

GLOBAL WELLNESS AND INTEGRATED HEALTH CONGRESS

MARCH 15-16, 2023 | (Hotel Crowne Plaza Dubai - Deira)



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Myhre syndrome in Jordan

Myhre syndrome is an extremely rare inherited disorder that affects connective tissue.

In theory, it affects males and females in equal numbers. caused by mutations (pathologic variants) in the SMAD4 gene. it has been reported that Myhre syndrome can be transmitted from parent to child. A person with Myhre syndrome would have a 50-50 chance of transmitting this to each child.

Myhre syndrome is a rare, but increasingly diagnosed genetic disorder characterized by short stature, characteristic facial features, a tendency for the buildup of scar tissue (fibrosis) in the skin and internal organs, distinctive heart and aorta problems, mild to moderate intellectual disability, autism/autistic-like behavior, and various bone and joint abnormalities.

Signs and symptoms:

It is important to note that affected individuals may not have all of the symptoms discussed below and that every patient is unique:

1- Growth deficiency: before birth resulting in low birth weight, after birth resulting in short stature and pubertal irregularities (early or delayed puberty)

Affected females may experience absence of menstruation (amenorrhea), irregular menstruation, or early-onset heavy menstruation. Affected males may experience failure of the testes to descend into the scrotum (cryptorchidism). Some have premature puberty.

2- Distinctive facial features:

including eyes which are small and narrow (blepharophimosis, short palpebral fissure), underdeveloped midfacial structures (maxillary hypoplasia), and prominent nose. Prognathism, a shorter than normal groove in the upper lip (philtrum), thin lips, an unusually narrow mouth, and small ears.

3- Fibrosis can occur in the absence of injury (spontaneously) or develop following surgery or trauma. Affected individuals typically have stiff, thickened skin, usually beginning in childhood. Typically, the skin changes first appear on the palms of the hands, the soles of the feet, the back of the elbows, and the front of the knees. Eventually the skin thickens on other parts of the body. As a result of the thicker skin, affected individuals typically have fewer facial creases (wrinkles) than others of their age. Scars may be more noticeable or become unusually thickened after healing (keloids or hypertrophic scars).

4- airway can be affected at various levels such as narrowing or blockage of the nasal airway by tissue (choanal atresia), leading to stridor, croup.

5- Most individuals with Myhre syndrome develop hearing loss. In some, it is due to the accumulation of wax-like material or from recurrent ear infections.

6- Developmental delays occur in all people with Myhre syndrome.

Mild to moderate intellectual disability occurs in most people. Behavioral abnormalities including difficulties with social interactions, poor communication skills, hyperactivity, stubbornness, and/or repetitive behaviors are common. Many patients are considered to be on the autism spectrum.

Several skeletal abnormalities; abnormally thick skull bones, broad and wide ribs, brachydactyly ,hypo plastic iliac wings

7- Most people with Myhre syndrome have a least one problem involving the heart or aorta. Here we present this case to highlight about this rare syndrome and the proper follow-up for this syndrome.

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Biography:

Hajar Abu-eleinin, dermatology consultant with years of experience diagnosing and treating a variety of skin conditions. Has completed Bachelor's Degree - Medicine & Surgery from University of Aleppo, Syria and achieved the Jordanian Board exams in skin diseases March 2002.

She is a dermatology consultant at al Mouwasat hospital in Dammam, Saudi Arabia. She served as a dermatologist and a dermatology consultant at several hospitals and health centers of the ministry of health in Jordan.

Moreover, certified Member at The Scientific Committee of the Jordanian Medical Council, ex member of biological pharmaceutical committee and clinical Instructor for The Hashemite University medical students.

She had been selected to write the Jordanian Board exams in skin diseases by the Scientific Committee of the Jordanian Medical Council.

Furthermore, she did both in-service(hold the position of Section Head/Dermatology Consultant at Prince Hamza Hospital(MOH)) as well as she was responsible for instructing medical students and teaching Dermatology residents.

Also, Invited speaker of different medical and dermatological conferences.