

# CURRICULUM VITAE

## Mustafa Abdalla Mohamed Salih

### 1. PERSONAL:

- 1.1 *Date of birth* : 5<sup>th</sup> January 1950
- 1.2 *Place of birth* : Kosti, Sudan
- 1.3 *Nationality* : Sudanese
- 1.4 *Marital Status* : Married with 3 children
- 1.5 *E-mail* : [a] mustafa@ksu.edu.sa ; [b] mustafa\_salih05@yahoo.com
- 1.6 *Internet Biolinks* :
  - <http://fac.ksu.edu.sa/mustafa/cv>
  - <http://faculty.ksu.edu.sa/66414/default.aspx>
  - [https://www.researchgate.net/profile/Mustafa\\_Salih](https://www.researchgate.net/profile/Mustafa_Salih)
  - <http://newcastle-muscle.org/contact/mustafa-salih/>
  - <http://alumni.uofk.edu/index.php/article-categories/116-mustafa-salih>

### 2. EDUCATION AND CERTIFICATION:

- 2.1 *1956 - 1964* : Elementary and intermediate schools (Kosti). Entered the Secondary School with the second best result in the Sudan (The President of the Sudan Prize).
- 2.2 *1964 - 1968* : Kosti Secondary School. Entered the University with Grade I, distinctions in seven subjects, and second top credit in the eighth.
- 2.3 *July 1968* : Joined University of Khartoum (U of K).
- 2.4 *November 1974*: Graduated from the Faculty of Medicine with distinctions in Microbiology and Obstetrics and Gynecology; and distinctions and prizes in Social and Preventive Medicine and in Paediatrics.
- 2.5 *April 1980* : Master of Paediatrics and Child Health (U of K) with Distinction.
- 2.6 *December 1982* : Doctor of Medicine with Distinction (U of K).

- 2.7 *October 1985* : Educational Commission for Foreign Medical Graduates (ECFMG) Certificate.
- 2.8 *May 1990*: Doctor of Medical Sciences (Dr. Med Sci, Uppsala University, Sweden).
- 2.9 *August 2005*: Elected Fellow to the Royal College of Paediatrics and Child Health (FRCPCH, UK).
- 2.10 *March 2015*: Elected Fellow to the American Academy of Neurology (FAAN, USA).

### **3. CLINICAL APPOINTMENTS:**

- 3.1 *1974 - 1975* : House Officer, Khartoum Teaching Hospital (KTH) and Soba University Hospital (SUH) in the Departments of Surgery, Obstetrics and Gynecology, Medicine and Paediatrics.
- 3.2 *1975 - 1976* : Medical Officer in Paediatrics, El Buluk Teaching Hospital, Omdurman.
- 3.3 *1977 - 1980* : Paediatric Registrar and Teaching Assistant, KTH, SUH and Children's Emergency Hospital (CEH).
- 3.4 *1980 - Dec 1991*: Consultant Paediatrician at KTH, SUH and CEH.
- 3.5 *Jan 1992 - present* : Joined the Department of Pediatrics, College of Medicine, King Saud University (KSU), Consultant Paediatric Neurologist, King Khalid University Hospital (KKUH), Riyadh, Saudi Arabia.

### **4. ACADEMIC APPOINTMENTS:**

- 4.1 *April 1980*: Appointed Lecturer in the Department of Paediatrics and Child Health, Faculty of Medicine, U of K.
- 4.2 *July 1985*: Promoted to Associate Professor (U of K).
- 4.3 *July 1990*: Promoted to status of Professor of Paediatrics (U of K).
- 4.4 *1993 – present*: Professor of Pediatrics, College of Medicine, KSU.

### **5. UNDERGRADUATE TEACHING AND POSTGRADUATE TEACHING:**

- 5.1 *1982 - Dec 1991*: Undergraduate teaching in paediatrics and paediatric neurology, Faculty of Medicine, U of K.

- 5.2 *1992 – present:* Undergraduate teaching in paediatrics and paediatric neurology, College of Medicine, KSU, Riyadh.
- 5.3 *August 1980 :* Teacher, Intensive Clinical Paediatrics Courses sponsored by the Arab Board for Medical Specialization, SUH, Khartoum.
- 5.4 *1982 - Dec 1991:* Postgraduate Lectures within the teaching courses for Part I Students of the Master of Paediatrics and Child Health (MPCH) and the Joint Part I Courses for Postgraduate students in Clinical Sciences (Medicine, Obstetrics and Gynecology and Surgery), Faculty of Medicine, U of K.
- 5.5 *1982 - Dec 1991:* Theoretical and clinical teaching for Part II MPCH students.
- 5.6 *1988 - Dec 1991:* Lectures, Master of Clinical Pathology (MD Clinical Pathology), U of K.
- 5.7 *1987 - Dec 1991:* Coordinator, Part II MPCH Programme, U of K.
- 5.8 *1987 - Dec 1991:* Coordinator of theoretical and clinical parts of Part II MPCH Exam, U of K.
- 5.9 *1980 - Dec 1991:* Shared in the professional training of 68 postgraduate students in paediatrics who were preparing for their MPCH (U of K), each spending a period of 6 months on rotation basis.
- 5.10 *1992 - to present:* Professional training in paediatric neurology (periods of 1-2 months on rotation basis). Arab Board, Saudi Board and KSU Fellowship students in paediatrics, KKHU, College of Medicine, KSU.
- 5.11 *1992 - :* Adhoc teaching rounds and case discussion in clinics for Postgraduate Residents preparing for MRCP/MRCPC (UK) and MRCP (Ireland).
- 5.12 *Jan 1995 – present:* Professional training in paediatric neurology (periods of 3 months on rotation basis), Fellowship Programme in Neurology, College of Medicine, KSU.
- 5.13 *1993 - present:* Lectures and case discussions in paediatric neurology during annual vacations for postgraduate students preparing for MD (clinical paediatrics), U of K, MRCP/MRCPC and Sudan Board Certificate in Paediatrics.
- 5.14 *2015 - :* Professional training in paediatric neurology for King Saud University Fellowship Degree in Pediatric Neurology.

## **6. VISITING LECTURES:**

- 6.1 *February 1993, July 1995, June 1999:* King Fahad Hospital, Hofuf, Saudi Arabia.
- 6.2 *April 1994, Nov 1995:* King Fahad Specialist Hospital, Buraidah, Saudi Arabia.
- 6.3 *Nov 1994:* Madina Maternity and Children Hospital, Al Madina Al Munawara, Saudi Arabia.
- 6.4 *Nov 1995:* Course on “Specialty Review in Paediatrics” Department of Continuing Education, Arab Development Institute, Riyadh.
- 6.5 *Oct / Nov. 1996-2001:* “Intensive Review Course in Paediatrics”, Dallah Hospital and Rawnaa Training Center, Riyadh.
- 6.6 *July 2001:* “New Neuromuscular Diseases from the Old World”, Institute for Neurological Diseases / Neurosciences, University of Iowa, Iowa City, USA
- 6.7 *August 2001 :* “New Neuromuscular Diseases from the Old World”, Department of Neurology, University of Minnesota Medical School, USA.
- 6.8 *June 2002:* “Weak Hands and Feet: From Microbiology to Molecular Biology”, Department of Neurology, University of Minnesota Medical School, USA.
- 6.9 *July 2007:* “New Neurogenetic Disorders from the Old World”, Division of Genetics, Harvard Medical School, Boston, USA.
- 6.10 *April 2015:* “The Era of Pediatric Neurogenetics”, Department of Pharmacology and Physiology, The George Washington University, Washington DC, USA.

## **7. EXAMINATIONS:**

- 7.1 *1986, 1987, 1990 – Dec. 1991:* Internal Examiner, Part I MPCH, U of K.
- 7.2 *March 1985:* External Examiner, Faculty of Medicine, Gezira University, Sudan.
- 7.3 *1987 - Dec 1991:* Internal Examiner, Part II MPCH, U of K.
- 7.4 *April 1991:* Internal Examiner, Part II Clinical Paediatrics Examination, Arab Board for Medical Specialization, SUH, Khartoum.
- 7.5 *February 1992:* External Examiner, Arab Board Intensive Course in Paediatrics. The Joint Board of Postgraduate Medical Education (JBPME), Riyadh.
- 7.6 *April 1993:* Internal Examiner, Arab Board Intensive Course in Paediatrics, JBPME, Riyadh.
- 7.7 *Nov 1993-present:* Internal Examiner, Fellowship/Saudi Board Examinations in Paediatrics, KKHU, KSU, Riyadh.

- 7.8 *June 1995, June 1997, July 1998* : External Examiner, End of the Year Examination in Paediatrics, Saudi Board and Arab Board Residents, Sulaimania Children's Hospital, Ministry of Health, Riyadh.
- 7.9 *March 1996* : Examiner, Clinical Course in Paediatrics, Saudi Paediatric Association, Riyadh and Jeddah, Saudi Arabia.
- 7.10 *November 1996*: External Examiner, Training Course for Paediatric Arab Board Residents, Maternity and Children Hospital, Jeddah, Saudi Arabia.
- 7.11 *September 1997-1999*: Member, Examinations Committee, Saudi Board of Paediatrics, Saudi Council for Health Specialties.
- 7.12 *October 1997, October 1998, October 1999*: External Examiner, Training Course for Paediatric Saudi Board and Arab Board Residents, Maternity and Children Hospital, Jeddah
- 7.13 *February 1998, February 1999, November 2002*: Examiner, Final Examination for the Saudi Board in Paediatrics, Riyadh.
- 7.14 *April 1998* : External Examiner, Third Intensive Paediatric Course for Saudi Board, Arab Board and MRCP Part II candidates, Buraidah, Al-Qassim.
- 7.15 *June 1999* : External Examiner, End of the Year Examination in Paediatrics, Saudi Board and Arab Board Residents, King Fahad Hospital, Hofuf
- 7.16 *October 1999, January 2000, January 2001, January 2002* : External Examiner, Intensive Clinical Training Course for Paediatric Saudi Board and Arab Board Residents, Sulaimania Children's Hospital, Riyadh
- 7.17 *January 2001, January 2003*: External Examiner, Clinical Course in Paediatrics for Saudi Board, Arab Board and MRCPCH, Children's Hospital, Riyadh Medical Complex, Riyadh
- 7.18 *January 2001 - present*: Internal Examiner, Pediatric Preparatory Courses for KSU Fellowship, Saudi Board, MRCPCH and Arab Board.
- 7.19 *April 2003*: Examiner, Intensive Course for Part I MRCPCH, College of Medicine, KSU, Riyadh.
- 7.20 *January 2004*: External Examiner, Comprehensive Pediatric Clinical Course for Saudi Board, MRCPCH and Arab Board Candidates, King Fahad National Guard Hospital, Riyadh.
- 7.21 *August 2004*: External Examiner for two theses, MD (Clinical Paediatrics), Faculty of Medicine, U of K.

- 7.22 *December 2005, November 2006:* External Examiner, Pediatric Clinical Course for Saudi Board, MRCPCH and Arab Board Candidates, Children's Hospital, King Fahad Medical City, Riyadh.
- 7.23 *April 2006:* External Examiner, Pediatric Clinical Course for MRCPCH, Prince Salman Hospital, Riyadh.
- 7.24 *November 2007:* External Examiner, Final Examination in Paediatrics and Child Health, Sudan Medical Specialization Board, Khartoum, Sudan.
- 7.25 *October 2008:* External Examiner, Final Examination in Paediatrics and Child Health, University of Medical Sciences and Technology, Khartoum, Sudan.
- 7.26 *October 2008:* External Examiner, Final Examination, MD (Clinical Paediatrics), Faculty of Medicine, University of Khartoum, Sudan.
- 7.27 *December 2009:* External Examiner, First Intensive Course in Clinical Pediatrics (Saudi Board, MRCPCH and Arab Board Candidates), College of Medicine, King Saud University, Riyadh.
- 7.28 *February 2010 :* External Examiner, Final Clinical Pediatrics Examination, Arab Board for Medical Specialization, Alyamama Hospital, Riyadh.
- 7.29 *November/December 2010:* Examiner, Second Clinical Course in Pediatrics (Saudi Board, MRCPCH and Arab Board Candidates), College of Medicine, King Saud University, Riyadh.
- 7.30 *December 2011:* Examiner, Third Clinical Course in Pediatrics (Saudi Board, MRCPCH and Arab Board Candidates), College of Medicine, King Saud University, Riyadh.
- 7.31 *December 2012:* Examiner, Fourth Clinical Course in Pediatrics (Saudi Board, MRCPCH and Arab Board Candidates), College of Medicine, King Saud University, Riyadh.
- 7.32 *November 2013:* Examiner, Fifth Clinical Course in Pediatrics (Saudi Board, MRCPCH and Arab Board Candidates), College of Medicine, King Saud University, Riyadh.
- 7.33 *June 2014:* External Examiner, Pediatrics Clinical MD Graduation Examination, College of Medicine and Medical Sciences, Arabian Gulf University, Kingdom of Bahrain.
- 7.34 *November 2016:* Examiner, Eighth Clinical Course in Pediatrics (Saudi Board, MRCPCH and Arab Board Candidates), College of Medicine, King Saud University, Riyadh.

## **8. POSTGRADUATE STUDENTS SUPERVISION, TRAINING AND THESES:**

### **8.1 University of Khartoum:**

- 8.1.1 Supervisor, MPCH Thesis: “Acute Lower Respiratory Tract Infections in Sudanese Children : A Clinical and Aetiological Study”. Thesis passed in *May 1989*.
- 8.1.2 Supervisor, MPCH Thesis: “The Selenium Status of Malnourished Sudanese Children”. Thesis passed in *April 1991*.
- 8.1.3 Supervisor, MPCH Thesis: “Post -epidemic Acute Childhood Bacterial Meningitis in Sudanese Children: An Epidemiological and Clinical Study”. Thesis passed in *November 1991*.
- 8.1.4 Supervisor, MPCH Thesis: “Whooping Cough in Sudanese Children - An Epidemiological and Clinical Study”. Thesis passed in *November 1991*.
- 8.1.5 Co-supervisor, Master in Clinical Pathology Thesis: “Urinary Tract Infection in Pre-school Children”. Thesis passed in *June 1992*.
- 8.1.6 Supervisor, MD (Clinical Paediatrics) Thesis: “The Acute Phase Reactants in Sudanese Children with Severe Protein-Energy Malnutrition” Thesis passed in *June 1994*.
- 8.1.7 Co-supervisor, PhD (Physiology) Thesis: “Childhood-onset Hereditary Peripheral Neuropathy, Cerebellar Ataxia and Spastic Paraplegia in Saudi and Sudanese Populations: A Clinical, Electrophysiological and Genetic Study” Thesis passed in *October 2012*.

### **8.2 OTHER UNIVERSITIES: Collaborative studies formed part of the following theses:**

- 8.2.1 Eltom, M, 1984. **Endemic Goitre in the Sudan**. Doctoral Theses at Uppsala University, Sweden.
- 8.2.2 Fredlund, H, 1993. **Serum Factors and Polymorphonuclear Leukocytes in Human Host Defense Against *Neisseria Meningitidis*. Studies of Interactions with Special Reference to a Chemiluminometric Technique**. Doctoral theses at Uppsalla University (Scandinavian Journal of Infectious Diseases, Supplement 87, 1993).
- 8.2.3 Dajani, F, 1994. **Development of an ELISA for Detection of IgG Antibodies to Respiratory Pathogens and their Seroprevalence in Sudan and Ethiopia**. M Phil thesis at University of Liverpool, UK.

- 8.2.4 Herrmann, B, 1995. **Chlamydial Infections in the Genital and Respiratory Tracts.** Epidemiological and Diagnostic Studies. Doctoral thesis at Uppsala University, Sweden.
- 8.2.5 Backman , A, 1999. **Neisseria Meningitidis and Diagnosis of Bacterial Meningitis : Genotypic and Phenotypic Characterization, Antibiotic Susceptibility and PCR Detection.** Doctoral thesis at Linkoping University, Sweden.
- 8.2.6 Azzedine, H, 2006. **[Molecular Genetics of Autosomal Recessive Forms of Charcot Marie Tooth Disease] French.** Doctoral thesis at the University of Paris VI (Pierre and Marie Curie University), France.
- 8.2.7 Gribba, M, 2006. **[Novel Genetic and Molecular Forms of Recessive Progressive Atxias] French.** Doctoral thesis at the University Louis Pasteur-Strasbourg, France.
- 8.2.8 Anheim, M, 2009. **[Identification and Characterization of Novel Genes Responsible of Novel Forms of Recessive Ataxias] French.** Doctoral thesis at the University of Strasbourg, France.
- 8.2.9 Assoum, M, 2010. **[Identification of Novel Recessive Ataxia Genes: Implication of Mitochondria and Novel Pathophysiologies] French.** Doctoral thesis at the University of Strasbourg, France.
- 8.2.10 Mallaret, M, 2015. **[dentification of Novel Recessive Cerebellar Ataxia Genes and Usefulness of High Throughput Sequencing for the Diagnosis of Inherited Ataxias] French.** Doctoral thesis at the University of Strasbourg, France.
- 8.2.11 Elsayed, L, 2016. **Hereditary spastic paraplegias: clinical spectrum in Sudan, further deciphering of the molecular bases of autosomal recessive forms and new genes emerging.** Doctoral thesis at Sorbonne University of Paris VI [Paris 6], France.

## **9. ON MEDICAL AND HEALTH EDUCATION:**

- 9.1 1976 - 1980 : Teaching courses in Child Health and Medicine, School of Nurses, El Buluk Teaching Hospital, SUH and KTH.
- 9.2 1976 - 1980 : Interviews on Nutrition Programme (a National Radio Programme on Health Education organized by the Department of Nutrition, Ministry of Education).
- 9.3 February 1983: Organizer, Intensive Paediatric Neurology Course for postgraduate students in paediatrics and medicine. Course conducted by Dr. David Gradner-Medwin, Regional Neurological Centre, Newcastle Upon Tyne, UK.



- 9.4 *July 1987:* Initiated and organized a postgraduate course in clinical paediatrics, conducted by Dr. Fraser W. Alexander , Newcastle General Hospital, Newcastle Upon Tyne, UK.
- 9.5 *1988 – 1989 :* Lectures, Immunization Course for Health Visitors, organized by Expanded Programme on Immunization (EPI) Central Office, Khartoum.
- 9.6 *1989:* Three interviews, Sudan National Radio programme on immunization, organized by EPI Central Office, Khartoum.
- 9.7 *April 1991:* Organizer, Part II Clinical Paediatrics Examination, Arab Board for Medical Specialization (ABMS) [First Clinical Examination Centre for ABMS to be opened in Sudan].
- 9.8 *1992 – present :* Interviews on children health issues, The Kingdom of Saudi Arabia National Radio and Television Programs.
- 9.9 *2003 - present:* Interviews on children health and pediatric neurology. Sudan National Television and Blue Nile Television.
- 9.10 *April 2006:* Co-organizer, Scientific Exhibition, The First Meeting for The Founders and Sponsors, Prince Salman Centre for Disability Research, **Riyadh, Saudi Arabia.**
- 9.11 *November 2007:* Co-organizer, Pre-Congress Workshop on Epilepsy in Childhood; The 15<sup>th</sup> Congress of the Union of Arab Paediatricians and the 15<sup>th</sup> Conference of the Sudanese Association of Paediatricians, **Khartoum, Sudan.**
- 9.12 *October 2009:* The 16<sup>th</sup> Conference of the Sudanese Association of Paediatricians, **Khartoum, Sudan.** (Member of the Scientific Committee and Co-organizer of pre-congress workshop on Epilepsy in Childhood and Co-chairperson, Neurology Session).
- 9.13 *October 2011:* The 17<sup>th</sup> Conference of the Sudanese Association of Paediatricians, **Khartoum, Sudan.** (Member of the Scientific Committee and Co-organizer of pre-congress workshop on Epilepsy in Childhood and Co-chairperson, Neurology Session).
- 9.14 *January 2015:* The International Child Neurology Association (ICNA) Educational Meeting, **Khartoum, Sudan.** (Member of the Scientific Committee and Co-chairperson of one session:  
[\(https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4949866/](https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4949866/),  
<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4949860/>).

## ***10. ACADEMIC AND ADMINISTRATIVE RESPONSIBILITIES:***

- 10.1 *1983* : Coordinator of Learning Resources Office, Education Development Centre (EDC), Faculty of Medicine, University of Khartoum (U of K).
- 10.2 *1985* : Convenor, Students' Affairs Committee, Faculty Board Adhoc Committee on Students' Evaluation, Faculty of Medicine, U of K.
- 10.3 *1985 - 1986* : Member, Curriculum Revision Committee, Faculty of Medicine, U of K.
- 10.4 *1982 - Dec 1991* : Member, MPCH Degree Committee, Faculty of Medicine, U of K.
- 10.5 *1986 – 1991* : Co-opted Member, Central Committee for Maternity and Child Health, Ministry of Health.
- 10.6 *Jul 1990 - Dec 1991*: Convenor, MPCH Degree Committee, Faculty of Medicine, U of K.
- 10.7 *Jul 1990 - Dec 1991* : Head, Department of Paediatrics, Faculty of Medicine, U of K. Became, in this capacity, a member of the Senate, U of K; Faculty of Medicine Board, Faculty Research Board, Heads of Departments Board, Board of Postgraduate Medical Studies (BPMS) Convenors Committee, BPMS Board; and Academic Council for the High Nursing School.
- 10.8 *Sept 1990 - Dec 1991*: President, Scientific Committee, Sudan National Expanded Programme on Immunization.
- 10.9 *Nov 1990 - Dec 1991*: Member of the Central Research Committee, U of K. Member of Adhoc Subcommittee which devised regulations that govern distribution of research funds in the various colleges.
- 10.10 *1992 – 1994*: Member of Departmental Residency Training Committee (DRTC), Department of Paediatrics, College of Medicine, King Saud University (KSU), Riyadh.
- 10.11 *1992 - 1998*: Member, Department of Paediatrics Development and Planning Committee, and some Adhoc committees formulated by the Dean, College of Medicine, KSU, Riyadh.
- 10.12 *1992 – 1998 and 2005-present*: Member, College of Medicine Research Centre (CMRC) Board, KSU, Riyadh.
- 10.13 *1993 - 1998*: Member, Committee for KSF Written Examination in Paediatrics, KSU, Riyadh.

- 10.14 *1994 - 1998*: Member, Research Advisory Committee, Department of Paediatrics, College of Medicine, KSU, Riyadh.
- 10.15 *1998 – 2000 and 2004 - present*: Member, Department of Paediatrics Postgraduate and Continuous Medical Education Committee, College of Medicine, KSU, Riyadh.
- 10.16 *1998 – present*: Member, Department of Paediatrics Research and Development Committee, College of Medicine, KSU, Riyadh.
- 10.17 *Dec. 1998 - present*: Member, the Neurogenetics Group of the Pan Arab Union of Neurological Sciences (PAUNS) Research Committee.
- 10.18 *2008 -2010*: Member, Prince Salman Center for Disability Research (PSCDR) Research Board, KSU, Riyadh.

## **11. FELLOWSHIPS AND AWARDS:**

- 11.1 *1980 - 1982* : Clinical Research Fellow, Regional Neurological Centre, Newcastle General Hospital (NGH) and Fellow to the Department of Child Health, University of Newcastle Upon Tyne, UK (15 months).
- 11.2 *1984* : Fellow to the Department of Paediatrics (International Child Health and Paediatric Neurology Units) and Neurophysiology, University Hospital, Uppsala, Sweden (4 months).
- 11.3 *1986* : Fellow and Clinical Assistant, Department of Neurophysiology, Regional Neurological Centre, NGH and Fellow to the Department of Child Health, University of Newcastle Upon Tyne, UK (6 months).
- 11.4 *1985 - 1990* :Fellow to the Department of Paediatrics (International Child Health Unit), Uppsala University and the Department of Clinical Microbiology and Immunology, Orebro Medical Centre Hospital, Sweden. Within this fellowship, a joint Sudanese/Swedish project on childhood bacterial meningitis in the Sudan was conducted. Travels to Sweden were arranged within annual vacations and a sabbatical leave.
- 11.5 *1996* : The 1996 Riyadh Neurosciences Club Award (The First Annual Awards Day).
- 11.6 *2007*: The Medal of Excellence. Presented at a special ceremony by The President of Sudan at the Republican Palace, Khartoum, Sudan.
- 11.7 *2008*: The Saudi Neuroscience Society Award for Pioneers and Promoters of Neurosciences in Saudi Arabia.
- 11.8 *2010*: King Saud University Gold Medal.

11.9 *2011*: College of Medicine Active Researcher Award 2010.

11.10 *2014*: King Saud University Life-time Achievement Award.

## **12. INTERNATIONAL COLLABORATIONS:**

12.1 Howard Hughes Medical Institute, Department of Physiology and Biophysics, Internal Medicine, and Neurology, University of Iowa College of Medicine, Iowa City, IA, **USA.**

12.2 Department of Pathology, The University of Iowa, Iowa City, IA, **USA.**

12.3 Department of Neurology, Children's Hospital Boston, Boston, Harvard Medical School, Boston, Massachusetts, **USA.**

12.4 Division of Genetics, The Manton Center for Orphan Disease Research, Children's Hospital Boston, Boston, Massachusetts, **USA.**

12.5 Howard Hughes Medical Institute, Boston Children's Hospital, Boston, MA, **USA.**

12.6 Department of Genetics, Harvard Medical School, Boston, MA, **USA.**

12.7 Department of Neurology and Program in Genomics, Children's Hospital Boston, and Program in Neuroscience, Harvard Medical School, Boston, MA, **USA.**

12.8 Department of Molecular and Human Genetics, Baylor College of Medicine, One Baylor Plaza, Houston, Texas, **USA.**

12.9 Departments of Neurology and Human Genetics, UCLA School of Medicine, Los Angeles, CA, **USA.**

12.10 Department of Pharmacology and Physiology, The George Washington University School of Medicine, Washington, DC 20037, **USA.**

12.11 Provincial Medical Genetics Program, Child and Family Research Institute, Department of Medical Genetics, University of British Columbia, Vancouver, **Canada.**

12.12 Neuromuscular/Neurology Division, Hospital de Clinicas, Universidade Federal do Parana, Curitiba, **Brazil.**

12.13 Genethon, Centre, National de la Recherche Scientifique, Unite' Mixte de Recherche, Evry, **France.**

12.14 Institut de Myologie, Groupe Hospitalier Pitie'-Salpetriere, Paris, **France.**

12.15 Institut de Génétique et de Biologie Moléculaire et Cellulaire, CNRS/INSERM/Université de Strasbourg, et Collège de France, 67404 Illkirch, **France.**

12.16 Université Pierre et Marie Curie-Paris, Paris, **France.**

- 12.17 University Institute of Clinical Research, INSERM UMR\_S 827 and Laboratoire de Genetique Moleculaire, University Hospital, Montpellier, **France**.
- 12.18 The John Walton Muscular Dystrophy Research Centre, Newcastle upon Tyne, **UK**.  
(<http://newcastle-muscle.org/contact/mustafa-salih/>)
- 12.19 Wellcome Trust Centre for Human Genetics, Oxford, **UK**.
- 12.20 Division of Clinical Neurology and Department of Molecular Pathogenesis, Institute of Neurology, Queen Square, London, WC1N 3BG, **UK**.
- 12.21 Neurosciences Group, Nuffield Department of Clinical Neurosciences, Weatherall Institute of Molecular Medicine, University of Oxford, Oxford OX3 9DS, **UK**.
- 12.22 MRC Mitochondrial Biology Unit, Cambridge CB2 0XY, **UK**.
- 12.23 Institute of Genetic Medicine, Newcastle University, International Centre for Life, Newcastle NE1 3BZ, **UK**.
- 12.24 Guy's and St Thomas' NHS Foundation Trust, London, **UK**.
- 12.25 St John's Institute of Dermatology, King's College London (Guy's campus), London, **UK**.
- 12.26 Department of Human Genetics 855, Radboud University Nijmegen Medical Center, Box 9101, 6500 HB Nijmegen, **The Netherlands**.
- 12.27 Department of Child Neurology, VU University Medical Center, Amsterdam, **The Netherlands**.
- 12.28 Molecular Genetics Department, Flanders Interuniversity Institute for Biotechnology, University of Antwerp, **Belgium**.
- 12.29 Department of Human Genetics, Université de Liège, Liège, **Belgium**.
- 12.30 Department of Pediatrics and Pediatric Neurology, University Hospital of Essen, Essen, **Germany**.
- 12.31 Friedrich-Baur-Institute, Department of Neurology, Ludwig-Maximilians-University, Munich, **Germany**.
- 12.32 Department of Ropers–Molecular Human Genetics, Max-Planck Institute of Molecular Genetics, Berlin, **Germany**.
- 12.33 Department of Molecular Neurogenetics, Institute of Neurology Besta, 23888 Milan, **Italy**.

- 12.34 IRCCS Casa Sollievo della Sofferenza, Mendel Laboratory San Giovanni Rotondo, San Giovanni Rotondo, **Italy**.
- 12.35 Division of Neuromuscular Diseases and Neuroimmunology, Fondazione IRCCS Istituto Neurologico C. Besta, Milan, **Italy**.
- 12.36 International Child Health Unit, Department of Pediatrics, University Hospital, Uppsala, **Sweden**.
- 12.37 Department of Clinical Microbiology and Immunology, Orebro Medical Center Hospital, Orebro, **Sweden**.
- 12.38 Department of Methodology, National Institute of Public Health, Oslo, **Norway**.
- 12.39 Folkhalsan Institute of Genetics and Department of Medical Genetics, Haartman Institute, University of Helsinki, Helsinki, **Finland**.
- 12.40 Neuromuscular Research Center, Tampere University and University Hospital, Tampere, Helsinki, **Finland**.
- 12.41 Departments of Pediatrics and Child Health, Biochemistry, Physiology and Microbiology, College of Medicine, University of Khartoum, Khartoum, **Sudan**.
- 12.42 Molecular Biology Laboratory, Institute of Endemic Diseases, University of Khartoum, Khartoum, **Sudan**.
- 12.43 School of Mathematical Sciences, University of Khartoum, Khartoum, **Sudan**.
- 12.44 National Metabolic Service, Starship Children's Hospital, Auckland, **New Zealand**.
- 12.45 Department of Neurology and Geriatrics, Kagoshima University Graduate School of Medical and Dental Sciences, Kagoshima, **Japan**.
- 12.46 The Institute of Basic Medical Sciences, Department of Molecular Medicine, University of Oslo, Oslo, **Norway**.
- 12.47 Department of Medical Genetics, Oslo University Hospital and University of Oslo, **Norway**.
- 12.48 National Centre for Rare Epilepsy-related Disorders, Oslo University Hospital, Oslo, **Norway**.

### 13. Membership of International Consortia:

13.1 The International Charcot-Marie-Tooth (CMT) Disease Consortium.

13.2 The International Network of hereditary forms of SPastic paraplegias and cerebellar ATAXias (SPATAX).

13.3 The International Joubert Syndrome and Related disorders (JSRD) Study Group.

13.4 The Autosomal Recessive Cutis Laxa Debry-type Study Group.

### 14. Novel Scientific Contribution, Patents and Invention:

**14.1 Novel neurologic disorders and newly identified causative genes (investigated in collaboration with an international group of scientists):**

***14.1.1 Syndromes and diseases with gene / locus identification:***

14.1.1.1 **Salih myopathy:** Autosomal recessive titinopathy causing early onset myopathy/dystrophy with dilated cardiomyopathy  
(<http://www.ncbi.nlm.nih.gov/books/NBK83297/>).

14.1.1.2 Charcot-Marie-Tooth Disease Type 4B1 (OMIM 601382).

14.1.1.3 A new form of childhood-onset, autosomal recessive spinocerebellar ataxia and epilepsy due to tumour suppressor gene *WWOX* mutation  
(<http://brain.oxfordjournals.org/cgi/content/full/130/7/1921>,  
<http://www.ncbi.nlm.nih.gov/pubmed/24369382>).

14.1.1.4 Spinocerebellar ataxia with axonal neuropathy (SCAN1; OMIM 607250;  
<http://www.ncbi.nlm.nih.gov/books/NBK1105/>)

14.1.1.5 Horizontal gaze palsy and progressive scoliosis (OMIM 607313).

14.1.1.6 Walker-Warburge syndrome due to *LARGE* gene mutation (OMIM 613154).

**14.1.1.7 Bosley-Salih-Alorainy syndrome (OMIM 601536)**

14.1.1.8 **Salih ataxia**  
(<http://brain.oxfordjournals.org/content/133/8/2439.full.pdf+html>)  
(<http://www.ncbi.nlm.nih.gov/pubmed/23728897>).

- 14.1.1.9 Emery-Dreifuss muscular dystrophy 6 (EDMD6, OMIM 300696) due to *FHL1* gene mutation.
- 14.1.1.10 Mitochondrial disease due to two novel genes: *MFF* (OMIM 614785) and *FARS2* (OMIM 611592).
- 14.1.1.11 Hereditary spastic paraplegia due to *CYP2U1* gene mutations (SPG49) (<http://www.sciencedirect.com/science/article/pii/S0002929712005794>).
- 14.1.1.12 Mainzer-Saldino syndrome (OMIM 266920): A ciliopathy caused by IFT140 gene mutations.
- 14.1.1.13 A novel form of congenital myasthenic syndrome due to *AL2G2* gene mutations (<http://brain.oxfordjournals.org/content/136/3/944.long> )
- 14.1.1.14 A novel form of congenital muscular dystrophy due to *B3GALNT2* gene mutations (<http://www.cell.com/AJHG/retrieve/pii/S0002929713000694>)
- 14.1.1.15 A novel autosomal recessive form of intellectual disability with strabismus due to single ancient founder missense mutation in *ADAT3* gene affecting eight seemingly unrelated Arab families from divergent regions. (<http://jmg.bmj.com/content/50/7/425.long>).
- 14.1.1.16 Encephalomyopathic mitochondrial DNA depletion syndrome-13 (MTDPS13, OMIM 615471) due to *FBXL4* gene mutation
- 14.1.1.17 A novel gene for migrating partial seizures in infancy (<http://onlinelibrary.wiley.com/doi/10.1002/ana.23998/pdf>).
- 14.1.1.18 Limb-girdle muscular-dystroglycanopathy type C12 (MDDGC12, OMIM 616094) due to *POMK* gene mutation.
- 14.1.1.19 Confirming the clinical utility of molecular karyotyping by investigating 584 patients with neurogenerative phenotypes and finding that at least 21% tested cases had chromosomal abbreviations (<http://www.ncbi.nlm.nih.gov/pubmed/25503496>).
- 14.1.1.20 Using whole-exome sequencing in pre-screened multiplex consanguineous families, 33 novel candidate genes were described for various neurogenetic conditions ([http://www.cell.com/cell-reports/pdf/S2211-1247\(14\)01044-4.pdf](http://www.cell.com/cell-reports/pdf/S2211-1247(14)01044-4.pdf)).



14.1.1.21 Accelerating matchmaking of novel dysmorphology syndromes through clinical and genomic characterization of a large cohort of 31 families. This study identified 10 novel genes  
([https://scholar.google.com/scholar?cluster=11441892540088267610&hl=en&as\\_sdt=0,5](https://scholar.google.com/scholar?cluster=11441892540088267610&hl=en&as_sdt=0,5)).

14.1.1.22. A novel locus for intellectual disability with null mutation in *TNIK* gene (Mental retardation, autosomal recessive 54; OMIM 617028).

14.1.1 23. A novel epileptic-dyskinetic encephalopathy syndrome due to mutations in *FRRS1L* gene (Epileptic encephalopathy, early infantile, 37; OMIM 616981).

14.1.1.24. Through clinical genomics, the morbid genome of intellectual disability was expanded by investigating a large cohort of 337 patients. This study identified 3 novel candidate genes and likely causal variants in another 32 potential candidate genes  
(<http://www.nature.com/mp/journal/vaop/ncurrent/full/mp2016113a.html>).

#### **14.1.2 Syndromes and diseases with gene/locus still unidentified:**

14.1.2.1 Autosomal recessive epidemolysis bullosa simplex (OMIM 226670).

14.1.2.2 Muscular dystrophy, congenital, with severe central nervous system atrophy and absence of large myelinated fibers (OMIM 601170).

14.1.2.3 **Dincsoy Salih Patel syndrome:** midline malformations, multiple, with limb abnormalities and hypopituitarism ([http://www.orpha.net/consor/cgi-bin/Disease\\_Search\\_Simple.php?lng=EN](http://www.orpha.net/consor/cgi-bin/Disease_Search_Simple.php?lng=EN); OMIM 601016)

## **14.2 Patents and Invention:**

### **14.2.1 United States Patent No.: 6, 136, 546**

(<http://www.freepatentsonline.com/6136546.html>)

**Inventors:** Kevin P. Campbell, Iowa City Iowa; Valerie Allamand, Iowa City Iowa; Youshihide Sunada, Kawaguchi, Japan; Volker Straub, Iowa City Iowa; **Mustafa Salih**, Riyadh, Saudi Arabia.

**Title:** Method for Aiding in the Diagnosis of In-Frame Deletion Type Congenital Muscular Dystrophy.

### **14.2.2 United States Patent No.: US20140335021 A1 (pending)**

(<http://www.google.com/patents/US20140335021>)

**Inventors:**

**Mustafa A.M. Salih**, Mohamed A.M. Salih, Ali A. Mustafa, Thomas M. Bosley.

**Title:** Ocular Neostigmine Drops for Diagnosing Myasthenia Gravis.

## **15. EDITORIAL ACTIVITIES, PROFESSIONAL AFFILIATIONS AND SCIENTIFIC REFERENCE:**

### **15.1 Editorial Activities:**

15.1.1 *1985 – 1991*: Editor, Sudanese Journal of Paediatrics.

15.1.2 *1983 - 1986*: Member of the Editorial Board of Sudan Medical Journal (invited Editor of an issue).

15.1.3 *2003-present*:- Member, Advisory Board, Journal of Pediatric Neurology.

15.1.4 *2006*: Guest Editor: Saudi Medical Journal, Supplement 1 (March 2006), Volume 27 on “Childhood Stroke”.

15.1.5 *2008-present*: Member of the Editorial Board of The Open Journal of Pediatric Medicine.

15.1.6 *2008-present*: Member of the Editorial Board of The Open Neurology Journal.

15.1.7 *2009-present*: International Editor, Sudan Medical Journal.

15.1.8 *2010-present*: International Editor, Sudanese Journal of Paediatrics.

15.1.9 *2013-present*: Member, Advisory Board, Journal of Taibah University Medical Sciences.

15.1.10 *2014- present*: Member of the Editorial Board of International Journal of Pediatrics and Adolescent Medicine.

15.1.11 *2014*: Guest Editor: Saudi Medical Journal, Supplement (December 2014), Volume 35 on “Neural Tube Defects”.

### **15.2 Professional Affiliations:**

15.2.1 *1983 -* : Member of the Sudan Association of Paediatricians.

15.2.2 *1983 - 1991* : Member of the Council of Sudan Association of Paediatricians.

15.2.3 *1983 -* : Member of the World Federation of Neurology.

15.2.5 *1992 -* : Member of Saudi Pediatric Association.

15.2.5 *1992-* : Member of Saudi Neuroscience Society.

13.2.6 *1997-* : Member of the American Association for the Advancement of Science.

15.2.7 *1997-* : Member of the National Geographic Society (USA).

15.2.8 *1998-* : Member of the International Child Neurology Association.

15.2.13 2007-: Member of the American Academy of Neurology.

15.2.14 2007-: Member of the American Epilepsy Society.

### **15.2 Scientific Reference:**

15.2.1 Scientific referee for the promotion to the status of Professor in Neurology for:

15.2.2 King Faisal University, Dammam, Saudi Arabia.

15.2.3 King Abdulaziz University, Jeddah, Saudi Arabia.

### **15.3.2 Refereed original articles for the following scientific periodicals:**

15.3.2.1 Sudanese Journal of Paediatrics.

15.3.2.2 Sudan Medical Journal.

15.3.2.3 Acta Paediatrica Scandinavica.

15.3.2.4 Saudi Medical Journal.

15.3.2.5 Annals of Saudi Medicine.

15.3.2.6 Saudi Journal of Kidney Diseases and Transplantation.

15.3.2.7 The Saudi Journal of Sport Medicine.

15.3.2.8 Journal of Pediatric Neurology.

15.3.2.9 Journal of Pediatric Infectious Diseases.

15.3.2.10 Epilepsy & Behavior.

15.3.2.11 The Open Journal of Pediatric Medicine.

15.3.2.12 The Open Neurology Journal.

15.3.2.13 PEDIATRICS.

15.3.2.14 Plos Neglected Tropical Diseases.

### 15.3.3 Refereed and reviewed scientific research projects for the following institutes:

- 15.3.3.1 King Salman Center for Disability Research.
- 15.3.3.2 Medical Research Section, Ministry of Health, Saudi Arabia.
- 15.3.3.3 Medical Research Centre, Sciences and Medical Studies, Department for Women Students, King Saud University (KSU), Saudi Arabia.
- 15.3.3.4 Centre for Research on Education, KSU, Saudi Arabia.
- 15.3.3.5 Office of Research Affairs, King Faisal Specialist Hospital and Research Centre (KFSH&RC), Riyadh, Saudi Arabia.
- 15.3.3.6 College of Medicine Research Board (CMRC), KSU, Riyadh, Saudi Arabia.

## 16 RESEARCH GRANTS AND COMPLETED PROJECTS

16.1 **Principal Investigator**, Research Project (No. B/M/4/15): Stroke in Saudi Children: Clinical Features, Etiology, Risk Factors, and Prognosis; **King Salman Center for Disability Research**.

16.2 **Co-Investigator**, Pilot Study (No. PSCDR/244/402): Risk Factors for Neural Tube Defects in Riyadh City, Saudi Arabia; **King Salman Center for Disability Research**.

16.3 **Principal Investigator**, Research Project (No. 05-495): Risk Factors for Neural Tube Defects in Riyadh City, Saudi Arabia; College of Medicine Research Board (CMRC), **King Saud University, Riyadh, Saudi Arabia**: A study exploring the risk factors for neural tube defects in Saudi children including folic acid supplementation and the role of consanguinity.

16.4 **Co-Investigator**, Research Project (AT-29-31): Genetics of Hereditary Visual Loss; **King Abdulaziz City for Science and Technology, Riyadh, Saudi Arabia**.

16.5 **Co-Investigator**, Research Project (AT-30-20): Genetics of Ocular Motility; **King Abdulaziz City for Science and Technology, Riyadh, Saudi Arabia**.

16.6 **Principal Investigator**, Research Group Project (No. RGP-VPP-301), Deanship of Scientific Research, **King Saud University, Riyadh, Saudi Arabia**.

**17. CONFERENCES, SEMINARS & SYMPOSIA: SCIENTIFIC WORK PRESENTED, ORGANIZATION AND CHAIRMANSHIP:**

- 17.1 The 6<sup>th</sup> Conference of the Sudan Association of Physicians, **Khartoum**, February 1978 (presented one paper).
- 17.2 The XIIth Congress of the Union of the Middle Eastern and Mediterranean Paediatric Societies, **Khartoum**; December 1979 (presented one paper).
- 17.3 British Paediatric Neurology Association Meeting, **Manchester**, January 1981 (presented one paper).
- 17.4 53<sup>rd</sup> Annual Meeting of the British Paediatric Association, **York**; April 1981 (presented one paper).
- 17.5 The XIVth Congress of the Union of Middle Eastern and Mediterranean Paediatric Societies. **Kuwait**; December 1982 (presented two papers and chaired the session on Genetics).
- 17.6 Workshop on Child Health Priorities and Development in Primary Health Care (VIIth Conference of the Sudan Association of Paediatricians), **Fao Town**; December 1982 (presented the Final Report of Immunization Panel).
- 17.7 63<sup>rd</sup> Annual Meeting of British Association of Dermatologists, **London** ; July 1983 (presented one paper).
- 17.8 XVII International Congress of Paediatrics, **Manila** ; November 1983 (presented one paper and co-chaired a symposium on "New syndromes in Pediatric Dermatology").
- 17.9 The 9<sup>th</sup> Conference of the Sudan Association of Physicians, **Khartoum** ; February 1984 (presented one paper).
- 17.10 The XVth Congress of the Union of the Middle Eastern and Mediterranean Paediatric Societies, **Cairo** ; November 1984 (presented one paper).
- 17.11 The 8<sup>th</sup> Conference of the Sudan Association of Paediatricians, **Khartoum**; February 1985 (presented three papers).
- 17.12 The XIIth World Congress of Neurology, **Hamburg**; September 1985 (presented one paper).
- 17.13 58<sup>th</sup> Annual Meeting of the British Paediatric Association, **York** ; April 1986 (presented one paper).
- 17.14 The 9<sup>th</sup> Annual Conference of the Sudan Association of Paediatrician in Collaboration with the 23rd Arab Medical Congress, **Khartoum** ; February 1987 (presented three papers and edited the Scientific Programme of the Paediatric Panel).

- 17.15 The International Symposium on Work in a Hot Environment and Heat Related Disorders, **Khartoum** ; January 1988 (presented an invited review) ; Member of Scientific Committee.
- 17.16 The 11<sup>th</sup> Conference of the Sudan Association of Physicians; **Khartoum**; February 1988 (presented one paper).
- 17.17 The International Seminar on the Importance of Nutrition and Early Stimulation for the Education of Children in the Third World, **Stockholm**; April 1988 (organized by Wenner-Gren Centre Foundation and UNESCO Sweden. Presented an invited review).
- 17.18 International Conference on the Impact of Viral Diseases on Health Care and Medical Service in Saudi Arabia and the Middle East, **Riyadh**; June 1988 (presented two papers).
- 17.19 The XVIII Congress of the Union of Middle - Eastern and Mediterranean Paediatric Societies, **Damascus**: September 1988 (presented two papers).
- 17.20 The 4<sup>th</sup> Annual Paediatric Symposium, **Riyadh**; March 1989 (presented two papers).
- 17.21 The Sixth International Congress on Rapid Methods and Automation in Microbiology and Immunology, **Helsinki**; June 1990 (presented one paper).
- 17.22 The First Scientific Conference, Soba University Hospital, U of K, **Khartoum**; February 1991 (presented an invited review).
- 17.23 The 6<sup>th</sup> Annual Paediatric Symposium, **Riyadh**; February 1992 (presented an invited review).
- 17.24 International Congress for Infectious Diseases, **Nairobi**; June 1992 (contributed one paper).
- 17.25 Third Annual Paediatric Symposium on Recent Advances in Paediatrics, **Al Madina Al Munawara**; October 1992 (presented an invited review).
- 17.26 The First Congress of the Saudi Benevolent Association for Disabled Children, **Riyadh**; November 1992 (contributed two papers; Member of Paediatric Neurology Reviewers Committee; Co-chairman of a scientific session).
- 17.27 International Paediatric Conference: Modern Trends in Paediatrics, **Bisha**, Saudi Arabia; June 1993 (presented an invited review).
- 17.28 International Workshop on Epilepsy and Sleep Disorders, **Riyadh** ; October 1993 (presented an invited review).

- 17.29 The 9<sup>th</sup> Annual Neuroscience Symposium, **Riyadh** ; March 1994 (contributed three papers ; Chairperson of a scientific session).
- 17.30 1994 Joint Meeting, European Group for Rapid Viral Diagnosis and European Society Against Virus Diseases, **Stockholm**; August 1994 (contributed one paper).
- 17.31 The VIIth International Child Neurology Congress and 23rd National Meeting of the American Child Neurology Society, **San Francisco**; October 1994 (contributed one paper).
- 17.32 The 7<sup>th</sup> Annual Paediatric Symposium, **Riyadh**; December 1994 (presented one paper).
- 17.33 The International and National Symposium on Paediatrics Neurosurgery, **Al Medina Al Munawara**; October 1995 (presented one paper).
- 17.34 Symposium on Psychosomatic Medicine and Child Psychiatry, **Riyadh**; April 1996 (contributed two papers and chaired a scientific session).
- 17.35 The 8<sup>th</sup> Annual Paediatric Symposium, **Riyadh**; May 1996 (presented one paper).
- 17.36 The 5<sup>th</sup> Asian and Oceanian Congress of Child Neurology, **Istanbul**; October 1996 (presented two papers).
- 17.37 The 6<sup>th</sup> National Meeting on Neuromuscular Diseases, the Association Francaise contre les Myopathies (AFM), **Versailles**; October 1996 (invited; contributed two papers).
- 17.38 The American Society of human Genetics Meeting, **San Francisco**; October 1996 (contributed one paper).
- 17.39 The Second Congress of the World Muscle Society, **Tunis**; October 1997 (contributed one paper).
- 17.40 Symposium on Genetic Diseases in Arab Populations - A wealth of Information, **Riyadh**; November 1997 (presented two papers).
- 17.41 Workshop on Paediatric Radiology, **Riyadh**; February 1998 (Co-chairperson, Session II).
- 17.42 Symposium on Recent Advances in Neurology, **Riyadh**; April 1998 (presented an invited paper and an invited video presentation).
- 17.43 The 8<sup>th</sup> International Child Neurology Congress , **Ljubljana (Slovenia)**; September 1998 (presented one paper).
- 17.44 The 4<sup>th</sup> Workshop of the European CMT - Consortium - 62nd ENMC International Workshop : Rare Forms of Charcot - Marie - Tooth Disease and Related Disorders, **Soestduinen (The Netherlands)**; November 1998 (invited; presented one paper).

- 17.45 International Symposium on Recent Advances in Pediatrics and Neonatology, **Dhahran**; March 1999 (invited; presented one paper).
- 17.46 The 8<sup>th</sup> Annual Symposium of the European Charcot - Marie - Tooth Consortium, **Antwerp**; July 1999 (invited; presented two papers).
- 17.47 Symposium on Genetics at Bench and Bedside, King Faisal Specialist Hospital and Research Centre (KFSH & RC), **Riyadh**; October 1999 (invited; presented one paper and contributed another two).
- 17.48 First Annual Meeting of the Saudi Chapter of Epilepsy and Epilepsy Workshop, **Riyadh**; October 1999 (invited; presented one paper and contributed another one).
- 17.49 The 4<sup>th</sup> International Congress of the World Muscle Society, **Antalya, Turkey**; October 1999 (presented one paper and contributed another two).
- 17.50 International Pediatric Symposium, Riyadh Al Kharj Hospital (RKH), **Riyadh**; February 2000 (co-chaired the session on Molecular Genetics of Neurologic Disorders).
- 17.51 The 9<sup>th</sup> Annual Symposium of the European Charcot – Marie – Tooth Consortium, **Antwerp**; July 2000 (invited, presented one paper).
- 17.52 The Second International Conference on Disability and Rehabilitation, **Riyadh** October 2000 (invited, presented one paper and co-chaired another session).
- 17.53 Symposium on Pediatrics in Saudi Arabia in the New Millennium, **Riyadh**, January 2001 (invited, presented one paper).
- 17.54 The Fourth International Conference of the ICDR, **Khartoum & Wad Madani**; February 2001 (invited, presented one paper).
- 17.55 The XVII World Congress of neurology, **London**, June 2001 (presented one paper and contributed another two).
- 17.56 Second Scientific Symposium on Autism, **Riyadh**, October 2001 (invited, presented one paper).
- 17.57 Child Health International Congress and The 9<sup>th</sup> Congress of the Union of Arab Pediatric Societies, **Jeddah**, November 2001 (presented one paper and chaired the session on Pediatric Neurology).
- 17.58 The 10<sup>th</sup> Annual Symposium of the European Charcot-Marie-Tooth Consortium, **Antwerp**; November 2001 (contributed one paper).
- 17.59 Autosomal Recessive Charcot-Marie-Tooth (CMT) Network Seminar, **Paris**; June 2002 (invited, presented one paper).



- 17.60 The Xth Internal Congress on Neuromuscular Diseases. **Vancouver**; June 2002 (contributed one paper).
- 17.61 Riyadh Al-Kharj Hospital (RKH) Paediatric Symposium (Paediatric Neurology and General Paediatric Update). **Riyadh**; September 2002 (invited, presented two papers).
- 17.62 The 9<sup>th</sup> International Child Neurology. **Beijing**; September 2002 (presented one paper).
- 17.63 Symposium on Current Problems and Controversies in the Practice of Paediatrics, **Riyadh**; October 2002 (invited, presented one paper).
- 17.64 The 13<sup>th</sup> Conference of the Sudan Association of Paediatricians. **Khartoum**; July 2003 (invited, presented two papers).
- 17.65 Muscular Dystrophy and Related Conditions Symposium (Organised by King Faisal Specialist Hospital and Research Centre). **Riyadh**; September 2003 (Member of the Organizing Committee, presented two invited papers).
- 17.66 The Joint Conference of King Saud University and Taiba Medical Centre on Recent Advances in Neurology and Psychiatry Disorders. **Al Madina Al Munawara**, October 2003 (presented an invited paper).
- 17.67 The Joint Scientific Meeting of Charcot-Marie-Tooth (CMT) and Spastic Paraplegia/Ataxia (SPATAX) International Networks. **Paris**; October 2003 (invited, presented two papers).
- 17.68 The 11<sup>th</sup> Pan Arab Pediatric Conference and the 3<sup>rd</sup> Pan Arab Pediatric Neurology Meeting. **Bahrain**; December 2003 (invited, presented two papers, Chairperson of a Scientific Session).
- 17.69 Symposium on Recent Advances in Medical Imaging, **Riyadh**, February 2004 (contributed one paper).
- 17.70 The First Conference of Pan Arab Faculties of Medicine. **Amman, Jordan**; April 2004 (invited, presented two papers).
- 17.71 The European Human Genetics Conference 2004. **Munich**, June 2004 (contributed one paper).
- 17.72 Paediatric Update Symposium (Organised by Department of Paediatrics, College of Medicine, KSU and Saudi Pediatric Association). **Riyadh**; September 2004 (Members of Organising Committee, presented one paper).
- 17.73 First International Symposium on Autism and Related Disorders. **Riyadh**; September 2004 (Scientific Consultant to the Organising Committee).

- 17.74 The 8<sup>th</sup> Asian and Oceanian Congress of Child Neurology, **New Delhi**; October 2004 (presented two papers, Chairperson for the session on Neurotransmitter Disorder).
- 17.75 International Symposium of Saudi Pediatric Association, **Dhahran**; December 2004 (invited, presented two papers).
- 17.76 Symposium on Epilepsy, King Fahad Hofuf Hospital, **Hofuf**, Saudi Arabia; April 2005 (invited, presented two papers).
- 17.77 The 14<sup>th</sup> Conference of the Sudan Association of Paediatricians. **Khartoum**, July 2005 (invited, presented one paper).
- 17.78 55<sup>th</sup> Annual Meeting of the American Society of Human Genetics, **Salt Lake City, Utah**, October 2005 (contributed one platform presentation).
- 17.79 The XVIII World Congress of Neurology, **Sydney**; November 2005 (presented two papers).
- 17.80 International Conference on Child Health, King Fahad Medical City, **Riyadh**; December 2005 (invited, presented two papers).
- 17.81 The 14<sup>th</sup> Saudi Neuroscience Symposium, **Riyadh**; March 2006 (invited, presented one paper).
- 17.82 XVIII Symposium Neuroradiologicum, World Federation of Neuroradiological Sciences, **Adelaide, Australia**, March 2006 (contributed an Award Winning Poster).
- 17.83 The Joint Scientific Meeting of Charcot-Marie-Tooth (CMT) and Spastic Paraplegia / Ataxia (SPATAX) International Networks, **Paris**; June 2006 (invited, presented two papers)
- 17.84 The 3<sup>rd</sup> International Conference on Child Health, **Jeddah**; November 2006 (invited presented one paper).
- 17.85 The 12<sup>th</sup> Manchester Birth Defects Conference, **Manchester, UK**; November 2006 (contributed one paper).
- 17.86 The 33<sup>rd</sup> Annual Meeting of the American Association for Pediatric Ophthalmology and Strabismus, **Seattle, Washington**; April 2007 (contributed one paper).
- 17.87 59<sup>th</sup> Annual Meeting of the American Academy of Neurology, **Boston**; April/May 2007 (contributed one paper).
- 17.88 The 12<sup>th</sup> International Congress of the World Muscle Society, **Sicily, Italy**; October 2007 (presented one paper)

17.89 The 5<sup>th</sup> Annual Meeting of the American Society of Human Genetics, **San Diego, California**, October 2007 (contributed one paper)

17.90 The 5<sup>th</sup> Congress of the Union of Arab Paediatricians and the 15<sup>th</sup> Conference of the Sudanese Association of Paediatricians, **Khartoum, Sudan**; November 2007. (Member of the Scientific Committee and Co-organizer of pre-congress workshop on Epilepsy in Childhood and Co-chairperson, Neurology Session II (Invited, presented one paper in Plenary Session I and 2 papers in the Epilepsy Workshop).

17.91 The 61<sup>st</sup> American Epilepsy Society Meeting, **Philadelphia, Pennsylvania**; November/December, 2007. (Invited, Special Guest Speaker at the Botanicals Special Interest Group Session).

17.92 The 16<sup>th</sup> Annual Saudi Neuroscience Symposium in Association with the 1<sup>st</sup> Annual Saudi Stroke Association Meeting, the 1<sup>st</sup> Joint Meeting of Saudi Neurosurgical Society and Asian-Australasian Society of Neurological Surgeons, the 5<sup>th</sup> Saudi Chapter of Epilepsy Workshop and Commission on East Mediterranean Affairs (CEMA) Course; and the 6<sup>th</sup> Gulf Neurology Meeting, **Riyadh**, January 2008 (invited, presented two papers).

17.93 The 60<sup>th</sup> Annual Meeting of the American Association of Neurology, **Chicago**; April 2008 (contributed one platform presentation).

17.94 The 2<sup>nd</sup> Al-Baha Advanced Pediatric Conference, **Al-Baha**, Saudi Arabia; April/May 2008 (invited, presented two papers).

17.95 Eighteenth Meeting of the European Neurological Society, **Nice**, France; June 2008, (contributed one paper).

17.96 The 5<sup>th</sup> Latin American on Epilepsy, **Montevideo**, Uruguay; November 2008 (presented one paper).

17.97 The American Academy of Ophthalmology Annual Meeting; **Atlanta, GA, USA**; November 2008 (contributed one paper).

17.98 The 58<sup>th</sup> American Society of Human Genetics Meeting, **Philadelphia, Pennsylvania**, USA; November 2008 (contributed one paper).

17.99 The American Epilepsy Society 62nd Annual Meeting; **Seattle, Washington**; December 2008 (contributed one paper).

17.100 The 17<sup>th</sup> Annual Saudi Neuroscience Symposium in Association with the 2<sup>st</sup> Annual Saudi Stroke Association Meeting, **Almadinah Al-Munawarah**, February 2009 (invited, presented one paper).

- 17.101 The European Society of Human Genetics Meeting , **Vienna**, Austria, May 2009 (contributed two papers).
- 17.102 The Joint Scientific Meeting of Spastic Paraplegia / Ataxia (SPATAX) International Network, **Paris**; June 2009 (invited, presented one paper).
- 17.103 The 3<sup>rd</sup> International Charcot-Marie-Tooth Consortium Meeting, **Antwerp**, Belgium; July 2009 (invited, presented one paper and contributed another).
- 17.104 The 19<sup>th</sup> World Congress of Neurology, **Bangkok**, Thailand; October 2009 (contributed two papers including one platform presentation).
- 17.105 The 16<sup>th</sup> Conference of the Sudanese Association of Paediatricians, **Khartoum, Sudan**; November 2009. (invited, presented one paper).
- 17.106 Human and Medical Genetics Meeting, **Strasbourg**, France, January 2010 (contributed one paper).
- 17.107 The 3<sup>rd</sup> Pan Arab Human Genetics Conference, **Dubai**, UAE, March 2010 (contributed two papers).
- 17.108 The XI<sup>th</sup> International Child Neurology Congress, **Cairo**, May 2010 (contributed one platform presentation).
- 17.109 BIT' 1st World Congress of (NeuroTalk 2010), **Singapore**, June 2010 (invited, presented one paper).
- 17.110 The 15<sup>th</sup> International Congress of The World Muscle Society, **Kumamoto, Japan**, October 2010 (presented one paper).
- 17.111 The 63<sup>rd</sup> Annual Meetig of the American Academy of Neurology, **Honolulu, Hawaii, USA**, April 2011(presented one paper and contributed another).
- 17.112 The 1<sup>st</sup> Medical Journalism Symposium, **Riyadh**, May 2011 (co-chaired two sessions).
- 17.113 European Human Genetics Conference 2011, **Amsterdam**, May/June 2011 (contributed one platform presentation).
- 17.114 The 29<sup>th</sup> International Epilepsy Congress, **Rome**, August/September 2011 (contributed one platform presentation).
- 17.115 The 12th International Congress of Human Genetics / 61st Annual Meeting of the American Society of Human Genetics, **Montreal (Quebec)**, October 2011(contributed one platform presentation).

17.116 The 16<sup>th</sup> Conference of the Sudanese Association of Paediatricians, **Khartoum, Sudan**, October 2009. (Member of the Scientific Committee and Co-organizer of pre-congress workshop on Epilepsy in Childhood and Co-chairperson, Neurology Session (Invited, presented one paper in Plenary Session and 2 papers in the Epilepsy Workshop).

17.117 The 17<sup>th</sup> Conference of the Sudanese Association of Paediatricians, **Khartoum, Sudan**, October 2011. (Member of the Scientific Committee and Co-organizer of pre-congress workshop on Epilepsy and Neurophysiology; Co-chairperson, Neurology Session (Invited, presented one paper in Plenary Session and 2 papers in the Epilepsy and Neurophysiology Workshop).

17.118 The 20th World Congress of Neurology, **Marrakesh, Morocco**, November 2011 (Presented one oral presentation, contributed another oral presentation, and Co-chaired a Child Neurology session.

17.119 The 19<sup>th</sup> Annual Saudi Neuroscience Symposium, **Dammam, Saudi Arabia**, November 2011 (Invited, presented one paper).

17.120 The 4<sup>th</sup> Conference and Exhibition of the Saudi Pediatric Association, **Riyadh**, April 2012 (Invited, presented one paper).

17.121 The European Human Genetics Conference 2012, **Nurnberg, Germany**, June 2012 (contributed one platform presentation).

17.122 The 17<sup>th</sup> Lebanese Epidemiological Association Annual Conference, **Beirut, Lebanon**, November 2012 (Co-invited by the WHO Regional Office, presented one paper).

17.123 The 1<sup>st</sup> International Pediatric Neurology Conference, **Doha, Qatar**, March 2013 (Invited, presented one paper).

17.124 The Joint Scientific Meeting of Spastic Paraplegia / Ataxia (SPATAX) International Network, **Paris**, June 2013 (contributed one platform presentation).

17.125 The 17th International Congress of Parkinson's Disease and Movement Disorders, **Sydney**, June 2013 (contributed one paper).

17.126 The 12<sup>th</sup> Asian and Oceanian Congress of Child Neurology, **Riyadh**, September 2013 (Invited, presented one paper, Chairperson for the session on Neuromuscular Disorders).

17.127 The 18<sup>th</sup> Conference of the Sudanese Association of Paediatricians, **Khartoum, Sudan**, November 2013. (Member of the Scientific Committee, Chairperson, Neurology Session (Invited, presented one paper in Plenary Session).

17.128 The Third Annual Emirates International Pediatric Neurology Conference (EIPN). **Dubai , United Arab Emirates**, February 2014. (Invited, presented two papers and Co-chaired a session).

17.129 The 66<sup>th</sup> Annual Meetig of the American Academy of Neurology, **Philadelphia, PA, USA**, April/May 2014 (presented one platform paper).

17.130 The 13th International Child Neurology Congress, **Foz do Iguaçu, Brazil**, May 2014. (Presented one platform paper and poster and contributed another two posters).

17.131 The 18th International Congress of Parkinson's Disease and Movement Disorders, **Stockholm**; June 2014 (contributed one paper).

17.132 The First Saudi Neuromuscular Conference and Electro-diagnostics Workshop, **Al Khobar, Saudi Arabia**, December 2014. (Invited, presented one paper in Plenary Session).

17.133 The International Child Neurology Association (ICNA) Educational Meeting, **Khartoum, Sudan**, January 2015. (Invited, Member of the Scientific Committee; Co-chairperson, Platform Session; Presented two papers in Plenary Sessions).

17.134 The Sixth Annual Conference of The Post-Graduate & Scientific Research Conference, University of Khartoum, **Khartoum, Sudan**, February 2015. (Invited, Co-Chairperson, Young Researcher Award Session; presented two papers in Platform Sessions).

17.135 The 67<sup>th</sup> American Academy of Neurology Annual Meeting, **Wahington DC**, April 2015. (Presented one poster).

17.136 The XXII World Congress of Neurology, **Santiago, Chile**, October/November 2015. (Presented one paper in Platform Session and one poster).

17.137 The 6<sup>th</sup> International Conference of Sudanese Society of Neurological Sciences (SSNS), **Khartoum, Sudan**, January 2016 (Invited ; Co-chairperson, Platform Session; Presented one paper in Plenary Session).

17.138 The First Saudi Pediatric Neurology Society Symposium, **Riyadh, Saudi Arabia**, March 2016. (Invited, presented one paper in Plenary Session).

## **18. OTHER SCIENTIFIC LECTURES:**

18.1 *January 1982*: "Unusual Muscular Dystrophy in an Extended Sudanese Kindred"; Clinical Meeting, Department of Child Health, University of Newcastle Upon Tyne, UK.

18.2 *June 1984*: Two lectures on diphtheria and acute bacterial meningitis in Sudanese children, International Child Health Unit, Department of Paediatrics, Uppsalla University, Sweden.

18.3 *Feb. 1986* : "Neurological Problems from Sudan" Meeting of Division of Neurological Medicine and Surgery, Regional Neurological Centre, Newcastle General Hospital, Newcastle Upon Tyne, UK.

18.4 *June 1986*: "Neurocutaneous Syndromes in Sudanese Children" Clinical Meeting, Department of Child Health, University of Newcastle Upon Tyne, UK.

18.5 *Sept. 1987*: "Infections and Childhood Priorities in Sudan" Clinical Meeting, Department of Paediatrics, Orebro Medical Centre Hospital, Sweden.

18.6 *1989 – 1991*: Seven lectures on childhood acute bacterial meningitis, spectrum of infectious diseases in Sudan and medical problems in African Refugees. Presented at the Department of Clinical Microbiology and Immunology, Paediatrics and Infectious Diseases, Orebro Medical Centre Hospital, Orebro; International Child Health Unit, Department of Paediatrics, Department of Infectious Diseases and Institute of Clinical Bacteriology, Uppsala University, Sweden.

18.7 *June 1992*: "Acute Bacterial Meningitis: New Diagnostic Methods and Molecular Epidemiology" Riyadh Microbiology/Infectious Diseases/Tropical Medicine Club Meeting, Saudi Arabia.

18.8 *Oct. 1992*: "Mitochondrial Myoencephalopathy in Infancy and a Family Study" Riyadh Paediatric Club Meeting, Saudi Arabia.

18.9 *Dec. 1992*: "Muscle Biopsy : A Window to Paediatric Neurology" Riyadh Pathology Group Meeting, Riyadh, Saudi Arabia.

18.10 *Oct. 1994*: "The Story of 'Adhalin' and Severe Childhood Autosomal Recessive Muscular Dystrophy" Riyadh Paediatric Club Meeting, Saudi Arabia.

18.11 *Dec. 1995*: "The Face and Hands Predict the Brain" Riyadh Paediatric Club Meeting, Saudi Arabia.

18.12 *Dec. 1996*: "The Story of Adhalin" Tuesday Clinical Meeting, Security Forces Hospital, Riyadh, Saudi Arabia.

18.13 *May 1997*: "Where Can We Find a Muscle to Take a Biopsy from?" Neuromuscular Club Meeting, Riyadh Neurosciences Club, Riyadh, Saudi Arabia.

18.14 *Sept. 1997* : "Floppy Infant Syndrome" Neuromuscular Club Meeting, Riyadh Neurosciences Club, Riyadh, Saudi Arabia.

18.15 *Feb. 1998*: "Childhood Muscular Dystrophies: Pathophysiology, Clinical Features and Management" Handicap Children Centre for Saudi Benevolent Association for Disabled Children, Riyadh, Saudi Arabia.

18.16 *March 1998*: "Floppy Infant Syndrome" Al Hammadi Hospital, Riyadh, Saudi Arabia.

18.17 *April 1998*: “The Floppy Infant Syndrome” Sulaimania Children’s Hospital, Riyadh, Saudi Arabia.

18.18 *May 1998*: “The Floppy Infant Syndrome” Children’s Hospital, Riyadh Medical Complex, Riyadh, Saudi Arabia.

18.19 *Oct. 1998*: “Family with a Novel Form of Congenital Muscular Dystrophy” Neuromuscular Club Meeting, Riyadh Neurosciences Club, Riyadh, Saudi Arabia.

18.20 *Nov. 1998*: “Recent Advances in the Pathology of Muscular Dystrophy” Riyadh Pathology Club Meeting, Riyadh, Saudi Arabia.

18.21 *Nov. 1998*: “Diagnosis and Clinical Features of Epilepsy” Suleimania Children’s Hospital, Riyadh, Saudi Arabia.

18.22 *May 2000*: “Monotherapy in Childhood Epilepsy” Department of Paediatrics, Security Forces Hospital, Riyadh, Saudi Arabia.

18.23 *April 2001*: “Stroke in Saudi Children” Prince Salman Centre for Disability Research, Riyadh, Saudi Arabia.

18.24 *January 2004*: “New Childhood Muscular Dystrophies from the Old World” Department of Medical Genetics Grand Round, King Faisal Specialist Hospital and Research Centre, Riyadh, Saudi Arabia

18.25 *March 2006*: “Stroke in Saudi Children: Epidemiology and Risk Factors” Prince Salman Centre for Disability Research Academic Lecture, Riyadh, Saudi Arabia

18.26 *January 2010*: “ Publishing in *Nature* and *Science*” King Saud University Public Lecture organized by the Deanship for Scientific Research.

18.27 *May 2012*: “The Era of Pediatric Neurogenetics” Department of Medical Genetics Grand Round, King Faisal Specialist Hospital and Research Centre, Riyadh, Saudi Arabia.

18.28 *November 2013*: Lecture, Fifth Clinical Course in Pediatrics (Saudi Board, MRCPC and Arab Board Candidates), College of Medicine, King Saud University, Riyadh.

18.29 *August 2015*: “Approach to Hereditary Ataxia/Spastic Paraplegia: Phenotype Leading to Genotype” Department of Medical Genetics Grand Round, King Faisal Specialist Hospital and Research Centre, Riyadh, Saudi Arabia.



## 19. PUBLICATIONS:

- Publications were highlighted by *Nature* (*Nature Index*):

[http://www.nature.com/nature/journal/v532/n7600\\_supp\\_ni/full/532S19a.html](http://www.nature.com/nature/journal/v532/n7600_supp_ni/full/532S19a.html)

### 19.1 Theses, Supplements and Book Chapters

19.1.1 **MPCH Thesis.** Unusual Muscular Dystrophy in an Extended Sudanese Kindred. (A thesis submitted in partial fulfillment of the requirement for the Degree of MPCH, University of Khartoum, April 1980).

19.1.2 **MD Thesis.** Unusual Muscular Dystrophy in an Extended Sudanese Kindred - Doctor of Medicine Thesis (Degree granted with distinction by the University of Khartoum, December 1982).

19.1.3 **Doctor of Medical Science Thesis.** Childhood Acute Bacterial Meningitis in the Sudan: An epidemiological, clinical laboratory study. *Scandinavian Journal of Infectious Diseases* 1990; Supplement 66:1 - 103. (Uppsala University, Sweden, May 1990).

19.1.4 **Salih MAM.** Neuromuscular Disorders Among Arabs. In: Teebi AS, Farag TF, eds. *Genetic Disorders Among Arab Populations*. London : Oxford University Press, 1997.

19.1.5 **Salih MAM.** Genetic disorders in the Sudan. In : Teebi AS, Farag TF, eds. *Genetic Disorders Among Arab Populations*. London : Oxford University Press, 1997.

19.1.6 **Salih MA.** Childhood Stroke. *Saudi Medical Journal* 2006; Supplement 1(Vol.27): S1- S111.

19.1.7 Boerkoel C, Hirano R, **Salih M**, Takashima H (October 2007) Spinocerebellar Ataxia with Axonal Neuropathy in: GeneReviews at GeneTests: Medical Genetics Information Resource [database online]. Copyright, University of Washington, Seattle, 1997-2007. Available at: <http://www.ncbi.nlm.nih.gov/books/NBK1105/>

19.1.8 Cheryl Walton, Heidrun Interthal, Ryuki Hirano, **Mustafa A.M. Salih**, Hiroshi Takashima and Cornelius Boerkel. . Spinocerebellar Ataxia with Axonal Neuropathy. In: Ahmad, Shamim, ed. *Diseases of DNA Repair*. New York: Springer, 2010.

19.1.9 **Salih MAM.** Muscular Dystrophies and Myopathies in Arab Populations. In : Teebi AS, ed. *Genetic Disorders Among Arab Populations* (Second edition). New York: Springer, 2010.

19.1.10 **Salih MAM.** Genetic Disorders in Sudan. In: Teebi AS, ed. *Genetic Disorders Among Arab Populations* (Second edition). New York: Springer, 2010.

19.1.11 **Salih MAM.** **Seven chapters** in: Elzouki AY, ed. *the Textbook of Clinical Pediatrics* (Second edition). New York: Springer, 2012.

- 19.1.12 Carmignac V, Suominen T, Hackman P, Udd B, **Salih MA**. Salih Myopathy (January 2012) in: GeneReviews at GeneTests: Medical Genetics Information Resource [database online]. Copyright, University of Washington, Seattle , 1997-2010. Available at: <http://www.ncbi.nlm.nih.gov/books/NBK83297/>
- 19.1.13 Fam HK, **Salih MAM**, Takashima H, Boerkoel CF: Spinocerebellar Ataxia with Axonal Neuropathy, Autosomal Recessive (April 2012) in: GeneReviews at GeneTests: Medical Genetics Information Resource [database online]. Copyright, University of Washington, Seattle, 1997-2010. Available at: <http://www.ncbi.nlm.nih.gov/books/NBK1105/>
- 19.1.14 **Salih MA**. Neural Tube Defects. Saudi Medical Journal 2014; Supplement (Vol.35): S1- S90.
- 19.1.15 Azzedine H, LeGuern E, **Salih MA**. Charcot-Marie-Tooth Neuropathy Type 4C. 2008 Mar 31 [Updated 2015 Oct 15]. In: Pagon RA, Adam MP, Ardinger HH, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2015. Available at: <http://www.ncbi.nlm.nih.gov/books/NBK1340/>

## 19.2Papers

- 19.2.1 **Salih MAM**, Hashem N. Pendred syndrome in a Sudanese family. Sudanese Journal of Paediatrics 1978 ; 2 : 25-34.
- 19.2.2 El Karim O, **Salih MAM**. Morbidity and mortality from measles in an urban community of the Sudan. Annals of Tropical Paediatrics, Medicine and Parasitology 1980 ; 75 : 227-30.
- 19.2.3 El Karim OA, **Salih MAM**. Extra-alveolar air collection in measles. Annals of Tropical Paediatrics 1981 ; 1 : 57-60.
- 19.2.4 **Salih MAM**, Suliman GI, Hassan HS. Complications of diphtheria seen during the 1978 outbreak in Khartoum. Annals of Tropical Paediatrics 1981 ; 1 : 97-101.
- 19.2.5 **Salih MAM**, Omer MIA, Karrar O, Bayoumi RA, Johnson M. Severe autosomal recessive muscular dystrophy in an extended Sudanese kindred. Developmental Medicine and Child Neurology 1983 ; 25 : 43-52.
- 19.2.6 **Salih MAM**, Roberts DF, Omer MIA. Reflections on muscular dystrophy in Sudanese kindred. Clinical Genetics 1983 ; 23 : 325- 328.
- 19.2.7 **Salih MAM**, Ekmejian A, Omer MIA. Respiratory insufficiency in a severe autosomal recessive form of muscular dystrophy. Annals of Tropical Paediatrics 1984 ; 4: 45-48.
- 19.2.8 **Salih MAM**. Laboratory diagnosis of meningitis : An overview in relation to the Sudan. Sudanese Journal of Paediatrics 1984 ; 3 : 10-19.
- 19.2.9 **Salih MAM**, Suliman GI, Hassan HS. Unusual sites of diphtheritic membrane and cervical oedema. Sudanese Journal of Paediatrics 1984 ; 3 : 52-62.

- 19.2.10 **Salih MAM**, Lake BD, El Hag MA, Atherton DJ. Lethal epidermolytic epidermolysis bullosa : A new autosomal recessive type of epidermolysis bullosa. *British Journal of Dermatology* 1985 ; 11 : 135-143 (Selected for abstracting by the Editors of International Synopses).
- 19.2.11 Eltom M, El Mahdi EMA, **Salih MAM**, Mukhtar E, Omer MIA. A new focus of endemic goitre in the Sudan . *Tropical and Geographical Medicine* 1985 ; 37 : 15-21.
- 19.2.12 **Salih MAM**, El Hakeem HS, Suliman GI, Khatim AS. An epidemiological study of the 1978 outbreak of diphtheria in Khartoum province. *Journal of Tropical Paediatrics* 1985; 31 : 8-12.
- 19.2.13 Eltom M, **Salih MAM**, Bostrom H, Dahlberg PA. Differences in aetiology and thyroid function in endemic goitre between rural and urban areas of the Darfur Region of the Sudan. *Acta Endocrinologica* 1985 ; 108 : 356-360.
- 19.2.14 **Salih MAM**. Childhood muscular dystrophy : An African review. *Annals of Tropical Paediatrics* 1985 ; 5 : 167-173.
- 19.2.15 **Salih MAM**, Bender DA, McCreanor GM. A lethal familial pellagra. - like skin lesion associated with neurological and developmental impairment and the development of cataracts. *Pediatrics* 1985 ; 76 : 787-793.
- 19.2.16 **Salih MAM**. A clinical profile of diphtheria in Sudanese children. *Sudanese Journal of Paediatrics* 1986 ; 5 : 31-36.
- 19.2.17 **Salih MAM**, Ahmed HS, Hofvander Y, Danielsson D, Olcen P. Rapid diagnosis of bacterial meningitis by an enzyme immunoassay of cerebrospinal fluid. *Epidemiology and Infection* 1989 ; 103 : 301- 310. (Selected by the Editors of the Clinical Digest Series as outstanding in its field).
- 19.2.18 **Salih MAM**, Ahmed HS, Karrar ZA, Kamil I, Osman KA, Palmgren H, Hofvander Y, Olcen O. Features of the major epidemic of group A meningococcal meningitis in Khartoum, Sudan in 1988. *Scandinavian Journal of Infectious Diseases* 1990 ; 22 : 161- 170.
- 19.2.19 **Salih MAM**, El Haq AI, Ahmed HS, Bushara M, Yassin I, Omer MIA, Hofvander Y, Olcen P. Endemic bacterial meningitis in Sudanese children : aetiology, clinical findings, treatment and short term outcome. *Annals of Tropical Paediatrics* 1990 ; 10 : 203-210.
- 19.2.20 **Salih MAM**, Ahlsten G, Stalberg E, Schmidt R, Sunnegardh J, Michaelsson M, Gamstrop I. Friedreich's ataxia in 13 children : Clinical, neurophysiological and echocardiographic features. *Journal of Child Neurology* 1990 ; 5 : 321-326. (Selected by the Editors of the Clinical Digest Series as outstanding in its field).

19.2.21 **Salih MAM**, Ahmed HS, Osman KA, Kamil I, Palmgren H, Hofvander Y, Olcen P. Clinical features and complications of epidemic group A meningococcal disease in Sudanese children. *Annals of Tropical Paediatrics* 1990 ; 10 :231-238.

19.2.22 **Salih MAM**, Danielsson D, Backman A, Caugant DA, Achtman M, Olcen P. Characterization of epidemic and non-epidemic *Neisseria meningitidis* serogroup A strains from Sudan and Sweden. *Journal of Clinical Microbiology* 1990 ; 28 : 1711-19.

19.2.23 Karrar ZA, Bashaar S, **Salih MAM**. Mortality among children with meningococcal disease : An analysis of deaths during the 1988 epidemic. *Sudan Medical Journal* 1990 ; 28 : 1-4.

19.2.24 **Salih MAM**, Tuvemo T. Diabetes insipidus, diabetes mellitus, optic atrophy and deafness (DIDMOAD Syndrome). A clinical study in two Sudanese families. *Acta Paediatrica Scandinavica* 1991 ; 8 : 567-572.

19.2.25 **Salih MAM**, Khaleefa OH, Bushara M, Taha ZB , Musa ZA, Kamil I, Hofvander Y, Olcen P. Long term sequelae of childhood acute bacterial meningitis in a Developing Country : A study from the Sudan. *Scandinavian Journal of Infectious Diseases* 1991; 23 : 175-182.

19.2.26 **Salih MAM**, Fredlund H, Hugosson S, Bodin L, Olcen P. Different sero-prevalences of antibodies against *Neisseria meningitidis* serogroup A and *Haemophilus influenzae* type b in Sudanese and Swedish children. *Epidemiology and Infection* 1993 ; 110 : 307-316.

19.2.27 **Salih MAM**, El Bushra HM, Satti SA, Ahmed MEA, Kamil I. Attitudes and practices of breast feeding in Sudanese urban and rural communities. *Tropical and Geographical Medicine* 1993 ;45 : 171-174.

19.2.28 Al-Herbish AS, **Salih MAM**, Al-Husain M, Al-Jurayyan NAM, Patel PJ, Palkar V. X-linked adrenoleukodystrophy in the Arab ethnic group : presentation and management of a child. *Medical Science Research* 1993 ; 21 : 439-441.

19.2.29 Herrmann B, **Salih MAM**, Yousif BE, Abdelwahab O, Mardh PA. Chlamydial etiology of acute lower respiratory tract infections in children in the Sudan. *Acta Paediatrica Scandinavica* 1994 ; 83 : 169-172.

19.2.30 **Salih MAM**, Mohamed EH, Galgan V, Jones B, Hellsing K, Bani IA, Alasha E. Selenium in malnourished Sudanese children : Status and interaction with clinical features. *Annals of Nutrition and Metabolism* 1994 ; 38 : 68-74.

19.2.31 Naim-Ur-Rahman, **Salih MAM**, Jamjoom AH, Jamjoom ZA. Congenital intramedullary lipoma of the dorso-cervical spinal cord with intracranial extension : Case report. *Neurosurgery* 1994 ; 34 : 1081-1084.

- 19.2.32 **Salih MAM**, Herrmann B, Grandien M, El Hag MM, Yousif BE, Abdelbagi M, Mardh P-A, Ahmed HS. Viral pathogens and clinical manifestations associated with acute lower respiratory tract infections in children of the Sudan. *Clinical and Diagnostic Virology* 1994; 2:201-209.
- 19.2.33 El Bushra HM, **Salih MAM**, Satti SAR, Ahmed MEA, Kamil IA. Infant feeding practices in urban and rural communities of the Sudan. *Tropical and Geographical Medicine* 1994; 46 : 309-312.
- 19.2.34 Dincsoy MY, **Salih MAM**, Al-Jurayyan N, Al Saadi M, Patel PJ. New Syndrome: Multiple congenital malformations in two sibs reminiscent of hydrolethrus and pseudotrismus 13 syndromes. *American Journals of Medical Genetics* 1995; 56 : 317-321.
- 19.2.35 Patel PJ, Kolawole TM, Malabarey TM, Al-Herbish AS, Al-Jurayyan NAM, **Salih M**. Adrenoleukodystrophy : CT and MRI findings. *Paediatric Radiology* 1995; 25: 256-258.
- 19.2.36 **Salih MAM**, Andeejani AMI , Gader AMA, Kolawole T, Palkar V. Moyamoya syndrome associated with protein C deficiency. *Medical Science Research* 1995; 23 : 573-575.
- 19.2.37 **Salih MAM**, Kabiraj M, Gascon GG, Al Jarallah AS, Al Zamil FA. Typical and atypical presentations of pyridoxine-dependent seizures. *Saudi Medical Journal* 1995; 16 ; 347-351.
- 19.2.38 Jamjoom A, Tjan T, Jamjoom Z, **Salih MAM**, Rahman N. Embolization of intramedullary arteriovenous malformation of spinal cord. *Saudi Medical Journal* 1995; 16:565-568.
- 19.2.39 **Salih MAM**, Ahmed AA, Ahmed HS, Olcen P. An ELISA assay for the rapid diagnosis of acute bacterial meningitis. *Annals of Tropical Paediatrics* 1995; 15 : 273-278.
- 19.2.40 **Salih MAM**, Mahdi A, Al-Rikabi AC, Al-Bunyan M, Roberds SL, Anderson RD, Campbell KP. Clinical and molecular pathological features of severe childhood autosomal recessive muscular dystrophy in Saudi Arabia. *Developmental Medicine and Child Neurology* 1996; 38 : 262-270.
- 19.2.41 Haider N, **Salih MAM**, Al-Rasheed S, Al-Mofada S, Krahn PM, Kabiraj M. Non-ketotic hyperglycinemia : A life threatening disorder in Saudi Newborns. *Annals of Saudi Medicine* 1996; 16:400-404.
- 19.2.42 Al Husain MA, Zaki OK, **Salih MAM**. Case report : Deletion of short arm of chromosome 3 in a Saudi girl. *Annals of Saudi Medicine* 1996; 16 :455-457.

19.2.43 **Salih MAM**, Mahdi AH, Al-Jarallah AA, Al-Jarallah AS, Al-Saadi M, Hafeez MA, Aziz SA. Childhood neuromuscular disorders in Saudi Arabia : A decade's experience in a tertiary hospital. *Annals of Tropical Paediatrics* 1996;16:271-280.

19.2.44 Al Jarallah AA, **Salih MAM**, Al Nasser MN, AL Zamil FA, Al Gethmi J. Rett syndrome in Saudi Arabia : Report of six patients. *Annals of Tropical Paediatrics* 1996;16:347-352.

19.2.45 **Salih MAM**, Sunada Y, Al Nasser M, Ozo CO, Al Turaiki MHS, Akbar M. Campbell KP. Muscular dystrophy associated with  $\beta$  - dystroglycan deficiency. *Annals of Neurology* 1996;40:925-928. (Rated by Current Opinion in Neurology [1997;10:436-442] to be of outstanding interest).

19.2.46 Seidahmed MZ, Sunada Y, Ozo CO, Hamid F, Campbell KP, **Salih MAM**. Lethal congenital muscular dystrophy in two sibs with arthrogryposis multiplex : New entity or variant of cobblestone lissencephaly syndrome? *Neuropediatrics* 1996;27:305-310.

19.2.47 Ahmed AA, **Salih MAM**, Ahmed HS. Post-epidemic acute bacterial meningitis in Sudanese children. *East African Medical Journal* 1996;73:527-532.

19.2.48 Allamand V, Sunada Y, **Salih MAM**, Straub V, Ozo CO, Al Turaiki MHS, Akbar M, Kolo T, Colognato I, Zhang X, Sorokin LM, Yurchenco PD, Tryggvasson K, Campbell KP. Mild congenital muscular dystrophy in two patients with an internally deleted laminin  $\alpha 2$  - chain. *Human Molecular Genetics* 1997;6:747-752.

19.2.49 **Salih MAM**, Kabiraj M, Al-Jarallah AS, El Desouki M, Othman S, Palkar VA. Hemiconvulsion-hemiplegia-epilepsy syndrome: A clinical, electroencephalographic and neuroradiologic study. *Child's Nervous System* 1997;13:257-263.

19.2.50 Hashim MSK, **Salih MAM**, El Hag AA, Karrar ZA, Osman EM, El-Shiekh FS, El Tilib IA, Attala NE. AIDS and HIV infection in Sudanese children: A clinical and epidemiological study. *AIDS Patient Care and STDs* 1997;11:331-337.

19.2.51 Al-Herbish AS, Al-Jurayyan NA, Ba Abbad R, Jan M, **Salih MAM**. Epileptic laughter and precocious puberty due to hypothalamic hamartoma : a case report with review of the literature. *Medical Science Research* 1997;25:143-144.

19.2.52 Abdalla BA, **Salih MAM**, Yousif EA, Omer MIA. Whooping cough in Sudanese children. *East African Medical Journal* 1998; 75:51- 56.

19.2.53 Andeejani AMI, **Salih MAM**, Kolawole T, Gader AMA, Palkar V, Malabary TO, Al Damegh S, Al Boukai A. Moyamoya disease with unusual radiographic findings: Review of the literature. *Journal of the Neurological Sciences* 1998;159:11-16.

19.2.54 **Salih MAM**, Al Rayess M, Cutshall S, Urtizberea JA, Al Turaiki MHS, Ozo CO, Straub V, Akbar M, Abid M, Andeejani A, Campbell KP. A novel form of familial congenital muscular dystrophy in two adolescents. *Neuropediatrics* 1998;29:289-293.

- 19.2.55 Al-Tahan AY, Divakaran MP, Kabouris M, Bohlega S, **Salih M**, Ogunniyi A, Al-Ghanmi H. A novel autosomal recessive "Huntigton's disease - like" neurodegenerative disorder in a Saudi family. *Saudi Medical Journal* 1999;20:85-89.
- 19.2.56 Khan - Gori SN, Murshid WR, Samarkandi AHH, Al-Salman M, **Salih M**. Use of propofol and sevoflurane in moyamoya disease. Case reports and literature review. *Middle East Journal of Anesthesiology* 1999;15:73 - 81.
- 19.2.57 Al Rashed A, Al-Jarallah AA, **Salih M**, Kolawole T, Al-Jarallah J. Sotos syndrome (cerebral gigantism ): a clinical and radiological study of 14 cases from Saudi Arabia. *Annals of Tropical Paediatrics* 1999; 19 : 197 - 203.
- 19.2.58 Seidahmed MZ, Rooney DE, **Salih M**, Abdel Basit OB, Shaheed MM, Abdullah MA, Abomelha A. A case of partial trisomy 2q3 with clinical manifestations of Marshall - Smith syndrome. *American Journal of Medical Genetics* 1999; 85:185-188.
- 19.2.59 **Salih MAM**, Maisonobe T, Kabiraj M, Al Rayess M, Al Turaiki MHS, Akbar M, Tahan A, Urtizberea JA, Grid D, Hamadouche T, Guilbot A, Brice A, Leguern E. Autosomal recessive hereditary neuropathy with focally folded myelin sheaths and linked to chromosome 11q23 : A distinct and homogeneous entity. *Neuromuscular Disorders* 2000;10:10-15.
- 19.2.60 Nicole S, White PS, Topaloglu H, Beigthon P, **Salih MAM**, Hentati F, Fontaine B. The human CDC42 gene: genomic organization, evidence for the existence of a putative pseudogene and exclusion as a SJS1 candidate gene. *Human Genetics* 1999; 105:98-103.
- 19.2.61 Al Husain M, **Salih MAM**, Zaki OK, Al Othman L, Al Nasser MN. A clinical study of mentally retarded children with fragile X -syndrome in Saudi Arabia. *Annals of Saudi Medicine* 2000;20:16-19.
- 19.2.62 Al Nemri AR, Kilani RA, **Salih MAM**, Al Ajlan AA. Embryonal rhabdomyosarcoma and chromosomal breakage in a newborn infant with Dubowitz syndrome. *American Journal of Medical Genetics* 2000;92:107-110
- 19.2.63 Bolino A, Levy ER, Muglia M, Conforti FL, Le Guern E, **Salih MAM**, Georgiou DM, Christondoulou PK, Hausmanowa-Pertusewicz I, Mandich P, Gamberdella A, Quattrone A, Devoto M, Monaco AP. Genetic refinement and physical mapping of the CMT4B gene on chromosome 11q22. *Genomics* 2000; 63:271-278.

19.2.64 Bolino A, Muglia M, Conforti FL, Le Guern E, **Salih MAM**, Georgiou D- M, Christodoulou K, Haumanowa – Petrusiewicz I, Mandich P, Schnone A, Gambardella A, Bono F, Quattrone A, Devoto M, Monaco AP. Charcot – Marie – Tooth type 4B is caused by mutations in the gene encoding the myotubularin related protein 2, MTMR2, on chromosome 11q22. *Nature Genetics* 2000; 25:17-19. (The first identified gene in autosomal recessive hereditary motor and sensory neuropathy; Rated by *Current Opinion in Neurology* [2000;13:565-568] to be of outstanding interest).

19.2.65 Takashima H, Boerkoel CF, John J, Saifi GM, **Salih MAM**, Armstrong D, Mao Y. Quioco FA, Roa BB, Nakagawa M, Stockton DW, Lupski JR. Mutation of TDP1, encoding a topoisomerase 1-dependent DNA damage repair enzyme, in spinocerebellar ataxia with axonal neuropathy. *Nat Genet* ; 32 : 267-272.

19.2.66 Al-Faris EA, Abdulghani HM, Mahdi AH, **Salih MA**, Al-Kordi AG. Compliance with appointments and medications in a pediatric neurology clinic at a University Hospital in Riyadh, Saudi Arabia. *Saudi Med J.* 2002 ; 8 : 969-974.

19.2.67 Jen J, Coulin CJ, Bosley TM, **Salih MA**, Sabatti C, Nelson SF, Baloh RW. Familial horizontal gaze palsy with progressive scoliosis maps to chromosome 11q23-25. *Neurology* . 2002 ; 59 : 432-425 (Editorial Comment : 304-305).

19.2.68 Al-Jumah M, Majumdar R, Al-Rajeh S, Chaves-Carballo E, **Salih MM**, Awada A, Al-Shahwan S, Al-Uthaim S. Deletion mutations in the dystrophin gene of Saudi patients with Duchenne and Becker muscular dystrophy. *Saudi Med J.* 2002 ; 23 : 1478-1482.

19.2.69 Zaidan R, **Salih MAM**, Daif AK, Al-Tahan AR. Non paraneoplastic opsodonus myoclonus syndrome. *Neuroscience* 2003 ; 8 : 120-122.

19.2.70 Previtali SC, Zerega B, Sherman DL Brophy PJ, Dina G, King RH, **Salih MM**, Feltri L, Quattrini A, Ravazzolo R, Wrabetz L, Monaco AP, Bolino A. Myotubularin-related 2 protein phosphatase and neurofilament light chain protien, both mutated in CMT neuropathies, interact in peripheral nerve. *Hum Mol Genet.* 2003 ; 12 : 1713-1723.

19.2.71 Lee MJ, Stephenson DA, Groves MJ, Sweeney MG, Davis MB, An SF, Houlden H. **Salih MA**, Timmerman V, de Jonghe P, Auer-Grumbach M, Di Maria E, Scaravilli F, Wood NW, Reilly MM. Hereditary sensory neuropathy is caused by a mutation in the delta subunit of the cytosolic chaperonin-containing t-complex peptide-1 (Cct4) gene. *Hum Mol Genet,* 2003 ; 12 : 1917-1925.

19.2.72 Essa M, El-Medany Y, Hajjar W, Hariri Z, Al-Mulhim F, **Salih M**, Ashour M, Al-Kattan K. Maximal thymectomy in children with myasthenia gravis. *Eur J Cardiothorac Surg,* 2003 ; 24 : 187-189. (discussion 190-191).



19.2.73 Al-Jumah M, Majumdar R, Al-Rajeh S, Awada A, Chaves-Carbello E, **Salih M**, Al-Shahwan S, Al-Subiey K, Al-Uthaim S. Molecular analysis of the spinal muscular atrophy and neuronal apoptosis inhibitory protein genes in Saudi patients with spinal muscular atrophy. *Saudi Med J*, 2003 ; 24 : 1052-1054.

19.2.74 Jen JC, Chan W-M, Bosley TM, Wan J, Carr JR, Rub U, Shattuck D, Salamon G, Kudo LC, Ou J, Lin DDM, **Salih MAM** and 23 other authors. Mutations in a human ROBO gene hindbrain axon pathway crossing and morphogenesis. *Science*. 2004; 304 : 1509-1513. (Article featured on the cover of *Science*, highlighted in *Perspective*: 1455-1456 ; and as *Scienceexpress Report* on April 22, 2004).

19.2.75 Fernet M, Gribaa M, **Salih MA**, Seidahmed MZ, Hail J, Koenig M. Identification and functional consequences of a novel MRE11 mutation affecting 10 Saudi Arabian patients with the ataxia telangiectasia-like disorder. *Hum Mol Genet*. 2005 ; 14 : 307-318. [Epub 2004 Dec 01].

19.2.76 Rashed MS, Saadallah AA, Rahbeeni Z, Eyaid W, Seidahmed MZ, Al-Shahwan S, **Salih MA**, Osman ME, Al-Amoudi M, Al-AhaidibL, Jacob M. Determination of urinary S-sulphocysteine, xanthine and hypoxanthin by liquid chromatography-electrospray tandem mass spectrometry. *Biomed Chromatogr*. 2005;19 :223-230 [Epub 2004 Nov 23].

19.2.77 Bosley TM, **Salih MAM**, Jen JC, Lin DDM, Oystreck D, Abu-Amero KK, MacDonald DB, al-Zayed Z, Al Dhalaan H, Kansu T, Stigsby B, Baloh RW. Neurologic features of horizontal gaze palsy and progressive scoliosis with mutations in ROBO3. *Neurology* 2005;64:1196-1203.

19.2.78 Tischfield MA, Bosley TM, **Salih MA**, Alorainy IA, Sener EC, Nester MJ, Oystreck DT, Chan WM, Andrews C, Ericson RP, Engle EC. Homozygous HOXA1 mutations disrupt human brainstem, inner ear, cardiovascular and cognitive development *Nat Genet*. 2005; 37:1035-7. [Epub 2005 Sep 11]. The first characterization of homozygous HOX gene mutation in humans. Highlighted in several web sites and in *HotSpots of Clinical Genetics* (Coburn B. *Clinical Genetics* 2006; 69:205).

19.2.79 Alorainy IA, Sabir S, Seidahmed MZ, Farooqu HA, **Salih MA**. Brain stem and cerebellar findings in Joubert syndrome. *J Comput Assist Tomogr*. 2006; 30:116-121.

19.2.80 **Salih MA**, Zahraa JN, Al-Jarallah AA, Alorainy IA, Hasan HH. Stroke from systemic vascular disorders in Saudi children. The devastating role of hypernatremic dehydration. *Saudi Med J* 2006;27 Suppl 1:S97-102.

19.2.81 **Salih MA**, Abdel-Gader AG, Al Jarallah AA, Kentab AY, Al Nasser MN. Outcome of stroke in Saudi children. *Saudi Med J*. 2006;27 Suppl 1:S91-96.

19.2.82 **Salih MA**, Abdel-Gader AG, Zahraa JN, Al-Rayess MM, Alorainy IA, Hassan HH, Ruitenbeek W, Zeviani M. Stroke due to mitochondrial disorders in Saudi children. *Saudi Med J*. 2006; 27 Suppl 1:S81-90.

19.2.83 **Salih MA**, Murshid WR, Al-Salman MM, Abdel-Gader AG, Al Jarallah AA, Alorainy IA, Hassan HH, Kentab AY, Van Maldergem L, Othman SA, El-Desouki MI, Elgamal EA. Moyamoya syndrome as a risk factor for stroke in Saudi children. Novel and usual associations. *Saudi Med J*. 2006; 27 Suppl 1:S-69-80.

19.2.84 **Salih MA**, Al Jarallah AS, Abdel-Gader AG, Al-Jarallah AA, Al Saadi MM, Kentab AY, Alorainy IA, Hassan HH. Cardiac diseases as a risk factor for stroke in Saudi children. *Saudi Med J* 2006; 27 Suppl 1:S61-68.

19.2.85 **Salih MA**, Murshid WR, Zahraa JN, Abdel-Gader AG, Al Jarallah AA, Kentab AY, Alorainy IA, Hassan HH, Tjan GT. Congenital and genetic cerebrovascular anomalies as a risk factors for stroke in Saudi children. *Saudi Med J*. 2006; 27 Suppl 1:S53-60.

19.2.86 **Salih MA**, Abdel-Gader AG, Al Jarallah AA, Kentab AY, Gadelrab MO, Alorainay IA, Hassan HH, Zahraa JN. Infectious and inflammatory disorders of the circulatory system as risk factors for stroke in Saudi children. *Saudi Med J* 2006; 27 Suppl 1:S41-52.

19.2.87 **Salih MA**, Abdel-Gader AG. Diagnostic approach and management strategy of childhood stroke. *Saudi Med J*. 2006;27 Suppl 1:S4-11. Review.

19.2.88 **Salih MA**, Abdel-Gader AG, Al Jarallah AA, Kentab AY, Alorainy IA, Hassan HH, Al Nasser MN. Perinatal stroke in Saudi children. Clinical features and risk factors. *Saudi Ned J*. 2006; 27 Suppl 1:S35-40.

19.2.89 **Salih MA**, Abdel-Gader AG, Al Jarallah AA, Kentab AY, Alorainy IA, Hassan HH, Bahakim HM, Kurbaan KM, Zahraa JN, Murshid WR, El-Hazmi MA, Khoja WA. Hematologic risk factors for stroke in Saudi children. *Saudi Med J*. 2006;27 Suppl 1:S21-34.

19.2.90 **Salih MA**, Abdel-Gader AG, Al Jarallah AA, Kentab AY, Alorainy IA, Hassan HH, Bahakim HM, Kurbaan KM, Zahraa JN, Al Nasser MN, Nasir AA, Khoja WA, Kabiraj MM. Stroke in Saudi children. Epidemiology, clinical features and risk factors. *Saudi Med J*. 2006; 27 Suppl 1:S12-20.

19.2.91 **Salih MA**, Abdel-Gader AG, Al-Jarallah AA. Study project on stroke in Saudi children. Conclusions, recommendations and acknowledgements. *Saudi Med J*. 2006; 27 Suppl 1:S108-110.

19.2.92 Leutenegger AL, **Salih MA**, Ibanez P, Mukhtar MM, Lesage S, Arabi A, Lohmann E, Durr A, Ahmed AE, Brice A. Juvenile-onset Parkinsonism as a result of the first mutation in the adenosine triphosphate orientation domain of PINK 1. *Arch Neurol* 2006;1257-1261.

19.2.93 Dubourg O, Azzedine H, Verny C, Durosier G, Biroul N, Gouider R, **Salih MA**, Bouhouche A, Thiam A, Grid D, Mayer M, Ruberg M, Tazir M, Brice A, LeGuern E. Autosomal-recessive forms of demyelinating Charcot-MarieTooth disease. *Neuromolecular Med.* 2006; 8:75-86.

19.2.94 Carmignac V, **Salih MA**, Quijano-Roy S, Marchand S, Al Rayess MM, Mukhtar MM, Urtizberea JA, Labeit S, Guicheney P, Leturcq F, Gautel M, Fardeau M, Campbell K, Richard I, Estoumet B, Ferreiro A. C-terminal titin deletions cause a novel cause a novel early-onset myopathy with fatal cardiomyopathy. *Annals of Neurology* 2007;61:340-351.

19.2.95 Van Reeuwijk J, Grewal PK, **Salih MA**, de Bernabe D B-V, Mc Laughlan JM, Michielse CB, Herrmann R, Hewitt JE, Steinbrecher A, Seidahmed MZ, Shaheed MM, Abomelha A, Brunner HG, van Bokhoven H, Voit H. Intragenic deletion in *LARGE* gene causes Walker-Warburg Syndrome. *Human Genetics* 2007;121:685-690 [Epub 2007 Apr 14].

19.2.96 Gribba M, **Salih M**, Anheim M, Lagier-Tourenne C, H'mida D, Drouot N, Mohamed A, Elmalik S, Kabiraj M, Al-Rayess M, Almubarak M, Betard C, Goebel H, Koenig M. A new form of childhood onset, autosomal recessive spinocerebellar atascia and epilepsy is localized at 16q21-q23. *Brain* 2007;130:1921-1928 [Epub 2007 Apr 30].

19.2.97 Bosley TM, **Salih MA**, Alorainy IA, Oystreck DT, Nester M, Abu-Amero KK, Tischfield M, Engle EC. Clinical characterization of the *HOXA1* syndrome BSAS variant. *Neurology* 2007;69:1245-1253.

19.2.98 Hirano R, Interthal H, Huang C, Nakamura T, Deguchi K, Choi K, Bhattacharjee MB, Arimura K, Umehara F, Izumo S, Northrop JL, **Salih MA**, Inoue K, Armstrong DL, Champoux JJ, Takashima H, Boerkoel CF. Spinocerebellar ataxia with axonal neuropathy: consequence of a Tdp 1 recessive neomorphic mutation. *EMBO J* 2007: 4732-43 [Epub 2007 Oct 18].

19.2.99 **Salih MA**, Mustafa AA. A substance in broad beans (*Vicia faba*) is protective against experimentally induced convulsions in mice. *Epilepsy Behav.* 2008; 12: 25-29 [Epub 2007 Nov 5].

19.2.100 Khan AO, Oystreck DT, Koenig M, **Salih MA**. Ophthalmic features of ataxia telangiectasia-like disorder. *J AAPOS.* 2008; 12:186-9 [Epub 2007 Dec 21].

19.2.101 Kornak U, Reynders E, Dimopoulou A, van Reeuwijk J, Fischer B, Rajab A, Budde B, Nurnberg P, Foulquier F; the ARCL Debre-type Study Group [Dobyns WB, Queihias D, Vilarinho L, Leao-Teles E, Grealley M, Seemanova E, Simandlova M, **Salih M ...**] and 11 other authors. Impaired glycosylation and cutis laxa caused by mutations in the vesicularH(1)-ATPase subunit ATP6VOA2. *Nat Genet.* 2008; 40:32-4 [Epub 2007 Dec 23]

19.2.102 Mihaylova V, Muller JS, Vilchez JJ, **Salih MA**, Kabiraj MM, D'Amico A, Bertini E, Wolfe J, Schreimer F, Kurlmann G, Rasic VM, Siakova D, Colomer J, Herczegfalvi A, Fabriciova K, Weschke B, Scola R, Hoellen A, Schara U, Abicht A, Lochmuller H. Clinical and molecular genetic findings in 22 COLQ-mutant CMS patients. *Brain*. 2008;131:747-59. [Epub 2008 Jan 7].

19.2.103 Bosley TM, Alorainy IA, **Salih MA**, Aldhalaan HM, Abu-Amero KK, Oystreck DT, Tischfield MA, Engle EC, Erickson RP. The clinical spectrum of homozygous HOXA1 mutations. *Am J Med Genet A*. 2008;146:1235-40. (Article featured on the cover of AJMG).

19.2.104 Manzini MC, Gleason D, Chang BS, Hill RS, Barry BJ, Partlow JN, Poduri A, Currier S, Galvin-Parton P, Shapiro LR, Schmidt K, Davis JG, Basel-Vanagaite L, Seidahmed MZ, **Salih MA**, Dobyns WB, Walsh CA. Ethnically diverse causes of Walker-Warburg syndrome (WWS): FCMD mutations are a more common cause of WWS outside of the Middle East. *Hum Mutat*. 2008 Nov;29(11):E231-41.

19.2.105 Khan AO, Oystreck DT, Seidahmed MZ, Aldrees A, Elmalik SA, Alorainy IA, **Salih MA**. Ophthalmic features of joubert syndrome. *Ophthalmology*. 2008 Dec;115(12):2286-9.

19.2. 106 **Salih M A M**, El Khashab H Y, Hamdy H. Hassan H H, Kentab A Y, Sara S. Al Subaei S S, Zeidan R M, Al-Nasser M N, Othman S A. A Study on Herpes Simplex Encephalitis in 18 Children, Including 3 Relapses. *The Open Pediatric Medicine Journal*, 2009, 3, 48-57.

19.2.107 Cazeneuve C, Sâh C, Ibrahim SA, Mukhtar MM, Kheir MM, Leguern E, Brice A, **Salih MA**. A new complex homozygous large rearrangement of the PINK1 gene in a Sudanese family with early onset Parkinson's disease. *Neurogenetics*. 2009 Jul;10(3):265-70.

19.2.108 Gueneau L, Bertrand AT, Jais JP, **Salih MA**, Stojkovic T, Wehnert M, et al. Mutations of the FHL1 Gene Cause Emery-Dreifuss Muscular Dystrophy. *American Journal of Human Genetics*. 2009 Sep;85(3):338-53.

19.2.109 Anheim M, Monga B, Fleury M, Charles P, Barbot C, **Salih M**, et al. Ataxia with oculomotor apraxia type 2: clinical, biological and genotype/phenotype correlation study of a cohort of 90 patients. *Brain*. 2009 Oct;132(Pt 10):2688-98. Epub 2009 Aug 20.

19.2.110 Mihaylova V, **Salih MA**, Mukhtar MM, Abuzeid HA, El-Sadig SM, von der Hagen M, et al. Refinement of the clinical phenotype in musk-related congenital myasthenic syndromes. *Neurology*. 2009 Dec;73(22):1926-8.

- 19.2.111 Laugel V, Dalloz C, Durand M, .... **Salih M**, .... Lehmann AR, Sarasin A, Dollfus H. Mutation update for the CSB/ERCC6 and CSA/ERCC8 genes involved in Cockayne syndrome. *Hum Mutat.* 2010 Feb;31(2):113-26.
- 19.2.112 Abu-Amero KK, Hellani A, **Salih MA**, Hussain AA, Obailan MA, Zidan G, Alorainy IA, and Bosley TM. Ophthalmologic abnormalities in a de novo terminal 6q deletion. *Ophthalmic genetics* 2010 Mar; 31(1):1-11.
- 19.2.113 van der Knaap MS, Lai V, Köhler W, **Salih MA**, Fonseca MJ, Benke TA, et al. Megalencephalic leukoencephalopathy with cysts without MLC1 defect. *Annals of neurology.* 2010 Jun;67(6):834-7.
- 19.2.114 Boyden SE, **Salih MA**, Duncan AR, White AJ, Estrella EA, Burgess SL, Seidahmed MZ, Al-Jarallah AS, Alkhalidi HM, Al-Maneea WM, Bennett RR, Alshemmari SH, Kunkel LM, Kang PB. Efficient identification of novel mutations in patients with limb girdle muscular dystrophy. *Neurogenetics.* 2010 Oct;11(4):449-55. Epub 2010 Jul 13.
- 19.2.115 Walton C, Interthal H, Hirano R, **Salih MA**, Takashima H, Boerkoel CF. Spinocerebellar ataxia with axonal neuropathy. *Adv Exp Med Biol.* 2010;685:75-83.
- 19.2.116 Assoum M, **Salih MA**, Drouot N, H'mida-Ben Brahim D, Lagier-Tourenne C, Aldrees A, Elmalik SA, Ahmed TS, Seidahmed MZ, Kabiraj MM, Koenig M. Rundataxin, a novel protein with RUN and diacylglycerol binding domains, is mutant in a new recessive ataxia. *Brain.* 2010 Aug;133(Pt 8):2439-47.
- 19.2.117 Abu-Amero KK, Hellani A, **Salih MA**, Alorainy IA, Zidan G, Kern KC, Sicotte NL, Bosley TM. Optic disk and white matter abnormalities in a patient with a de novo 18p **partial** monosomy. *Ophthalmic Genet.* 2010 Sep;31(3):147-54.
- 19.2.118 Abu-Amero KK, Hellani AM, **Salih MA**, Seidahmed MZ, Elmalik TS, Zidan G, Bosley TM. A de novo marker chromosome derived from 9p in a patient with 9p partial duplication syndrome and autism features: genotype-phenotype correlation. *BMC Med Genet.* 2010 Sep 21;11:135.
- 19.2.119 Shaheen R, Al-Dirbashi OY, Al-Hassnan ZN, Al-Owain M, Makhssheed N, Basheeri F, Seidahmed M, **Salih MA**, Faqih E, Zaidan H, Al-Sayed M, Rahbeeni Z, Al-Sheddi T, Hashem M, Kurdi W, Shimozawa N, Alkuraya FS. Clinical, biochemical and molecular characterization of peroxisomal diseases in Arabs. *Clin Genet.* 2011 Jan;79(1):60-70. [Epub ahead of print].

- 19.2.120 **Salih MA**, Abu-Amero KK, Alrasheed S, Alorainy IA, Liu L, McGrath JA, Van Maldergem L, Al-Faky YH, AlSuhaibani AH, Oystreck DT, Bosley TM. Molecular and neurological characterizations of three Saudi families with lipoid proteinosis. *BMC Med Genet*. 2011 Feb 24;12:31.
- 19.2.121 **Salih MA**, Oystreck DT, Al-Faky YH, Kabiraj M, Omer MI, Subahi EM, Beeson D, Abu-Amero KK, Bosley TM. Congenital Myasthenic Syndrome Due to Homozygous CHRNE Mutations: Report of Patients in Arabia. *J Neuroophthalmol*. 2011 Mar;31(1):42-47. Epub 2010 Dec 9.
- 19.2.122 Seidahmed MZ, Alkuraya FS, Shaheed M, Al Zahrani M, Al Manea W, Mansour F, Mustafa T, Farid G, **Salih MA**. Ritscher-Schinzel (cranio-cerebello-cardiac, 3C) syndrome: Report of four new cases with renal involvement. *Am J Med Genet A*. 2011 Jun;155A(6):1393-7. Epub 2011 May 12.
- 19.2.123 Adachi Y, Poduri A, Kawaguch A, Yoon G, **Salih MA**, Yamashita F, Walsh CA, Barkovich AJ. Congenital microcephaly with a simplified gyral pattern: Associated findings and their significance. *AJNR Am J Neuroradiol*. 2011 Jun-Jul;32(6):1123-9. Epub 2011 Mar 31.
- 19.2.124 Salih MAM, Satti SA. Editorial: A new horizon for the Sudanese Journal of Paediatrics. *Sudanese Journal of Paediatrics* 2011; 11(1):4-5.
- 19.2.125 Suliman OS, **Salih MA**, Karrar ZA, Mohammed AO, Helsing C. Acute phase reactants in Sudanese children with severe protein-energy malnutrition. *Sudan J Paediatr* 2011;11(1):49-59.
- 19.2.126 Alazami AM, Monies D, Meyer BF, Alzahrani F, Hashem M, **Salih MA**, Alkuraya FS. Congenital disorder of glycosylation IIa: The trouble with diagnosing a dysmorphic inborn error of metabolism. *Am J Med Genet A*. 2011 Nov 21. doi: 10.1002/ajmg.a.34347. [Epub ahead of print].
- 19.2.127 Bosley TM, **Salih MA**, Alorainy IA, Islam MZ, Oystreck DT, Suliman OS, Malki SA, Suhaibani AH, Khiari H, Beckers S, van Wesenbeeck L, Perdu B, Aldrees A, Elmalik SA, Van Hul W, Abu-Amero KK. The neurology of carbonic anhydrase type II deficiency syndrome. *Brain*. 2011 Dec;134(Pt 12):3502-15. Epub 2011 Nov 26.
- 19.2.128 **Salih MAM**, Satti SA. Editorial: Commitment to the wellbeing of children worldwide. *Sudanese Journal of Paediatrics* 2011; 11(2):4-5.
- 19.2.129 Suliman OS, **Salih MA**, Karrar ZA, Mohammed AO, Helsing C. Infection and immunoglobulin levels in Sudanese children with severe protein-energy malnutrition. *Sudan J Paediatr* 2011;11(2):32-42.

- 19.2.130 Oystreck DT, **Salih MA**, Bosley TM. When straight eyes won't move: phenotypic overlap of genetically distinct ocular motility disturbances. *Can J Ophthalmol*. 2011 Dec;46(6):477-80.
- 19.2.131 Di Blasi C, Bellafigliore E, **Salih MA**, Manzini MC, Moore SA, Seidahmed MZ, Mukhtar MM, Karrar ZA, Walsh CA, Campbell KP, Mantegazza R, Morandi L, Mora M. Variable disease severity in Saudi Arabian and Sudanese families with c.3924 + 2 T > C mutation of LAMA2. *BMC Res Notes*. 2011 Dec 13;4(1):534.
- 19.2.132 AL-Otaibi HM, AL-Jurayyan NAM, Mohamed S, **Salih MAM**. Osteomalacia in adolescents presenting as proximal myopathy. *Curr Pediatr Res* 2012; 16 (1): 57-60.
- 19.2.133 Davidson GL, Murphy SM, Polke JM, Laura M, **Salih MA**, Muntoni F, Blake J, Brandner S, Davies N, Horvath R, Price S, Donaghy M, Roberts M, Foulds N, Ramdharry G, Soler D, Lunn MP, Manji H, Davis MB, Houlden H, Reilly MM. Frequency of mutations in the genes associated with hereditary sensory and autonomic neuropathy in a UK cohort. *J Neurol* 2012 Aug;259(8):1673-85. [Epub ahead of print].
- 16.2.134 Shamseldin HE, Alshammari M, Al-Sheddi T, **Salih MA**, Alkhalidi H, Kentab A, Repetto GM, Hashem M, Alkuraya FS. Genomic analysis of mitochondrial diseases in a consanguineous population reveals novel candidate disease genes. *J Med Genet*. 2012 Apr;49(4):234-41.
- 16.2.135 Perrault I, Saunier S, Hanein S, Filhol E, Bizet AA, Collins F, **Salih MA**, Gerber S, Delphin N, Bigot K, Orssaud C, Silva E, Baudouin V, Oud MM, Shannon N, Le Merrer M, Roche O, Pietrement C, Goumid J, Baumann C, Bole-Feysot C, Nitschke P, Zahrate M, Beales P, Arts HH, Munnich A, Kaplan J, Antignac C, Cormier-Daire V, Rozet JM. Mainzer Saldino Syndrome Is a Ciliopathy Caused by IFT140 Mutations. *Am J Hum Genet* 2012 May 4;90(5):864-70.
- 16.2.136 Alsaadi MM, Iqbal SM, Elgamal EA, **Salih MA**, Gozal D. Sleep-disordered breathing in children with craniosynostosis. *Sleep Breath*. 2013 Mar;17(1):389-93.
- 16.2.137 **Salih MA**, Salih MA, Mustafa AA, Oystreck DT, Attia KM, El-Sadig SM, Hamed AA, Hajjar WM, Bosley TM. Ocular Neostigmine Drops for Diagnosing Myasthenia Gravis. *Journal of Neurology & Neurophysiology* 01/2012; S11:1-4. DOI:10.4172/2155-9562.S11-004.
- 16.2.138 Al-Faky YH, Bosley TM, Al-Turki T, **Salih MA**, Abu-Amero KK, Alsuhaibani AH. Prominent corneal nerves: a novel sign of lipid proteinosis. *Br J Ophthalmol* 2012 Jul;96(7):935-40. Epub 2012 May 12.

- 16.2.139 Poduri A, Chopra SS, Neilan EG, Christina Elhosary P, Kurian MA, Meyer E, Barry BJ, Khwaja OS, **Salih MA**, Stöberg T, Scheffer IE, Maher ER, Sahin M, Wu BL, Berry GT, Walsh CA, Picker J, Kothare SV. Homozygous *PLCB1* deletion associated with malignant migrating partial seizures in infancy. *Epilepsia*. 2012 Aug;53(8):e146-e150. doi: 10.1111/j.1528-1167.2012.03538.x. Epub 2012 Jun 12.
- 16.2.140 **Salih MAM**, Satti SA, Swar MO. Editorial: Changing an old therapy of a historic human infection: Malaria. *Sudanease Journal of Paediatrics* 2012; 12(1):4-5.
- 16.2.141 Alazami AM, Alshammari MJ, **Salih MA**, Alzahrani F, Hijazi H, Seidahmed MZ, Safieh LA, Aldosary M, Khan AO, Alkuraya FS. Molecular characterization of Joubert syndrome in Saudi Arabia. *Hum Mutat*. 2012 Oct;33(10):1423-8. [Epub ahead of print].
- 16.2.142 Abu-Amero KK, Kondkar AA, **Salih MA**, Alorainy IA, Khan AO, Oystreck DT, Bosley TM. Partial chromosome 7 duplication with a phenotype mimicking the *HOXA1* spectrum disorder. *Ophthalmic Genet* 2013 Mar-Jun;34(1-2):90-6.
- 16.2.143 Manzini MC, Tambunan DE, Hill RS, Yu TW, Maynard TM, Heinzen EL, Shianna KV, Stevens CR, Partlow JN, Barry BJ, Rodriguez J, Gupta VA, Al-Qudah AK, Eyaid WM, Friedman JM, **Salih MA**, Clark R, Moroni I, Mora M, Beggs AH, Gabriel SB, Walsh CA. Exome sequencing and functional validation in zebrafish identify *GTDC2* mutations as a cause of Walker-Warburg syndrome. *Am J Hum Genet*. 2012 Sep 7;91(3):541-7.
- 16.2.144 Seidahmed MZ, **Salih MA**, Abdulbasit OB, Shaheed M, Al Hussein K, Miqdad AM, Al Rasheed AK, Alazami AM, Alorainy IA, Alkuraya FS. A novel syndrome of lethal familial hyperekplexia associated with brain malformation. *BMC Neurol*. 2012 Oct 27;12:125. doi: 10.1186/1471-2377-12-125. (Highly accessed).
- 16.2.145 Tesson C, Nawara M, **Salih MAM**, Rossignol R, Zaki MS, et al. Alteration of fatty-acid-metabolizing enzymes affects mitochondrial form and function in hereditary spastic paraplegia. *The American Journal of Human Genetics* 2012, Dec 7;91(6):1051-64. doi: 10.1016/j.ajhg.2012.11.001. Epub 2012 Nov 21.
- 16.2.146 **Salih MAM**, Satti SA, Swar MO. Editorial: Managing the common and rare in paediatrics. *Sudanese Journal of Paediatrics* 2012; 12(2):4-6.
- 16.2.147 **Salih MA**, Bosley TM, Alorainy IA, Sabry MA, Rashed MS, Al-Yamani EA, El-Akoum S, Mohamed SH, Abu-Amero KK, Hellani AM. Preimplantation genetic diagnosis in isolated sulfite oxidase deficiency. *Can J Neurol Sci*. 2013 Jan;40(1):109-12.
- 16.2.148 Cossins J, Belaya K, Hicks D, **Salih MA**, Finlayson S, Carboni N, Liu WW, Maxwell S, Zoltowska K, Farsani GT, Laval S, Seidhamed MZ; WGS500 consortium, Donnelly P, Bentley D, McGowan SJ, Müller J, Palace J, Lochmüller H, Beeson D. Congenital myasthenic syndromes due to mutations in *ALG2* and *ALG14*. *Brain*. 2013 Mar;136(Pt 3):944-56.[Epub ahead of print].



- 16.2.149 Abu-Amero KK, Kondkar AA, **Salih MA**, Al-Husain M, Al Shammari M, Zeidan G, Oystreck DT, Hellani AM, Kentab AY, Bosley TM. Ophthalmologic Observations in a Patient with Partial Mosaic Trisomy 8. *Ophthalmic Genet.* 2013 Dec;34(4):249-53.
- 16.2.150 Awad S, Al-Dosari MS, Alyacoub N, Colak D, **Salih MA**, Alkuraya FS, Poizat C. Mutation in *PHC1* Implicates Chromatin Remodeling in Primary Microcephaly Pathogenesis. *Hum Mol Genet* 2013 Jun 1;22(11):2200-13.
- 16.2.151 Stevens E, Carss KJ, Cirak S, Foley AR, Torelli S, Willer T, Tambunan DE, Yau S, Brodd L, Sewry CA, Feng L, Haliloglu G, Orhan D, Dobyns WB, Enns GM, Manning M, Krause A, **Salih MA**, Walsh CA, Hurles M, Campbell KP, Manzini MC; UK10K Consortium, Stemple D, Lin YY, Muntoni F. Mutations in *B3GALNT2* Cause Congenital Muscular Dystrophy and Hypoglycosylation of  $\alpha$ -Dystroglycan. *Am J Hum Genet.* 2013 Mar 7;92(3):354-65.
- 16.2.152 Alazami A, Alshammari M, Baig M, **Salih M**, Hassan H, Alkuraya F. *NPHP4* mutation is linked to cerebello-oculo-renal syndrome and male infertility. *Clin Genet* 85, no. 4 (2014): 371-375.
- 16.2.153 Alazami AM, Hijazi H, Al-Dosari MS, Shaheen R, Hashem A, Aldahmesh MA, Mohamed JY, Kentab A, **Salih MA**, Awaji A, Masoodi TA, Alkuraya FS. Mutation in *ADAT3*, encoding adenosine deaminase acting on transfer RNA, causes intellectual disability and strabismus. *J Med Genet* 2013 Jul;50(7):425-30.
- 16.2.154 Elgamal EA, Almotairi FS, Abdullah AM, **Salih MA**. Cerebrospinal fluid hydrothorax, an unusual complication of ventriculoperitoneal shunt. *Journal of Pediatric Neurology* 2013, 11(2):119-122.
- 16.2.155 **Salih MA**, Tzschach A, Oystreck DT, Hassan HH, AlDrees A, Elmalik SA, El Khashab HY, Wienker TF, Abu-Amero KK, Bosley TM. A newly recognized autosomal recessive syndrome affecting neurologic function and vision. *Am J Med Genet A.* 2013 Jun;161(6):1207-13.
- 16.2.156 Assoum M, **Salih MA**, Drouot N, Hnia K, Martelli A, Koenig M. The Salih Ataxia Mutation Impairs Rubicon Endosomal Localization. *Cerebellum* 2013 Dec;12(6):835-40.
- 16.2.157 Patel N, **Salih M**, Alshammari M, Abdulwahhab F, Adly N, Alzahrani F, Elgamal E, Elkhashab H, Al-Qattan M, Alkuraya F. Expanding the clinical spectrum and allelic heterogeneity in van den Ende-Gupta syndrome. *Clin Genet* 2014 May;85(5):492-4.
- 16.2.158 **Salih MAM**, Satti SA, Swar MO. Editorial: Building civilization starts from childhood. *Sudanese Journal of Paediatrics* 2013; 13(1):6-10.
- 16.2.159 **Salih MAM**. Professor Tigani El Mahi: Short biography and photos. *Sudanese Journal of Paediatrics* 2013; 13(1):75-78.

16.2.160 **Salih MAM.** An elegy on the memory of Professor Tigani El Mahi (Arabic poem). Sudanese Journal of Paediatrics 01/2013; 13(1):119-123.

16.2.161 Gai X, Ghezzi D, Johnson MA, Biagosch CA, Shamseldin HE, Haack TB, Reyes A, Tsukikawa M, Sheldon CA, Srinivasan S, Gorza M, Kremer LS, Wieland T, Strom TM, Polyak E, Place E, Consugar M, Ostrovsky J, Vidoni S, Robinson AJ, Wong LJ, Sondheimer N, **Salih MA**, Al-Jishi E, Raab CP, Bean C, Furlan F, Parini R, Lamperti C, Mayr JA, Konstantopoulou V, Huemer M, Pierce EA, Meitinger T, Freisinger P, Sperl W, Prokisch H, Alkuraya FS, Falk MJ, Zeviani M. Mutations in FBXL4, Encoding a Mitochondrial Protein, Cause Early-Onset Mitochondrial Encephalomyopathy. Am J Hum Genet. 2013 Sep 5;93(3):482-95.

16.2.162 **Salih MA**, Mundwiller E, Khan AO, Aldrees A, Elmalik SA, Hassan HH, Al-Owain M, Alkhalidi HM, Katona I, Kabiraj MM, Chrast R, Kentab AY, Alzaidan H, Rodenburg RJ, Bosley TM, Weis J, Koenig M, Stevanin G, Azzedine H. New Findings in a Global Approach to Dissect the Whole Phenotype of PLA2G6 Gene Mutations. PLoS One. 2013 Oct 9;8(10):e76831. doi: 10.1371/journal.pone.0076831.

16.2.163 Hijazi H, **Salih MA**, Hamad MH, Hassan HH, Salih SB, Mohamed KA, Mukhtar MM, Karrar ZA, Ansari S, Ibrahim N, Alkuraya FS. Pellagra-like condition is xeroderma pigmentosum/Cockayne syndrome complex and niacin confers clinical benefit. Clin Genet. 2015 Jan;87(1):56-61. (2013 Nov 21. doi: 10.1111/cge.12325. [Epub ahead of print]).

16.2.164 **Salih MAM**, Satti SA, Swar MO. Editorial: Biomedical research: Child health and nation's health. Sudanese Journal of Paediatrics 2013; 13(2):6-9.

16.2.165 **Salih MAM.** Remembering for tomorrow: Professor Mansour Ali Haseeb. Sudanese Journal of Paediatrics 2013; 13(2):76-83.

16.2.166 **Salih MAM.** An elegy on the memory of Professor Mansour Ali Haseeb (Arabic poem). Sudanese Journal of Paediatrics 2013; 13(2):129-131.

16.2.167 Kruer MC, **Salih MA**, Mooney C, Alzahrani J, Elmalik SA, Kabiraj MM, Khan AO, Paudel R, Houlden H, Azzedine H, Alkuraya F. C19orf12 mutation leads to a pallido-pyramidal syndrome. Gene. 2014 Mar 10;537(2):352-6.

16.2.168 Poduri A, Heinzen, EL, Chitsazzadeh V, Lasorsa F M, Elhosary P C, LaCoursiere CM, ... , **Salih MA** & Walsh, C. A. (2013). SLC25A22 is a novel gene for migrating partial seizures in infancy. Annals of Neurology, 74(6), 873-882.

16.2.169 Mallaret M, Synofzik M, Lee J, Sagum CA, Mahajnah M, Sharkia R, Drouot N, Renaud M, Klein FA, Anheim M, Tranchant C, Mignot C, Mandel JL, Bedford M, Bauer P, **Salih MA**, Schüle R, Schöls L, Aldaz CM, Koenig M. The tumour suppressor gene WWOX is mutated in autosomal recessive cerebellar ataxia with epilepsy and mental retardation. Brain. 2014 Feb;137(Pt 2):411-9.

- 16.2.170 Bosley TM, Alorainy IA, Oystreck DT, Hellani AM, Seidahmed MZ, Osman Mel F, Sabry MA, Rashed MS, Al-Yamani EA, Abu-Amro KK, **Salih MA**. Neurologic injury in isolated sulfite oxidase deficiency. *Can J Neurol Sci*. 2014 Jan;41(1):42-8.
- 16.2.171 Poduri A, **Salih M**, Walsh CA. Reply: To PMID 24243345. *Ann Neurol* 2014 Feb;75(2):326. 2012. doi: 10.1002/ana.24055. Epub 2014 Jan 2.
- 16.2.172 Al-Faky YH, **Salih MA**, Mubarak M, Al-Rikabi AC. Bilateral Congenital Entropion with Cutis Laxa. *Pediatr Dermatol* 2014 May-Jun;31(3):e82-4.
- 16.2.173 Khan AO, Aldrees A, Elmalik SA, Hassan HH, Koenig M, Stevanin G, Azzedine H, **Salih MA**. Ophthalmic features of PLA2G6-related paediatric neurodegeneration with brain iron accumulation. *Br J Ophthalmol*. 2014 Feb 12. doi: 10.1136/bjophthalmol-2013-304527. [Epub ahead of print].
- 16.2.174 Mohamed S, Osman A, Al Jurayyan NA, Al Nemri A, **Salih MA**. Congenital toxoplasmosis presenting as central diabetes insipidus in an infant: a case report. *BMC Res Notes*. 2014 Mar 28;7:184. doi: 10.1186/1756-0500-7-184.
- 16.2.175 Di Costanzo S, Balasubramanian A, Pond HL, Rozkalne A, Pantaleoni C, Saredi S, Gupta VA, Sunu CM, Yu TW, Kang PB, **Salih MA**, Mora M, Gussoni E, Walsh CA, Manzini MC. POMK mutations disrupt muscle development leading to a spectrum of neuromuscular presentations. *Hum Mol Genet*. 2014 Jun 11. pii: ddu296. [Epub ahead of print].
- 16.2.176 **Salih MAM**, Swar MO. Editorial: Professionalism in medicine and hyposkillia. *Sudanese Journal of Paediatrics* 2014; 14(1):6 - 10.
- 16.2.177 Manzini MC, Xiong L, Shaheen R, Tambunan DE, Di Costanzo S, Mitisalis V, Tischfield DJ, Cinquino A, Ghaziuddin M, Christian M, Jiang Q, Laurent S, Nanjiani ZA, Rasheed S, Hill RS, Lizarraga SB, Gleason D, Sabbagh D, **Salih MA**, Alkuraya FS, Walsh CA. CC2D1A Regulates Human Intellectual and Social Function as well as NF- $\kappa$ B Signaling Homeostasis. *Cell Rep*. 2014 Aug 7;8(3):647-55. doi: 10.1016/j.celrep.2014.06.039. Epub 2014 Jul 24.
- 16.2.178 Al-Qattan SM, Wakil SM, Anazi S, Alazami AM, Patel N, Shaheen R, Shamseldin HE, Hagos ST, AlDossari HM, **Salih MA**, El Khashab HY, Kentab AY, AlNasser MN, Bashiri FA, Kaya N, Hashem MO, Alkuraya FS. The clinical utility of molecular karyotyping for neurocognitive phenotypes in a consanguineous population. *Genet Med*. 2014 Dec 11. doi: 10.1038/gim.2014.184. [Epub ahead of print].
- 16.2.179 **Salih MA**, Murshid WR, Seidahmed MZ. Epidemiology, prenatal management, and prevention of neural tube defects. *Saudi Med J*. 2014 Dec;35(12):S15-28.

- 16.2.180 **Salih MA**. Message from the guest editor. Saudi Med J. 2014 Dec;35(12):S2.
- 16.2.181 Seidahmed MZ, Abdelbasit OB, Shaheed MM, Alhussein KA, Miqdad AM, Khalil MI, Al-Enazy NM, **Salih MA**. Epidemiology of neural tube defects. Saudi Med J. 2014 Dec;35(12):S29-35.
- 16.2.182 **Salih MA**. Neural tube defects. Challenging, yet preventable. Saudi Med J. 2014 Dec;35(12):S3-4.
- 16.2.183 Seidahmed MZ, Abdelbasit OB, Alhussein KA, Miqdad AM, Khalil MI, **Salih MA**. Sirenomelia and severe caudal regression syndrome. Saudi Med J. 2014 Dec;35(12):S36-43.
- 16.2.184 Elgamal EA, Hassan HH, Elwatidy SM, Altwijri I, Alhabib AF, Jamjoom ZB, Murshid WR, **Salih MA**. Split cord malformation associated with spinal open neural tube defect. Saudi Med J. 2014 Dec;35(12):S44-8.
- 16.2.185 Seidahmed MZ, Abdelbasit OB, Shaheed MM, Alhussein KA, Miqdad AM, Samadi AS, Khalil MI, Al-Mardawi E, **Salih MA**. Genetic, chromosomal, and syndromic causes of neural tube defects. Saudi Med J. 2014 Dec;35(12):S49-56.
- 16.2.186 **Salih MA**, Murshid WR, Seidahmed MZ. Classification, clinical features, and genetics of neural tube defects. Saudi Med J. 2014 Dec;35(12):S5-S14.
- 16.2.187 Elgamal EA, Elwatidy SM, Alhabib AF, Jamjoom ZB, Murshid WR, Hassan HH, **Salih MA**. Agenesis of the corpus callosum associated with spinal open neural tube defect. Saudi Med J. 2014 Dec;35(12):S57-63.
- 16.2.188 **Salih MAM**, Swar MO. Remembering the first doctors worldwide who offered their lives fighting Ebola virus epidemic. Sudan J Paediatr 2014; 14(2):6 - 8.
- 16.2.189 **Salih MA**, Murshid MR, Mohamed AG, Lena C Ignacio LC, de Jesus JE, Baabbad R, El Bushra HM. Risk factors for neural tube defects in Riyadh City, Saudi Arabia: Case-control study. Sudan J Paediatr 2014; 14(2):49 - 60.
- 19.2.190 Alazami AM, Patel N, Shamseldin HE, Anazi S, Al-Dosari MS, Alzahrani F, Hijazi H, Alshammari M, Aldahmesh MA, **Salih MA**, et al. Accelerating Novel Candidate Gene Discovery in Neurogenetic Disorders via Whole-Exome Sequencing of Prescreened Multiplex Consanguineous Families. Cell Rep. 2015 Jan 13;10(2):148-61. doi: 10.1016/j.celrep.2014.12.015. Epub 2014 Dec 31.
- 19.2.191 Cecilia M, **Salih MA**, Nguyen K, Mallaret M, Leboucq N, Hassan HH, Drouot N, Labauge P, Koenig M. Cerebral Iron Accumulation Is Not a Major Feature of FA2H/SPG35. Movement Disorders Clinical Practice 2, no. 1 (2015): 56-60. Available from: [https://www.researchgate.net/publication/272569544\\_Cerebral\\_Iron\\_Accumulation\\_Is\\_Not\\_a\\_Major\\_Feature\\_of\\_FA2HSPG35](https://www.researchgate.net/publication/272569544_Cerebral_Iron_Accumulation_Is_Not_a_Major_Feature_of_FA2HSPG35) [accessed Mar 24, 2015].

19.2.192 **Saudi Mendeliome Group**. Comprehensive gene panels provide advantages over clinical exome sequencing for Mendelian diseases. *Genome Biol.* 2015 Jun 26;16(1):134. doi: 10.1186/s13059-015-0693-2.

19.2.193 **Salih MAM**, Swar MO. Paediatric neurology at the confluence of the Blue and White Niles. *Sudan J Paediatr* 2015; 15(1): 6 - 8.

19.2.194 **Salih MAM**. Welcome Speech of the International Child Neurology Association (ICNA) Educational Meeting in Khartoum, Sudan (January 27-31, 2015). *Sudan J Paediatr* 2015; 15(1):79 - 87.

19.2.195 **Salih MA**, Seidahmed MZ, El Khashab HY, Hamad MH, Bosley TM, Burn S, Myers A, Landsverk ML, Crotwell PL, Bilguvar K, Mane S, Kruer MC. Mutation in GM2A Leads to a Progressive Chorea-dementia Syndrome. *Tremor Other Hyperkinet Mov (N Y)*. 2015 Jul 9;5:306. doi: 10.7916/D8D21WQ0. eCollection 2015.

19.2.196 Mohamed S, Hamad MH, Hassan HH, **Salih MA**. Glutaric aciduria type 1 as a cause of dystonic cerebral palsy. *Saudi medical journal*. 2015 Nov;36(11):1354-1357.

19.2.197 Shaheen R, Patel N, Shamseldin H, Alzahrani F, Al-Yamany R, ALMoisheer A, Ewida N, Anazi S, Alnemer M, Elsheikh M, Alfaleh K, Alshammari M, Alhashem A, Alangari AA, **Salih MA**, et al. Accelerating matchmaking of novel dysmorphology syndromes through clinical and genomic characterization of a large cohort. *Genetics in Medicine*. 2015 Dec 3. doi: 10.1038/gim.2015.147. [Epub ahead of print].

19.2.198 **Salih MAM**, Swar MO. Lessons to the World: Sudanese Youth and the Wellbeing of Children. *Sudan J Paediatr* 2015; 15(2): 6 - 8.

19.2.199 Hassan S, Babiker A, Bashiri FA, Hassan H H, El husseini M, **Salih MA**. Sturge-Weber syndrome: Continued vigilance is needed. *Sudan J Paediatr* 2015; 15(2): 63 - 70.

19.2.200 Elsayed LE, Drouet V, Usenko T, Mohammed IN, Hamed AA, Elseed MA, **Salih MA**, Koko ME, Mohamed AY, Siddig RA, Elbashir MI. A novel nonsense mutation in DNAJC6 expands the phenotype of autosomal recessive juvenile-onset Parkinson disease. *Annals of neurology*. 2015 Dec 24. doi: 10.1002/ana.24591. [Epub ahead of print].

19.2.201 Shamseldin HE, Anazi S, Wakil SM, Fageih E, El Khashab HY, **Salih MA**, Al-Qattan MM, Hashem M, Alsedairy H, Alkuraya FS. Novel copy number variants and major limb reduction malformation: Report of three cases. *American Journal of Medical Genetics Part A*. 2016 Jan 8. doi: 10.1002/ajmg.a.37550. [Epub ahead of print].

- 19.2.202 Bosley TM, **Salih MA**, Alkhalidi H, Oystreck DT, El Khashab HY, Kondkar AA, Abu-Amero KK. Duane retraction syndrome in a patient with Duchenne muscular dystrophy. *Ophthalmic Genet*. 2016 Feb 5:1-5. [Epub ahead of print].
- 19.2.203 Anazi S, Shamseldin HE, AlNageb D, Abouelhoda M, Monies D, **Salih MA**, Al-Rubeaan K, Alkuraya FS. A null mutation in TNIK defines a novel locus for intellectual disability. *Hum Genet*. 2016 Apr 22. [Epub ahead of print].
- 19.2.204 Madeo M,..... **Salih MA**, Acevedo-Arozena A, Kruer MC. Loss-of-Function Mutations in FRRS1L Lead to an Epileptic-Dyskinetic Encephalopathy. *Am J Hum Genet*. 2016 Jun 2;98(6):1249-55. doi: 10.1016/j.ajhg.2016.04.008.
- 19.2.205 Seidahmed MZ, **Salih MA**, Abdelbasit OB, Alassiri AH, Hussein KA, Miqdad A, Samadi A, Rasheed AA, Alorainy IA, Shaheen R, Alkuraya FS. Gonadal mosaicism for ACTA1 gene masquerading as autosomal recessive nemaline myopathy *Am J Med Genet A*. 2016 Aug;170(8):2219-21. doi: 10.1002/ajmg.a.37768.
- 19.2.206 Bashiri FA, Al-Rasheed AA, Hassan SM, Hamad MH, El Khashab HY, Kentab AY, AlBadr FB, **Salih MA**. Auto-immune anti-N-methyl-D-aspartate receptor (anti-NMDAR) encephalitis: three case reports. *Paediatr Int Child Health*. 2016 Jun 22:1-5. [Epub ahead of print].
- 19.2.207 Seidahmed MZ, **Salih MA**, Abdulbasit OB, Samadi A, Al Hussien K, Miqdad AM, Biary MS, Alazami AM, Alorainy IA, Kabiraj MM, Shaheen R, Alkuraya FS. Hyperekplexia, microcephaly and simplified gyral pattern caused by novel ASNS mutations, case report. *BMC Neurol*. 2016 Jul 15;16:105. doi: 10.1186/s12883-016-0633-0.
- 19.2.208 Anazi S, Maddirevula S, Fageih E, .... **Salih MA**, Altassan N, Arold ST, Abouelhoda M, Wakil SM, Monies D, Shaheen R, Alkuraya FS. Clinical genomics expands the morbid genome of intellectual disability and offers a high diagnostic yield. *Mol Psychiatry*. 2016 Jul 19. doi: 10.1038/mp.2016.113. [Epub ahead of print].
- 19.2.209 Elsayed LE, Mohammed IN, Hamed AA, .... **Salih MA**, El-Sadig SM, Koko ME, ... Brice A, Ahmed AE, Stevanin G. Hereditary spastic paraplegias: identification of a novel SPG57 variant affecting TFG oligomerization and description of HSP subtypes in Sudan. *Eur J Hum Genet*. 2016 Sep 7. doi: 10.1038/ejhg.2016.108. [Epub ahead of print].
- 19.2.210 **Salih MA**, Swar MO. Another horizon and breakthrough for the Sudanese Journal of Paediatrics. *Sudan J Paediatr*. 2016;16(1):6-8.

19.2.211 Hamad MH, Adeel AA, Alhaboob AA, Ashri AM, **Salih MA**. Acute poisoning in a child following topical treatment of head lice (pediculosis capitis) with an organophosphate pesticide. Sudan J Paediatr. 2016;16(1):63-6.

19.2.212 Reddy HM, Cho KA, Lek M, .... **Salih MA**, Kunkel LM, MacArthur DG, Kang PB. The sensitivity of exome sequencing in identifying pathogenic mutations for LGMD in the United States. J Hum Genet. 2016 Oct 6. doi: 10.1038/jhg.2016.116. [Epub ahead of print].

19.2.213 Moen MN, Fjær R, Hamdani EH, .... **Salih MA**, El Khashab HY, Selmer KK, Chaudhry FA. Pathogenic variants in KCTD7 perturb neuronal K<sup>+</sup> fluxes and glutamine transport. Brain. 2016 Oct 14. pii: aww244. [Epub ahead of print].