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An unusual presentation of Menkes Syndrome in a female infant

Sarah Abdelbar

Cairo University, Egypt

Background:

Menkes Syndrome is rare, estimated to be of prevalence of 1:298000 births. It is an X-linked recessive disorder and thus presents mostly in males with only 18 female case reports.

Objective:

The objective of this clinical case report is to highlight the unusual presentation of Menkes Syndrome in females and to avoid incorrect diagnosis.

Case Presentation:

A 9 months old female infant from Aswan, Egypt, presented to hospital with failure to gain weight. The mother reported long standing history (starting at the age of 2 months) of hair changes, facial swelling, and irritability.

Clinical signs were highly suggestive of Menkes Syndrome including failure to thrive, delayed motor milestones, silver kinky hair, myopathic face with sagging cheeks, dry skin with areas of hypopigmentation and generalized hypotonia.

Further investigations were carried out for Menkes Syndrome. Serum Copper level came back low at 60 microgram/dl (70-150). Also serum Ceruloplasmin was low at 16 mg/dl (30-90). Radiographs showed severe osteopenia. On microscopic examination of scalp hair, abnormal hair shaft appearance was indicated with pili torti.

Oral Copper has no significant role in treatment of Menkes Syndrome. Parenteral Copper in the form of Copper Histidinate gives best results if administered early in life, within the first 28 days.

Biography:

My name is [Sarah Abdelbar](#). I was an IGCSE student. I obtained my MBChB at Faculty of Medicine, Cairo University in 2017 with a grade Excellent with High Honors. I have had 2 clinical electives abroad in Hungary and England before I started my work as a GP at the Egyptian Ministry of Health. I then started working as a resident doctor of Pediatrics and Neonatology, Abu ElReesh Pediatrics Hospital, [Cairo University](#). This is one of the largest Pediatrics Hospitals in the world. This subjected me to various experience, and the extremely high flow of patients to our hospital has made me come across multiple diseases and disorders, the common and the rare of them, and discuss the case of every patient with my staff whom are university professors and consultants of pediatrics. I also gained different practical skills during my work. I passed my Pediatrics MSc exams first part with excellence. I am a member of the [Royal College of Surgeons](#) and the Egyptian Medical Syndicate. I have GMC registration with full license to practice medicine at the UK.

I have had multiple courses over the years including adult resuscitation, immediate life support, first aid, trauma evaluation and management, medical ethics, research basics, soft skills for medical professionals, sudden cardiac arrest and a 5 day summer course at Weill Cornell Medical at which I received a certificate of outstanding achievement for my achievements during the course.

I have got multiple local publications at the Faculty of Medicine official medical magazine. I am also currently awaiting validation of my publication at LancetNeuro. I also have multiple volunteering experience. I joined the medical aid campaigns for the underprivileged, Ramadan food packaging and gave some lessons at an orphanage in Cairo.

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