Name: Mahmoud Reza Ashrafi DOB: 22 11 1960 Married with one son Place of Birth: Tehran

Education: Pediatrician - Pediatric Neurology

- 1 Board Certification in Child Neurology 1999 2001
  Shahid Beheshti University of Medical Sciences / Mofid
  Children's Hospital. Tehran / Iran
  Mitochondrial encephalopathies: Dr Ghofrani
- 2 Board Certification in Pediatrics 1987 1991 Tehran University of Medical Sciences / Children's Medical Center /Tehran / Iran
- 3 Degree of Medicine (M.D) 1978 1987 Tehran University – School of Medicine. Tehran / Iran

### UNIVERSITY APPOINTMENT:

Assistant professor of Pediatrics 1992 – 2004

Associate professor of Pediatrics. Pediatric Neurology 2004 - 2010

Professor of Pediatrics. Pediatric Neurology Since March 2010

#### ADMINISTRATION:

- 1. Member of executive committee of International Congress of pediatrics; 1991
- 2. Member of executive committee of Iranian Journal of Pediatrics; 1997 2003
- 3. Chief of OPD, Children's Medical Center; 1993 1999
- 4. Chief of PICU, Children's Medical Center; 2001 2007
- 5. Vice chancellor of research and education Children's Medical Center; 2002 2016
- Vice chancellor of education, Department of Pediatric Tehran University of Medical Sciences; 2002 June 2006 and 2008 – 2012
- 7. Member of Pediatric Neurology Board Exams. 2008
- 8. Executive member of Iranian Child Neurology Association. 2001
- 9. Active member of International Child Neurology Association (ICNA). 2005
- 10. Executive member and Vice chancellor of Iranian Neurometabolic Association 2017
- 11. Vice chancellor of Research, Children's Medical Center; 2016
- 12. Vice chancellor of Research, Department of Pediatrics, Tehran University of Medical Sciences; 03-2019

#### **TEACHING EXPERIENCES**

Duties entailed lecturing on several courses developed for medical students and pediatrics residents. I was chief of the OPD of Children's Medical Center. I was responsible for case Presentations to medical students as well as being involved in the diagnosis and treatment of children with neurologic problems in pediatric neurology department and PICU. 1992 Date.

Child Neurologist. Pediatric Neurology Department, Children's Medical Center, T.U.M.S. Sep 2001 – Date

#### **RESEARCH EXPERIENCES**

Editorial Board Member of Journal of Sleep Sciences Editorial Board Member of Iranian Journal of Child Neurology Awards and honors

#### Publication

1 - Hypertension and it's emergency treatment in children. Journal of Daru & Darman. May 10 (112); 1993.

2 - Schwartz-Jampel Syndrome and presentation of two cases from Children Medical Center. The Journal of Faculty of Medicine 56 the Year No 3; 1998.

3 - Status Epilepticus. Iranian Journal of Pediatrics. Dec Vol 103; 1998.

4 - Seckel Syndrome: A case report. Iranian Journal of Pediatrics. Aug Vol 102; 1998.

5 - Ohtahara Syndrome and IVF: A case report. Acta Medica Iranica. Vol 38 No 1; 2000.

6 - Study of breath holding spells and its triggering factors in Children Medical Center. The Journal of Faculty of Medicine 58 the Year No 1; 2000

7 – Auditory Brainstem Response (ABR) in Hyperbilirubinemic. Medical Journal of the Islamic Republic of Iran, Vol 16 No 2 Aug; 2002.

8 – Comparison of prevalence rates of obesity according to NCHS and NHANES among 7-10 old school children in Tehran. (Abstract), Annals of Nutrition & Metabolism; 45 (supplement 1) 227 – 228; 2001.

9-Clinical and Paraclinical study of Mitochondrial encephalopathies in Mofid Children's Hospital. Pejouhesh, Shahid Beheshti University of Medical Sciences Vol 26 No 3; 2002.

10 – Leigh Syndrome: Clinical and Paraclinical study of 15 cases. Acta Medica Iranica, Vol 40 No 4; 2002

11 – Chloride Channel Myotonia: Study of 5 cases. Iranian Journal of Pediatrics Vol 12 No 3; 2002.

12 – Trial of Clinical efficacy of Piracetam in treatment of Breath-holding Spells. Iranian Journal of Pediatrics Vol 12 No 4; 2002.

13 – Neurological manifestations of Shigellosis in Bahrami Children's Hospital. The Journal of Faculty of Medicine No 5; 2003.

14 – Anticonvulsant Drugs Effects on Thyroid function tests. Iranian Journal of Pediatrics Vol 13 No 2; Oct 2003.

15 – Mother awareness of civil vaccination program. Iranian Journal of Pediatrics Vol 13 No 2; Oct 2003.

16 – Sjogren – Larsson syndrome: A case report. Iranian Journal of Pediatrics Vol 14 No 2; Nov 2004.

17 – Sodium Valproate versus Propranolol in pediatric migraine Prophylaxis. European Journal of Pediatric Neurology (2005) 9, 333-338.

18 – Extensive Mongolian Spot: A clinical Sign Merits Special Attention. Pediatr Neurol 2006; 34: 143-145.

19 – Blood pressure distribution among healthy schoolchildren aged 6-13 years in Tehran. Eastern Mediterranean Health. Journal, Vol. 11, Nos 5/6, 2005.

20 - Study of Antiepileptic Drugs effects on bone metabolism. Iran J Pedatr, Vol 15, No 4, Dec 2005

21 – Myoclonic encephalopathy. Iran J Pediatr, Oct 2005, Vol 15, No 3

22 - Septo-Optic dysplasia syndrome: a case report. Iran J Pediatr, Vol 16, No 1, Mar 2006

23 – The prevalence of hypoparathyroidism among patients with major thalassemia aged above 10 years. Iran J Pediatr, Jul 2005, Vol 15, No 2

24 - Bilateral basal ganglia involvement in a patient with Griscelli Syndrome. European Journal of Pediatric Neurology 10(2006) 207-209.

25- Selenium and Intractable Epilepsy: Is There Any Correlation. Pediatr Neurol 2007; 36(1): 25-29.

26-Breath-holding spells: An analysis of 43 cases. Iranian Journal of Child Neurology Vol.1 No. 2 Oct.2006.

27-Can Homeopathy used simultaneously with Rehabilitation be effective in the Treatment of children with Cerebral Palsy. Iranian Journal of Child Neurology Vol.1 No. 3 Feb. 2007.

28- Moebius Syndrome and Corpus Callosum Agenesis; A Case Report. Acta Medica Iranica, Vol 44, No 6; 2006.

29- Association Between Total Serum Bilirubin Level and Manifestations of Kernicterus. Acta Medica Iranica, Vol44, No 6; 2006.

30-A Probable Causative Factor for an old problem: Selenium and Glutathione Peroxidase Appear to play important Roles in Epilepsy Pathogenesis. Epilepsia, Vol 48, No 9, 2007

- 31-Sleep Patterns and Sleep Problems among Preschool and School-aged Group Children in a Primary Care Setting. Iran J Ped Sep 2007
- 32-Reliability and validity of Persian version of BEARS pediatric sleep Questionnaire. Indian J sleep medicine 2008; 3.1, 14 19
- 33-Use of A-Interferon, Amantadin and Isoprinosine in SSPE: Comparing the effectiveness. Iranian Journal of Child Neurology Vo2 No 2 Feb. 2008.
- 34-Clinical Short-Term Outcome of Guillaine-Barre Syndrome in Children. Iranian Journal of Pediatrics. Vol 18 (No1), Mar 2008.
- 35-Incidence and common locations of Mongolian spots in newborns in Shariati Hospital. Tehran University Medical Journal; Vol.65, Supplement 1, 2007: 54-58.

36-Skull defects, alopecia and distinctive facies: A new syndrome. Clinical Dysmorphology 2008 17 203-205.

37- Neuronal Migration Disorder; A Case Report of Subcortical Band Heteropia Associated with Corpus CallosumAgenesis. Iranian Journal of Radiology 2008; 5(3):155-158

- 38-Neuromuscular Disease in Childhood, a Clinicopathological Study in Iran. Research Journal of Biological Sciences 3 (12) :1403-1407,2008.
- 39- Hemi Convulsion-Hemiplegia-Epilepsy Syndrome. Iran J Child Neurology Vol. 2 No. Oct 2008: 55-58
- 40- Executive dysfunction in treated phenylketonuric patients. Eur Child Adolesc Psychiatry 2009.

41- Melatonin versus chloral hydrate for recording sleep EEG.

European Journal of Pediatric Neurology. 14. 2010 235-238.

42- Adverse drug and medical instrument reactions in a pediatric intensive care unit. Pharmacology and Drug Safety 2009; 18: 761-762.

43-Effects of anti-epileptic drugs on serum immunoglobulin levels in children Acta Neurol, Belg, 2010, 110, 65-70 44-Effects and usability of buccal midazolam in controlling acute prolonged convulsive seizures in children. European Journal of pediatrics Neurology. 14(2010): 434-438

45-Incidence of Mongolian spots and its common sites at two university hospitals in Tehran, IRAN. Pediatric Dermatology. Vol. 27 No. 4 2010

46-Vaccine-associted paralytic poliomyelitis in immunodeficient children, Iran, 1995-2008 Emerging infectious Diseases. Vol. 16. No. 7, Jul 2010.

47- Severe cutaneous reactions caused by barbiturates in seven Iranian children. International Journal of Dermatology 2009, 48; 1254-1261

48- Effect of Antiepileptic-Drugs on serum level of IgG subclasses. Iran J Pediatr. Sep 2010 Vol 20 No 3 Pp 269-276.

49- Approach to Mental Retardation and Global Developmental Delay. Iranian Journal of Child Neurology. Vol 5 No1 winter 2011.

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51- Anterior Fontanelle size in Healthy Iranian Neonates on the first Day of life Acta Medica Iranica. 2011; 49(8):543-546.

52-Pelizaeus-Merzbacher Disease: The first genetically approved case report from Iran. Iranian Journal of Pediatrics. Sep 2011; Vol 21 (No 3), Pp 395-398.

53 –Psychomotor delay, a possible rare presentation of Moyamoya Disease. Iranian Journal of Radiology. 2011 8 (2). 54-L2 Hydroxy glutaric aciduria is a clinical indicator of Leukodystrophy. A case report. Iranian Journal of Child Neurology. 2011 5 (4); 37-38.

55-Efficacy and safety of cinnarizine in the prophylaxis of migraine headache in children: an open, randomized comparative trial with propranolol. ACTA Neurological Belgica. DOI 10, 10007/s 13760-012-0011-7.

56- Efficacy and safety of intravenous sodium valproate versus phenobarbital in controlling convulsive status epilepticus and acute prolonged convulsive seizures in children: a randomized trial. European Journal of Pediatric Neurology. Article in press.

57-Ataxia-telangiectasia or neurologic Wilson's disease: when strong family history becomes a diagnostic bias. Acta Neurol Belg published online :24 July 2012

58-Sleep Inducing for EEG Recording in Children: A Comparison between Oral Midazolam and Chloral Hydrate. Iranian Journal of Child Neurology. Vol 7 No 1 winter 2013.

59-Pseudotumor cerebri in a case of ulcerative colitis with sagittal sinus thrombosis. Iran J Pediatr Vol 23 No 1 Feb 2013.

60-Alexander Disease: Report of Two Unrelated Infantile Form Cases, Identified by GFAP Mutation Analysis and Review of Literature; The First Report from Iran. Iran J Pediatr Aug 2013; Vol 23 (No 4), Pp: 481-484

61-Aplasia Cutis Congenita (ACC) and seizure in a premature neonate. Iranian Journal of neonatology 2014; 5(1) 54-57

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63- Clinical Trial of Vigabatrin as Adjunctive Therapy in Children with Refractory Epilepsy Iranian Journal of Pediatrics, Volume 23 (Number 6), December 2013, Pages: 653-658

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Blind Placebo-Controlled Randomized Trial Pediatr Neurol 2014; 51: 503-508

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77- Brain on FIRES: Super Refractory Seizure in a 7 yr Old Boy. Iran J Child Neurol. AUTUMN 2016 Vol 10 No 4.

78- The effects of classic ketogenic diet on serum lipid profile in children with refractory Seizures. Acta Neurol Belg (2016) 116:529–534.

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80- The efficacy of the ketogenic diet in infants and young children with refractory epilepsies using a formula-based powder. Acta Neurol Belg. Published online 07 Dec 2016.

81- Clinical and Molecular Study of NPC in Iran: Report of 5 Novel Mutations. Iran J Child Neurol. 2015 Autumn vol 9 no 4 suppl.1.

82- Childhood leukodystrophies: A literature review of updates on new definitions, classification, diagnostic approach and management. Brain & Development 39 (2017) 369–385.

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84-The Efficacy of Cerebro lysin in Improvement of Spasticity in Children with Cerebral palsy: A Clinical Trial. Iran J Pediatr 2018 February; 28(1): e60840.

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86- The Relationship between Emotional Content and Word Processing in Normal Persian Speaking Children. Iranian Journal of Child Neurology, Autumn 2018 Vol. 12 No. 4

87- Overexpression of SMN2 Gene in Motoneuron-Like Cells Differentiated from Adipose-Derived Mesenchymal Stem Cells by Ponasterone a Journal of Molecular Neuroscience Published online 7 Dec 2018

88- Clinical presentation and outcome in infantile Sandhoff disease: a case series of 25 patients from Iranian neurometabolic bio registry with five novel mutations Orpha net Journal of Rare Diseases (2018) 13:130

89-The First Report of Relative Incidence of Inherited White Matter Disorders in an Asian Country Based on an Iranian Bio registry System Journal of Child Neurology (2018)

90-Hypomyelinating Leukodystrophy with Spinal Cord Involvement Caused by a Novel Variant in RARS:

Report of Two Unrelated Patients Neuropediatric

91- RNASET2-deficient leukoencephalopathy mimicking congenital CMV infection and Aicardi-Goutières syndrome: a case report with a novel pathogenic variant Orpha net Journal of Rare Diseases (2019) 14:184

92- Levetiracetam for prophylactic treatment of pediatric migraine: A randomized double-blind placebo-controlled trial Cephalalgia June 1, 2019 1–9

93-Clonidine Versus Chloral Hydrate for Recording Sleep EEG in Children. Iran J Child Neurol. Winter 2020;14(1): 85-92

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96-Neuralgic Amyotrophy with Cervical Root and Cranial Nerves Involvement in a Child Iran J Child Neurol. Autumn 2019; 13(4): 185-191

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99- We need well-designed multicenter studies to investigate neurologic manifestations of coronavirus disease 2019. Curr J Neurol 2020; 19(3): 150-1

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102- Novel disease-causing variants in a cohort of Iranian patients with metachromatic leukodystrophy and in silico analysis of their pathogenicity. Clinical Neurology and Neurosurgery 201 (2021) 106448.

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- 105- Clinical and imaging outcomes after intrathecal injection of umbilical cord tissue mesenchymal stem cells in cerebral palsy: a randomized double-blind sham-controlled clinical trial, Stem Cell Research & Therapy (2021) 12:439
- 106- An open-label phase 1 clinical trial of the allogeneic side population adipose-derived mesenchymal stem cells in SMA type 1 patients, Neurological Sciences, 2021
- 107- The safety and efficacy of umbilical cord blood mononuclear cells in individuals with spastic cerebral palsy: a randomized double-blind sham-controlled clinical trial, BMC Neurology, (2022) 22:123
- 108- The quality of life in children with spinal muscular atrophy: a case-control study, BMC Pediatrics (2022) 22:708
- 109- Novel phenotype and genotype spectrum of WDR62 in two patients with associated primary autosomal recessive microcephaly, Irish Journal of Medical Science (1971 -)
- 110- Characteristics of disease progression and genetic correlation in ambulatory Iranian boys with Duchenne muscular dystrophy, BMC Neurology, (2022) 22:162
- 111- Recommendations for Infantile-Onset and Late-Onset Pompe Disease: An Iranian Consensus, Frontiers in Neurology, September 2021 | Volume 12 | Article 739931
- 112- A cross-sectional, prospective ocular motor study in 72 patients with Niemann-Pick disease type C, A crosssectional, prospective ocular motor study in 72 patients with Niemann-Pick disease type C, European Journal of Neurology 2021
- 113- Genetic Analysis of Forty MLPA-Negative Duchenne Muscular Dystrophy Patients by Whole-Exome Sequencing, Journal of Molecular Neuroscience, 2022
- 114- Childhood Guillain–Barre syndrome in the SARS-CoV-2 era: Is there any causative relation? Clinical Case Reports, 2022
- 114- Defective complex III mitochondrial respiratory chain due to a novel variant in CYC1 gene masquerades acute demyelinating syndrome or Leber hereditary optic neuropathy, Mitochondrion, 60 (2021) 12–20
- 115- Epilepsia Partialis Continua a Clinical Feature of a Missense Variant in the ADCK3 Gene and Poor Response to Therapy, Journal of Molecular Neuroscience (2022) 72:1125–1132
- 116- Follow-up of 25 patients with treatable ataxia: A comprehensive case series study, Clin Case Rep. 2022;10: e05777
- 117- Clinical and Molecular Findings of Autosomal Recessive Spastic Ataxia of Charlevoix Saguenay: an Iranian Case Series Expanding the Genetic and Neuroimaging Spectra, The Cerebellum, June 2022

- 118- A comprehensive overview of SMN and NAIP copy numbers in Iranian SMA patients, Scientific Reports | (2023) 13:3202
- 119- The First Report of Iranian Registry of Patients with Spinal Muscular Atrophy, Journal of Neuromuscular Diseases 10 (2023) 211–225
- 120-Virtual and In-person Electroencephalography (EEG) Training among Pediatric and Adult Neurology Residents during the COVID-19Pandemic. Iran J Pediatr. In Press (In Press): e128717 Published online 2023 June 7.
- 121-Spinal Muscular Atrophy with Progressive Myoclonic Epilepsy (SMA-PME): three new cases and review of the mutational spectrum. Italian Journal of Pediatrics (2023) 49:64
- 122-Efficacy and safety of miglustat in the treatment of GM2 gangliosidosis: A systematic review. Eur J Neurol. 2023; 00:1–27.
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- 124-Novel homozygous frameshift variant in the ATCAY gene in an Iranian patient with Cayman cerebellar ataxia; expanding the neuroimaging and clinical features: a case report, BMC Medical Genomics (2023) 16:226.
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- 126-Clinical application of next generation sequencing for Mendelian disease diagnosis in the Iranian population. npj Genomic Medicine (2024) 9:12
- 127- A comparative study on prophylactic efficacy of cinnarizine and amitriptyline in childhood migraine: a randomized double-blind clinical trial. Cephalalgia 2024, Vol. 44(4) 1–1128-
- 128- The genetic basis of early-onset hereditary ataxia in Iran: results of a national registry of a heterogeneous population. Mahdieh et al. Human Genomics (2024) 18:35
- 129- An overview of early-onset cerebellar ataxia: a practical guideline. Sareh Hosseinpour, Maryam Bemanalizadeh,
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## Books

- 1- Approach to Floppy infants & Behavioral disorders in children.
- 1. Lecture Notes on Pediatrics. Tehran University of Medical Sciences. 1st. edition; 1996.
- 2- Step by step treatment in pediatrics.
  - Ashrafi M. R, Azimikia A (editors) .1st edition 1997.
  - Reprint 2000, 2003 and 2005.
  - Ashrafi M. R, Moghtaderi M, Nikkhah A, Azimikia A (editors) .2st edition (2021)
- 2. Encephalopathies & Neurodegenerative diseases of Childhood.
- Textbook of Pediatrics; Shahid Beheshti University of Medical Sciences 1st. edition; 2004. T.U.M.S, 1990 1991.
- 4. Therapeutic interventions in a child with Febrile Convulsion. Febrile Convulsion Feb 2008.
- 5. Mitochondrial Myopathies in: Neuromuscular Disorders in Children. Nov. 2008
- 6. Brain and Neurologic Disorders of Children for Parents. First edition. Sep 2012.
- 7. Brain and Neurologic Disorders of Children. First edition. Feb 2013
- 8. Step by step diagnosis of intellectual disability in children. Feb 2015

# Membership of the Scientific Organization and Association:

Iranian Child Neurology Society Iranian Neurometabolic Society Iranian Neurogenetic Society Iranian Headache Society International Child Neurology Association (ICNA) The Asian Oceanian Child Neurology Association (AOCNA) Iran SMA Association