



AMIRA TAHER MASRI

Dean of Deanship of Scientific Research

Professor of Pediatric Neurology

The University of Jordan

CONTACT INFORMATION

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WORK EXPERIENCE

Dean of Deanship of Scientific research: University of Jordan
2023-Present

Professor and Consultant of child Neurology: University of Jordan /Jordan University Hospital
2001-Present

Head of the pediatric neurology division at Jordan University Hospital
2014-Present

Chairman of the pediatric department at the University of Jordan
2007-2014

Part time Associate Professor of child Neurology ; child neurology course for students :
Mutah University, Amman-Jordan
2005-2007

Part time Associate professor of child Neurology; child neurology course for students:
Hashemite University, Amman-Jordan
2009-2014

EDUCATION

Pediatric Neurology- Montpellier–France
1998-2000

General Pediatrics - Jordan University Hospital& University of Jordan
1994 -1998

Internship: AL Bashir Hospital, Amman – Jordan
1993-1994

Bachelor of Medicine & surgery, University of Jordan
1987-1993

LANGUAGES

Arabic: fluent written and spoken

English: fluent written and spoken

French: fluent written and spoken

CERTIFICATIONS

Pediatric Neurology diploma (DIU and AFSA) University of Montpellier–France
2000

Diploma in EMG and Neuromuscular Diseases University of Montpellier-France
2000

Higher specialization degree in Pediatrics University of Jordan
1998

Jordanian Board of Pediatrics
1998

United States medical license examination ECFMG certificate
1996

Bachelor of Medicine & Surgery, Faculty of Medicine (MBBSB) - University of Jordan
1993

Certification in French language exams of the French embassy in Jordan
(1983) Brevet (1985) Certificat

PROFESSIONAL EXPERIENCE

Examiner at the Faculty of Medicine, The University of Jordan for undergraduate and postgraduate students including OSCE exams

2001-Present

Examiner at the Faculty of Medicine of Jordan University of Science & Technology, Mutah University, Hashemite University

2001-Present

Acting member at the Jordan Medical Council for the Jordanian Board exam in Pediatric neurology

2014-Present

Examiner at the Jordan Medical Council for the Jordanian Board exam in General Pediatrics

2007-present

Acting member at the Arab Medical Council for the Arab Board exam in General Pediatrics

2007-2014

Examiner at the Arab Medical Council for the Arab Board exam in General Pediatrics

2007-Present

Examiner at the Arab Medical Council for the Arab Board exam in Pediatric neurology

2018-Present

Academic director of the internship rotation in the pediatric department

2001-2002

Program director for the pediatric residency program

2002-2004

Program director for undergraduate students

2004-2006

Chairman of pediatric department at the University of Jordan

2007-2014

Head of the pediatric neurology division at Jordan University Hospital

2014-Present

Program director for the pediatric neurology fellowship program

2014-Present

Tempus program Trainer after taking a one-week work shop and doing a certifying Exam

2005

Others

King Abdullah II Fund for development: member of the technical committee

2021-Present

Scientific research support fund: member of technical committee

2016-2017

HOSPITAL AND ACADEMIC COMMITTEES

Current committees:

Institutional review board committee: Jordan university hospital

2015-Present

Head of the research committee at the Faculty of medicine-The University of Jordan

2015-Present

Previous committees:

Ethics committee

Patients 'rights committee

Privileges and credentials committee

Quality committee

Higher education committee

Residency training committee

Internship training committee

Students training committee

Pharmacy committee

Library committee

TRAINING COURSES AND WORKSHOPS

Scientific research skills, center of consultation, technical services and studies, Amman-Jordan

The Evidence-Based Medicine workshop, Center of Educational development, Amman-Jordan

Training Trainers for Tempus and other programs (TTT), Amman-Jordan

Workshops on writing research projects proposals, The Higher Council for Science and Technology (HCST), Amman-Jordan

Human Resources management. Center of consultation, Amman-Jordan

Leadership course, Consultation center, Amman-Jordan

Entrepreneurship training workshop, Empretec Jordan program, Amman-Jordan

Women's Business Leadership program, organized between BDC Jordan and THUNDERBRIDE-School of Global Management -USA

Others

Ultrasound-Guided BTX-A Injection Techniques Workshop

MEMBERSHIP

The Jordan Medical Council

Jordanian Neuroscience Society

Jordanian Epilepsy Society

European Pediatric Neurology Society

International child neurology association

Asian child neurology association

COMMUNITY SERVICES

Member of technical steering committee for developing National standards for Diagnostic Centers for persons with disabilities in Jordan that was issued by the higher council for the Rights of Persons with Disabilities and Health Care Accreditation Council (HCAC)

2018

Trainer for health workers in Ministry of Health on National Standards for Diagnostic centers for Persons with disabilities in Jordan

2019

Member of a steering committee to develop clinical guidelines for diagnosing people with intellectual disabilities and Autism Spectrum Disorders

2022

Acting member of several committees at the ministry of health related to improving services and health of children with neurological diseases

2007- present

Member of Al Hussein Society Jordan center for training and inclusion

2007- present

Education of the general community through lectures, workshops, and TV presentations regarding the manifestations, causes, treatment and prevention of neurodisabilities in children

Awards

Samia Mango award for distinguished researcher 2015

Jordan Medical Association Award for Outstanding Physicians in the field of child neurology 2012.

PUBLICATIONS

Beaman, M. Makenzie, Lucia Guidugli, Monia Hammer, Chelsea Barrows, Anne Gregor, Sangmoon Lee, Kristen L. Deak, Marie T. McDonald, Courtney Jensen, Maha S. Zaki, **Amira T. Masri**, Charlotte A. Hobbs, Joseph G. Gleeson, and Jennifer L. Cohen **2023**. "Novel Association of Dandy–Walker Malformation with CAPN15 Variants Expands the Phenotype of Oculogastrointestinal Neurodevelopmental Syndrome." *American Journal of Medical Genetics, Part A (May)*:2757–67.

Nasir, Arwa, **Amira Masri**, and Laeth Nasir. **2023**. "Pediatricians' Perspectives on Childhood Behavioral and Mental Health Problems in Jordan." *Middle East Current Psychiatry* 30(1).

Masri, Amira T., Liyana Oweis, Majd Ali, and Hanan Hamamy. **2023**. "Global Developmental Delay and Intellectual Disability in the Era of Genomics: Diagnosis and Challenges in Resource Limited Areas." *Clinical Neurology and Neurosurgery* 230(May):107799.

Masri, Amira T., Arwa K. Nasir, Aya G. Irshaid, Fatima Y. Irshaid, Farah K. Alomari, Faisal A. Khatib, Abdelkarim A. Al-Qudah, Omar A. Nafi, Miral A. Almomani, and Mahmoud A. Bashtawi. **2023**. "Autism Services in Low-Resource Areas." *Neurosciences* 28(2):116–22.

Al Jaber, Mira, Raghad Shihadat, and **Amira Masri**. **2022**. "Post SARS-CoV-2 Guillain-Barré Syndrome in a Child: Case Report and Review of the Literature." *Child's Nervous System* 38(10):2011–16.

Beaman, M. Makenzie, Lucia Guidugli, Monia Hammer, Chelsea Barrows, Anne Gregor, Marie

McDonald, Courtney Jensen, Sangmoon Lee, Maha Zaki, **Amira Masri**, Joseph Gleeson, and Jennifer L. Cohen. **2022**. "EP081: Dandy Walker Malformation in Three Unrelated Families with Biallelic Variants in CAPN15 Expands the Phenotypic Spectrum of Oculogastrointestinal Neurodevelopmental Disorder." *Genetics in Medicine* 24(3):S53–54.

Masri, Amira T., Arwa Nasir, Fatima Irshaid, Farah Alomari, Aya Irshaid, Abdelkarim Al-Qudah, Omar Nafi, and Miral Almomani. **2021**. "Genetic Evaluation of Children with Autism Spectrum Disorders in Developing and Low-Resource Areas." *Autism: the international journal of research and practice* 1491–98.

Masri, Amira T., Liyana Oweis, Abdelkarim Al Qudah, and Hatem El-Shanti. **2022**. "Congenital Muscle Dystrophies: Role of Singleton Whole Exome Sequencing in Countries with Limited Resources." *Clinical Neurology and Neurosurgery* 217.

Masri, Amira, Mira Al Jaber, Raghda Shihadat, Abdallah Rayyan, Mohammad AlMasri, Lina Abuna'meh, Majd Ali, Nosaiba Al ryalat, Azmy Hadidy, and Ashjan ahmad Al-Shakkah. **2022**. "Pseudotumor Cerebri Syndrome in Children: Clinical Characteristic and Re-Classification." *Brain & Development* 44(7):446–53.

Ababneh, Nidaa A., Raghda Barham, Ban Al-Kurdi, Dema Ali, Sabal Al Hadidi, Mohammad A. Ismail, Ahmed S. H. Muamar, Ahmed A. Abdulelah, Adan Madadha, Malik Sallam, Yazan Hassona, **Amira Masri**, and Abdalla Awidi. **2022**. "Generation of a Human Induced Pluripotent Stem Cell (iPSC) Line (JUJTCi019-A) from a Patient with Charcot-Marie-Tooth Disease Type 2A2 (CMT2A2) Due to a Heterozygous Missense Substitution c.2119C > T (p.Arg707Trp) in MFN2 Gene." *Stem Cell Research* 62.

Tamimi, Ahmad, Said Dahbour, Assma Al-Btush, Abdelkarim Al-Qudah, **Amira Masri**, Subhi Al-Ghanem, Malik E. Juweid, Yazan Olaimat, Amer Al Qaisi, Qutada Al-Soub, Maha Naim, Ali Sawalmeh, Rund Jarrar, Tala Tarawneh, Mai Bader, and Iskandar Tamimi. **2022**. "Facemask Wearing Does Not Impact Neuro-Electrical Brain Activity" *Scientific Reports* 2022 12:112(1):1-8.

Masri, Amira, Arwa Dwaikat, Nour Haroun, Lubna Haikal, Malik Kharabsheh, Amira Daher, Faris Bakri, and Abdelkarim Al Qudah. **2022**. "Aseptic Meningitis and Its Viral Etiologies, Clinical Characteristics and Management Practices in Children: A Retrospective Hospital-Based Study From Jordan." *Cureus* 14(4).

Wang, Lin, Dominik Aschenbrenner, Zhiyang Zeng, Xiya Cao, **Amira Masri**, Fawaz Alkazaleh, Deanna Guerra, **2022**. "Author Correction: Mutations in PYCR1 Cause Cutis Laxa with Progeroid Features." *Nature Genetics* 2022 54:2 54(2):213–213.

Masri, Amira, Areej M. Alfryjat, Dyala M. Alfryjat, Ro'Ya A. Ab. Saleem, and Shaima' A. Elha. **2022**. "Medical Students' Knowledge about Research Ethics." *Jordan Medical Journal* 56(1):55–63.

Tamimi, Ahmad, Faleh Tamimi, Malik Juweid, Abdelkarim A. Al-Qudah, **Amira Al Masri**, Said Dahbour, Yakub Al Bahou, and Abdelatif Shareef. **2021**. "Could Vagus Nerve Stimulation Influence Bone Remodeling?" *Journal of Musculoskeletal & Neuronal Interactions* 21(2):255.

Dodin, Yasmeen I., Maysa F. Suyagh, Mohammad I. Saleh, Ziad T. Nuseir, Salah M. Aburuz, Abdelkarim A. Al-Qudah, **Amira T. Masri**, Abdallah M. Younes, and Mutasim A. Al-Ghazawi. **2021**. "Population Pharmacokinetics Modeling of Lamotrigine in Jordanian Epileptic Patients Using Dried Blood Spot Sampling." *Drug Research* 71(8):429–37.

Masri, Amira and Hanan Hamamy. **2022**. "Cost Effectiveness of Whole Exome Sequencing for Children with Developmental Delay in a Developing Country: A Study from Jordan." *Journal of Pediatric Neurology* 20(1):20–23.

Polla, Daniel L., Mohammad Ali Farazi Fard, Zahra Tabatabaei, R. Fleming, Agnieszka Pollak, , **Amira T. Masri**, Hanan Hamamy. **2021**. "Biallelic Variants in TMEM222 Cause a New Autosomal Recessive Neurodevelopmental Disorder." *Genetics in Medicine : Official Journal of the American College of Medical Genetics* 23(7):1246.

Al Momani, Miral, Basima A. Almomani, and **Amira T. Masri**. **2021**. "The Clinical Characteristics of Primary Headache and Associated Factors in Children: A Retrospective Descriptive Study." *Annals of Medicine and Surgery* 65.

Masri, Amira, Mohammad Shboul, Aisha Khasawneh, Rama Jadallah, Asma ALmustafa, Nathalie Escande-Beillard, Hanan Hamamy, Faris Bakri, and Bruno Reversade. **2020**. "Congenital Insensitivity to Pain with Anhidrosis Syndrome: A Series from Jordan." *Clinical Neurology and Neurosurgery* 189.

Nasir, Arwa, Laeth Nasir, and **Amira Masri**. 2020. "Childhood Behavioral and Mental Health Problems in Jordan." *MedRxiv* **2020**.05.26.20113910.

Albsoul-Younes et al. 2020; A. Masri et al. 2020; **A. T. Masri** et al. 2020; Nasir, Nasir, and Masri **2020**; Trilokekar and El Masri 2020)Albsoul-Younes, Abla M., Amira T. Masri, Lobna F. Gharaibeh, Amer A. Murtaja, and Abdelkarim A. Al-Qudah. 2020. "Frequency of Antiepileptic Drugs and Response Change in Pediatric Patients Receiving 2 or More Antiepileptic Drugs." *Neurosciences* 25(4):269.

Masri, Amira T., Faisal Khatib, Abdelkarim Al Qudah, Omar Nafi, Miral Almomani, Mahmoud Bashtawi, Farah Alomari, Shahed Qutifan, and Ahed Qutifan. **2020**. "Parental Use of Conventional and Complementary Therapy for Autism in Jordan." *Complementary Therapies in Medicine* 48.

Alhafez, Laila and **Amira Masri**. 2019. "School Bullying: An Increasingly Recognized Etiology for Psychogenic Non-Epileptic Seizures: Report of Two Cases." *International Journal of Pediatrics & Adolescent Medicine* 6(4):155.

Azab, Belal, Zain Dardas, Loai Alzghoul, **Amira Masri**, Diya Hasan, Tareq Saleh, and Mohammad Alsalem. 2019. "Genotoxicity Assessment in Autism Spectrum Disorder Patients Using Sister Chromatid Exchange and Chromosomal Aberration Assays." *International Journal of Clinical and Experimental Medicine* 12(9):11476-11482.

Alsous, Mervat, Imad Hamdan, Mohammad Saleh, James McElnay, Robert Horne, and **Amira Masri**. 2018. "Predictors of Nonadherence in Children and Adolescents with Epilepsy: A Multimethod Assessment Approach." *Epilepsy & Behavior* 85:205–11.

Masri, Amira, Abeer Alassaf, Najwa Khuri-Bulos, Imad Zaq, Azmy Hadidy, and Faris G. Bakri. 2018. "Recurrent Meningitis in Children: Etiologies, Outcome, and Lessons to Learn." *Child's Nervous System : ChNS : Official Journal of the International Society for Pediatric Neurosurgery* 34(8):1541–47.

Khundakji, Yasmin, **Amira Masri**, and Najwa Khuri-Bulos. 2018. "Anti-NMDA Receptor Encephalitis in a Toddler: A Diagnostic Challenge." *International Journal of Pediatrics & Adolescent Medicine* 5(2):75.

Masri, Amira, Samah Aburahma, Aisha Khasawneh, Abdelkarim AL Qudah, Omar Nafi, Miral Al Momani, and Faisal Khatib. 2017. "Parental Knowledge and Attitudes towards Epilepsy -A Study from Jordan." *Seizure* 53:75–80.

Al-Qudah, Abdelkarim A., Abla Albsoul-Younes, **Amira T. Masri**, Samah K. AbuRahmah, Ibrahim A. Alabadi, Omar A. Nafi, Lubna F. Gharaibeh, Amer A. Murtaja, Lina H. Al-Sakran,

Haya A. Arabiat, and Abdallah A. Al-Shorman. 2017. "Type and Etiology of Pediatric Epilepsy in Jordan. A Multi-Center Study." *Neurosciences (Riyadh, Saudi Arabia)* 22(4):267–73.

Braun, Daniela A., Jia Rao, Geraldine Mollet, David Schapiro, Marie Claire Daugeron, Weizhen Tan, Olivier Gribouval, Olivia Boyer, Patrick Revy, , **Amira Masri**. 2017. "Mutations in KEOPS-Complex Genes Cause Nephrotic Syndrome with Primary Microcephaly." *Nature Genetics* 49(10):1529–38.

Masri, Amira, Seo Kyung Chung, and Mark I. Rees. 2017. "Hyperekplexia: Report on Phenotype and Genotype of 16 Jordanian Patients." *Brain & Development* 39(4):306–11.

Hamdan, Imad I., Mervat Alsous, and **Amira Taher Masri**. 2017. "Chromatographic Characterization and Method Development for Determination of Levetiracetam in Saliva: Application to Correlation with Plasma Levels." *Journal of Analytical Methods in Chemistry* 2017.

Assoum, Mirna, Christophe Philippe, Bertrand Isidor, Laurence Perrin, **Amira Masri**. 2016. "Autosomal-Recessive Mutations in AP3B2, Adaptor-Related Protein Complex 3 Beta 2 Subunit, Cause an Early-Onset Epileptic Encephalopathy with Optic Atrophy." *American Journal of Human Genetics* 99(6):1368–76.

Bakri, Faris G., Ayman Wahbeh, Awni Abu Sneina, Ali Al Khader, Fatima Obeidat, Izzat AlAwwa, Maryam Buni, Chang-Seok Ki, and Amira Masri. 2016. "Congenital Insensitivity to Pain and Anhydrosis Due to a Rare Mutation and That Is Complicated by Inflammatory Bowel Disease and Amyloidosis: A Case Report." *Clinical Case Reports* 4(10):997.

Scott, Eric M., Anason Halees, Yuval Itan, Emily G. Spencer, Yupeng He, **Amira Masri**. 2016. "Characterization of Greater Middle Eastern Genetic Variation for Enhanced Disease Gene Discovery." *Nature Genetics* 48(9):1071–79.

Albsoul-Younes, Abla, Lubna Gharaibeh, Amer A. Murtaja, **Amira Masri**, Ibrahim Alabbadi, and Abdelkarim A. Al-Qudah. 2016. "Patterns of Antiepileptic Drugs Use in Epileptic Pediatric Patients in Jordan." *Neurosciences (Riyadh, Saudi Arabia)* 21(3):264–67.

Masri, Amira and Iyad Al-Ammouri. 2016. "Clinical Presentation, Etiology, and Outcome of Stroke in Children: A Hospital-Based Study." *Brain & Development* 38(2):204–8.

Zaki, Maha S., **Amira Masri**, Anne Gregor, Joseph G. Gleeson, and Rasim Ozgur Rosti. 2015. "Dandy-Walker Malformation, Genitourinary Abnormalities, and Intellectual Disability in Two Families." *American Journal of Medical Genetics. Part A* 167A(11):2503–7.

Masri, Amira, Amani Jaafar, Rasha Noman, Almutez Gharaibeh, and Osama H. Ababneh. 2015. "Intracranial Hypertension in Children: Etiologies, Clinical Features, and Outcome." *Journal of Child Neurology* 30(12):1562–68.

Fischer-Zirnsak, Björn, Nathalie Escande-Beillard, Jaya Ganesh, Yu Xuan Tan, Mohammed Al Bughaili, , **Amira Masri**, and Uwe Kornak. 2015. "Recurrent De Novo Mutations Affecting Residue Arg138 of Pyrroline-5-Carboxylate Synthase Cause a Progeroid Form of Autosomal-Dominant Cutis Laxa." *American Journal of Human Genetics* 97(3):483–92.

Masri, Amira, Asmaa Semrin, Tariq Abdelghani, and Iyad Al-Ammouri. 2015. "Bilateral Pleural Effusion as a Complication of Central Venous Catheterization." *The Journal of Vascular Access* 16(4):e80–81.

Fairfield, Heather, Anuj Srivastava, Guruprasad Ananda, Rangjiao Liu, Martin Kircher, Anuradha Lakshminarayana, Belinda S. Harris, Son Yong Karst, Louise A. Dionne, Stephen

Murray, Robert Burgess, David E. Bergstrom, Leah Rae Donahue, Hanan Hamamy, **Amira Masri**, and Laura G. Reinholdt. 2015. "Exome Sequencing Reveals Pathogenic Mutations in 91 Strains of Mice with Mendelian Disorders." *Genome Research* 25(7):948

Kelmemi, W., M. E. Teeuw, Z. Bochdanovits, S. Ouburg, M. A. Jonker, F. Alkuraya, M. Hashem, H. Kayserili, A. van Haeringen, E. Sheridan, **A. Masri**, J. M. Cobben, P. Rizzu, P. J. Kostense, C. J. Dommering, L. Henneman, H. Bouhamed-Chaabouni, P. Heutink, L. P. ten Kate, and M. C. Cornel. 2015. "Determining the Genome-Wide Kinship Coefficient Seems Unhelpful in Distinguishing Consanguineous Couples with a High versus Low Risk for Adverse Reproductive Outcome." *BMC Medical Genetics* 16(1).

Akizu, Naiara, Vincent Cantagrel, Maha S. Zaki, **Amira Masri**, Laila Bastaki, Samia Temtamy, Ulrich Müller, Isabelle Desguerre, Jean Laurent Casanova, Ali Dursun, Murat Gunel, Stacey B. Gabriel, Pascale De Lonlay, and Joseph G. Gleeson. 2015. "Biallelic Mutations in SNX14 Cause a Syndromic Form of Cerebellar Atrophy and Lysosome-Autophagosome Dysfunction." *Nature Genetics* 47(5):528–34

Aburahma, Samah K., Firas Q. Alzoubi, Hanan M. Hammouri, and **Amira Masri**. 2015. "Vagus Nerve Stimulation Therapy in a Developing Country: A Long Term Follow up Study and Cost Utility Analysis." *Seizure* 25:167–72.

Zimoń, Magdalena, Esra Battaloğlu, Yesim Parman, Sevim Erdem, **Amira Masri**, Stephan Züchner, Vincent Timmerman, Haluk Topaloğlu, Peter De Jonghe, and Albena Jordanova. 2015. "Unraveling the Genetic Landscape of Autosomal Recessive Charcot-Marie-Tooth Neuropathies Using a Homozygosity Mapping Approach." *Neurogenetics* 16(1):33–42.

Alassaf, Abeer, **Amira Masri**, Etienne Mornet, Rasha Odeh, and Abdelkarim A. Al-Qudah. 2015. "Pseudotumor Cerebri as a Rare Presentation of Infantile Hypophosphatasia: A Case Report." *AACE Clinical Case Reports* 1(4):e221–24.

Hu, Wen F., Oz Pomp, Tawfeg Ben-Omran, Andrew Kodani, Katrin Henke, Ganeshwaran H. Mochida, Timothy W. Yu, Mollie B. Woodworth, Carine Bonnard, Grace Selva Raj, Thong Teck Tan, Hanan Hamamy, **Amira Masri**, Mohammad Shboul, Muna Al Saffar, Jennifer N. Partlow, Mohammed Al-Dosari, Anas Alazami, Mohammed Alowain, Fowzan S. Alkuraya, Jeremy F. Reiter, Matthew P. Harris, Bruno Reversade, and Christopher A. Walsh. 2014. "Katanin P80 Regulates Human Cortical Development by Limiting Centriole and Cilia Number." *Neuron* 84(6):1240.

Makrythanasis, Periklis, Mari Nelis, Federico A. Santoni, Michel Guipponi, Anne Vannier, Frédérique Béna, Stefania Gimelli, Elisavet Stathaki, Samia Temtamy, André Mégarbané, **Amira Masri**, Mona S. Aglan, Maha S. Zaki, Armand Bottani, Siv Fokstuen, Lorraine Gwanmesia, Konstantinos Aliferis, Mariana Bustamante Eduardo, Georgios Stamoulis, Stavroula Psoni, Sofia Kitsiou-Tzeli, Helen Fryssira, Emmanouil Kanavakis, Nasir Al-Allawi, Abdelaziz Sefiani, Sana' Al Hait, Siham C. Elalaoui, Nadine Jalkh, Lihadh Al-Gazali, Fatma Al-Jasmi, Habiba Chaabouni Bouhamed, Ebtessam Abdalla, David N. Cooper, Hanan Hamamy, and Stylianos E. Antonarakis. 2014. "Diagnostic Exome Sequencing to Elucidate the Genetic Basis of Likely Recessive Disorders in Consanguineous Families." *Human Mutation* 35(10):1203–10.

Masri, Amira, Jun Liao, Ruth Kornreich, and Alireza Haghighi. 2014. "Homozygous p.R284* Mutation in HEXB Gene Causing Sandhoff Disease with Nystagmus." *European Journal of Paediatric Neurology : EJPN : Official Journal of the European Paediatric Neurology Society* 18(3):399–403.

Masri, Amira, Stefania Gimelli, Hanan Hamamy, and Frédérique Sloan-Béna. 2014. "Microarray Delineation of Familial Chromosomal Imbalance with Deletion 5q35 and Duplication

10q25 in a Child Showing Multiple Anomalies and Dysmorphism." *American Journal of Medical Genetics. Part A* 164A(5):1254–61.

Novarino, Gaia, Ali G. Fenstermaker, Maha S. Zaki, Matan Hofree, Jennifer L. Silhavy, Andrew D. Heiberg, Mostafa Abdellateef, Basak Rosti, Eric Scott, Lobna Mansour, **Amira Masri**. 2014. "Exome Sequencing Links Corticospinal Motor Neuron Disease to Common Neurodegenerative Disorders." *Science (New York, N.Y.)* 343(6170):506–11.

Masri, Amira and Shourouk Al Wahsh. 2014. "Manifestations and Treatment of Epilepsy in Children with Neurometabolic Disorders: A Series from Jordan." *Seizure* 23(1):10–15.

Bode, Anna, Sian Elin Wood, Jonathan G. L. Mullins, Angelo Keramidias, Thomas D. Cushion, Rhys H. Thomas, William O. Pickrell, Cheney J. G. Drew, **Amira Masri**, Elizabeth A. Jones, Grace Vassallo, Alfred P. Born, Fusun Alehan, Sharon Aharoni, Gerald Bannasch, Marius Bartsch, Bulent Kara, Amanda Krause, Elie G. Karam, Stephanie Matta, Vivek Jain, Hanna Mandel, Michael Freilinger, Gail E. Graham, Emma Hobson, Sue Chatfield, Catherine Vincent-Delorme, Jubran E. Rahme, Zaid Afawi, Samuel F. Berkovic, Owain W. Howell, Jean François Vanbellinghen, Mark I. Rees, Seo Kyung Chung, and Joseph W. Lynch. 2013. "New Hyperekplexia Mutations Provide Insight into Glycine Receptor Assembly, Trafficking, and Activation Mechanisms." *The Journal of Biological Chemistry* 288(47):33745.

Hills, L. Benjamin, **Amira Masri**, Kotaro Konno, Wataru Kakegawa, Anh Thu N. Lam, Elizabeth Lim-Melia, Nandini Chandy, R. Sean Hill, Jennifer N. Partlow, Muna Al-Saffar, Ramzi Nasir, Joan M. Stoler, A. James Barkovich, Masahiko Watanabe, Michisuke Yuzaki, and Ganeshwaran H. Mochida. 2013. "Deletions in GRID2 Lead to a Recessive Syndrome of Cerebellar Ataxia and Tonic Upgaze in Humans." *Neurology* 81(16):1378–86.

Masri, Amira T., Najati Al Suluh, and Ramzi Nasir. 2013. "Diagnostic Delay of Autism in Jordan: Review of 84 Cases." *The Libyan Journal of Medicine* 8(1).

Eman F Badran, **Amira Masri**, Fawaz Al Kazaleh. 2012. "Fetal Intrauterine Transfusion Therapy: Neonatal Outcomes." *Journal of Blood & Lymph* 03(01).

Masri, Amira and Hanan Hamamy. 2013. "Clinical and Inheritance Profile of Familial Childhood Epilepsy in Jordan." *Seizure* 22(6):443–51.

Chung, Seo Kyung, Anna Bode, Thomas D. Cushion, Rhys H. Thomas, Charlotte Hunt, **Amira Masri**, Lokesh Lingappa, Owain W. Howell, Jean François Vanbellinghen, Jonathan G. Mullins, Joseph W. Lynch, and Mark I. Rees. 2013. "GLRB Is the Third Major Gene of Effect in Hyperekplexia." *Human Molecular Genetics* 22(5):927–40.

Carta, Eloisa, Seo Kyung Chung, Victoria M. James, **Amira Masri**, and Robert J. Harvey. 2012. "Mutations in the GlyT2 Gene (SLC6A5) Are a Second Major Cause of Startle Disease." *The Journal of Biological Chemistry* 287(34):28975.

Haghighi, Alireza, **Amira Masri**, Ruth Kornreich, and Robert J. Desnick. 2011. "Tay-Sachs Disease in an Arab Family Due to c.78G>A HEXA Nonsense Mutation Encoding a p.W26X Early Truncation Enzyme Peptide." *Molecular Genetics and Metabolism* 104(4):700–702.

Akl, Kamal F., **Amira T. Masri**, and Maali M. Hjazeen. 2011. "Acute Urine Retention Induced by Ceftriaxone." *Saudi Journal of Kidney Diseases and Transplantation: An Official Publication of the Saudi Center for Organ Transplantation, Saudi Arabia* 22(6):1226–28.

Masri, Amira, Hanan Hamamy, and Amal Khreisat. 2011. "Profile of Developmental Delay in Children under Five Years of Age in a Highly Consanguineous Community: A Hospital-Based Study--Jordan." *Brain & Development* 33(10):810–15.

Masri, Amira T., Amal M. Abu-Libdeh, Osama H. Ababneh, and Azmy M. Al-Hadidy. 2011. "Septo-Optic Dysplasia Syndrome with Schizencephaly and Sudden Visual Loss. A New Observation." *Neurosciences Journal* 16(3).

Masri, Amira, Faris G. Bakri, Ralf Birkenhäger, Abeer Alassaf, Awni F. Musharbash, Azmy Haroun, and Imad Zak. 2011. "Mondini Malformation Associated with Diastematomyelia and Presenting with Recurrent Meningitis." *Journal of Child Neurology* 26(5):622–24.

Amira Masri and Mayada Abu Shanab. 2011. "Misdiagnosis of Paroxysmal Non-Epileptic Disorders in Children." *Journal of Pediatric Neurology* 9(2):203–8.

Tian, Jing, Ling Ling, Mohammad Shboul, Hane Lee, Brian O'Connor, Barry Merriman, Stanley F. Nelson, Simon Cool, Osama H. Ababneh, Azmy Al-Hadidy, **Amira Masri**, Hanan Hamamy, and Bruno Reversade. 2010. "Loss of CHSY1, a Secreted FRINGE Enzyme, Causes Syndromic Brachydactyly in Humans via Increased NOTCH Signaling." *American Journal of Human Genetics* 87(6):768–78.

Kamal F., **Amira T. Masri**, Khaled Al-Baiti, Iyad Ammouri, and Asem Shehabi. 2010. "Dilated Cardiomyopathy with Entamoeba Histolytica Associated Hemolytic Uremic Syndrome in a Child: A Case Report with Review of the Literature(Case Reports)." *Jordan Medical Journal* 44(2):223–26.

Chung, Seo Kyung, Jean François Vanbellinghen, **Amira Masri**, and Mark I. Rees. 2010. "Pathophysiological Mechanisms of Dominant and Recessive GLRA1 Mutations in Hyperekplexia." *The Journal of Neuroscience : The Official Journal of the Society for Neuroscience* 30(28):9612–20.

Reversade, Bruno, Nathalie Escande-Beillard, Aikaterini Dimopoulou, **Amira Masri**, and Uwe Kornak. 2009. "Mutations in PYCR1 Cause Cutis Laxa with Progeroid Features." *Nature Genetics* 2009 41:9 41(9):1016–21.

Masri, Amira T., Eman F. Badran, Mohammad M. Saleem, and Abdelkarim A. Al-Qudah. 2009. "Lead Levels in Children with Developmental Delay. A Hospital-Based Study." *Neurosciences Journal* 14(3).

Masri, Amira, Faris G. Bakri, Abeer Assaf, Awni Musharbash, Azmy A. Haroun, and Imad Zak. 2009. "Intra-Axial Dermoid Tumor Mimicking Pilocytic Astrocytoma." *Child's Nervous System* 25(4):395–96.

Masri, Amira, Faris G. Bakri, Maissa Al-Hussaini, Azmy Al-Hadidy, Rania Hirzallah, Geneviève de Saint Basile, and Hanan Hamamy. 2008. "Griscelli Syndrome Type 2: A Rare and Lethal Disorder." *Journal of Child Neurology* 23(8):964–67.

Masri, Amira, Eman Badran, Hanan Hamamy, Abeer Assaf, and Abdelkarim A. Al-Qudah. 2008. "Etiologies, Outcomes, and Risk Factors for Epilepsy in Infants: A Case-Control Study." *Clinical Neurology and Neurosurgery* 110(4):352–56.

Masri, Amira T., Farouq M. Shakhathreh, Nada A. Yasein, Farihan F. Barghouti, and Abdelkarim A. Al-Qudah. 2008. "Familiarity, Knowledge, and Attitudes towards Epilepsy among Attendees of a Family Clinic in Amman, Jordan." *Neurosciences* 13(1):53–56.

Mohamed, Basly, Ben Mleh Hajer, Ben Jemaa Samia, **Masri Amira**, Chibani Mounir, Ben Ali Issam, Massoudi Lotfi, and Rachdi Radhouane. 2008. "Ovarian Hyperstimulation Syndrome: Report of Six Cases." *La Tunisie Medicale* 94–96.

Masri, Amira Taher and Hanan A. Hamamy. 2007. "Clinical and Inheritance Profiles of

Hyperekplexia in Jordan." *Journal of Child Neurology* 22(7):895–900.

Badran, Eman F., **Amira T. Masri**, Hanan Hamamy, and Abdulkarim A. Al-Qudah. 2007. "Etiological and Clinical Profile of Neonatal Seizures in a Highly Consanguineous Population." *Journal of Pediatric Neurology* 5(4):305–9.

Hamamy, Hanan A., **Amira T. Masri**, Azmy M. Al-Hadidy, and Kami M. Ajlouni. 2007. "Consanguinity and Genetic Disorders. Profile from Jordan." *Saudi Medical Journal* 28(7):1015–17.

E Badran, **A. Masri**, H. Hamamy , AA Al Qudah. 2007. "Neonatal Seizures in a Highly Consanguineous Population-Jordan University Hospital experience, *Journal of Pediatric Neurology* 5:305–9.

Masri, Amira T. , Faris G. Bakri, Azmy M. Al-Hadidy, Awni F. Musharbash, and Maisa Al-Hussaini. 2006. "Ataxia-Telangiectasia Complicated by Craniopharyngioma--a New Observation." *Pediatric Neurology* 35(4):287–88.

Masri, Amira T. 2006. "Neural Tube Defects in Jordan: A Hospital Based Study." *Journal of Pediatric Neurology* 4(4):245–49.

Masri, Amira and Hanan Hamamy. 2006. "Canavan Disease: A First Case from Jordan." *Journal of Pediatric Neurology* 4(2):143–46.

Masri, Amira. 2006. "Eye Myoclonus: A Rare Manifestation of Vitamin B12 Deficiency." *Journal of Pediatric Neurology* 4(1):49–51.

Masri, Amira. 2005. "Role of Maternal Factors in the Etiology of Neural Tube Defects in Jordan." *Saudi Medical Journal* 26(12).

Hamamy, H., **A. Masri**, and K. Ajlouni. 2005. "Wrinkly Skin Syndrome." *Clinical and Experimental Dermatology* 30(5):590–92.