53- Cardiomyopathy in Vici syndrome Egyptian Journal of Medical Human Genetics 2018; 19 (1), 49–50
54- Letter to the Editor: Are we missing fucosidosis? Egyptian Journal of Medical Human Genetics 2018; 19 (2), 151-152
55- Professor Rabah Mohamed Shawky, 1943–2021. Egyptian Journal of Medical Human Genetics 22 (1), 1-1