



## Mustafa Abdalla M. Salih

### Biography links:

- 1) <http://faculty.ksu.edu.sa/66414/default.aspx>
- 2) <http://fac.ksu.edu.sa/mustafa/cv>
- 3) <http://biography.marquiswhoswho.com/pediatric-neurologist/mustafa-abdalla-mohamedsalih/5059399>
- 4) [https://www.researchgate.net/profile/Mustafa\\_Salih/?ev=hdr\\_xprf](https://www.researchgate.net/profile/Mustafa_Salih/?ev=hdr_xprf)

- Pediatric neurologist Mustafa Abdalla M. Salih was born in Kosti, White Nile Province, Sudan.
- Professor Salih earned an MBBS degree in 1974, an MPCH degree in 1980 (renamed MD in Clinical Pediatrics), a Doctor of Medicine with Distinction in 1982, all from the University of Khartoum.
- He also earned a Doctor of Medical Science in 1990 from Uppsala University in Sweden.
- In 2005, he was elected Fellow to The Royal College of Paediatrics and Child Health (FRCPCH, UK).
- Currently, Prof. Salih serves as Professor of Pediatrics and Consultant Pediatric Neurologist at the College of Medicine, King Saud University, Riyadh, Saudi Arabia.
- He previously served as Lecturer, Associate Professor and Professor of Pediatrics with the Faculty of Medicine at the University of Khartoum in Sudan from 1980 to 1992.
- He has published (as of March 2014) 176 scientific articles in peer-reviewed medical journals, two supplements in the Scandinavian Journal of Infectious Diseases and the Saudi Medical Journal. He also authored two chapters in the textbook, Genetic Disorders Among Arab Populations (two editions: 1997 and 2010), one chapter in the textbook, Diseases of DNA Repair (2010) and seven chapters on pediatric neurology in the Textbook of Clinical Pediatrics (2012).
- He is a former editor of the Sudanese Journal of Paediatrics (1985-91), a former member of the editorial board of the Sudan Medical Journal (1983-86) and Guest Editor to a Saudi Medical Journal Supplement (March 2006). Currently, he is the International Editor of Sudanese Journal of Paediatrics and Sudan Medical Journal. He is also a member of the editorial boards of The Journal of Pediatric Neurology, The Open Pediatric Medicine Journal and The Open Neurology Journal; and Member of the Advisory Board of the Journal of Taibah University Medical Sciences.
- He holds a United States patent on a diagnostic method for congenital muscular dystrophy, and has evaluated new techniques for the rapid diagnosis of bacterial meningitis.

- Prof. Salih was awarded the President of Sudan Prize for Distinguished Students in 1964, 2 awards from the Faculty of Medicine at the University of Khartoum, the Riyadh Neuroscience Award in 1996, the Medal of Excellence by the President of Sudan in 2007, and The Saudi Neurosciences Society Award for Pioneers and Promoters of Neurosciences in Saudi Arabia in 2008, King Saud University Gold Medal in 2010, and King Saud University Life-time Scientific Achievement Award in 2014.
- His biography is listed in numerous International Biographical Centre, American Biographical Institute, and Marquis' Who's Who publications.

## Abstract

# Approach to hereditary ataxia/spastic paraplegia in childhood

Mustafa Abdalla M. Salih

Spinocerebellar ataxias are characterized by disturbances of the body posture and coordination. They may result from one or any combination of dysfunction of the cerebellum and its associated systems, lesions in the spinal cord, or peripheral sensory loss. Other nervous system structures that are usually affected in spinocerebellar ataxia include the basal ganglia and brainstem nuclei.

According to the mode of inheritance and gene in which causative mutations occur or chromosomal locus, spinocerebellar ataxias can be subdivided into autosomal dominant, autosomal recessive, X-linked, and mitochondrial. The underlying genetic defect remains unknown in about 40 % of suspected genetically determined ataxia cases. Nevertheless, the high incidence of consanguineous marriages in North Africa, including Sudan, and the Arabian Peninsula is reflected on the high prevalence of autosomal recessive (AR) disorders, in contrast to the situation in North America and Europe. The current presentation outlines a diagnostic clinical and investigational algorithm for hereditary ataxia/spastic paraplegia in childhood.

Utilizing this algorithm and the power of family-based genetic studies combined with emerging DNA technology, new syndromes and diseases were identified. Those with gene identification included:

1. A new form of childhood-onset, autosomal recessive spinocerebellar ataxia and epilepsy:  
(<http://brain.oxfordjournals.org/cgi/content/full/130/7/1921>)  
(<http://www.ncbi.nlm.nih.gov/pubmed/24369382>).
2. Spinocerebellar ataxia with axonal neuropathy (SCAN1; OMIM 607250;  
<http://www.ncbi.nlm.nih.gov/books/NBK1105/>).
3. Salih ataxia:  
(OMIM #615705, <http://omim.org/entry/615705>)  
(<http://brain.oxfordjournals.org/content/133/8/2439.full.pdf+html>)  
(<http://www.ncbi.nlm.nih.gov/pubmed/23728897>).
4. A new form of childhood-onset autosomal recessive hereditary spastic paraplegia (SPG56) caused by a novel gene (CYP2U1) mutations (OMIM # 615030).  
(<http://omim.org/entry/615030>)  
(<http://www.cell.com/AJHG/abstract/S0002-9297%2812%2900579-4>).

These advances of pediatric neurogenetics helped in refashioning the prognosis and differential diagnosis of these diseases. It also made possible presymptomatic, prenatal, and pre-implantation genetic diagnoses for affected families.

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## Abstract

# Diagnostic approach to the floppy infant syndrome

Mustafa Abdalla M. Salih

The complex of floppiness and hypotonia is a common neurologic symptom in infancy; and the floppy infant syndrome refers to an infant with generalized hypotonia presenting at birth or in early life. The diagnostic work up is often challenging, if a systematic clinical evaluation is not followed.

It has been a standard practice to decide first, whether the hypotonia is a manifestation of a motor unit disease or if it is due to a disorder of the central nervous system (CNS), or another system in the body. The lower motor neuron, which composes the motor unit, has four subunits. These subunits consist of a motor neuron in the brainstem or ventral horn of the spinal cord and its axon, which together with other axons form the peripheral nerve; the neuromuscular junction; and the group of muscle fibers innervated by a single motor neuron.

Disorders of the CNS manifesting as floppy infant syndrome include cerebral dysgenesis/dysplasia, perinatal hypoxic/ischemic insult, kernicterus and intracranial hemorrhage; congenital infections; genetic syndromes and chromosomal abnormalities; and inherited metabolic disorders.

Spinal cord lesions, either congenital or following birth injury, can also manifest as floppy infant syndrome. Disorders of the motor unit associated with the floppy infant syndrome include diseases of the anterior horn cell, nerve, neuromuscular junction and muscle.

The diagnostic approach of the floppy infant syndrome should be guided by the clinical evaluation followed by pertinent neurophysiologic and laboratory tests.

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