Neonatal Progeroid Syndrome

Alternative Names
Wiedemann-Rautenstrauch Syndrome
Neonatal Pseudo-Hydrocephalic Progeroid Syndrome of Wiedemann-Rautenstrauch
Rautenstrauch-Wiedemann Type Neonatal Progeria
Rautenstrauch-Wiedemann Syndrome

WHO International Classification of Diseases
Congenital malformations, deformations and chromosomal abnormalities

OMIM Number
264090

Mode of Inheritance
Autosomal recessive

Description
Neonatal progeroid syndrome is an extremely rare autosomal recessive disorder that is characterized by progeroid features from birth with multiple somatic anomalies and paucity of subcutaneous fat, causing the skin to appear abnormally thin, fragile, and wrinkled. In addition, affected individuals also have distinctive craniofacial malformations including frontal bossing and parietal bossing, causing pseudohydrocephalus; hypoplastic bones of the face and abnormally small facial features; a small "beak-shaped" nose that becomes more pronounced with advancing age; and sparse scalp hair, eyebrows, and eyelashes.

Epidemiology in the Arab World

Lebanon
[See also: Palestine > Megarbane and Loiselet, 1997].

Palestine
Megarbane and Loiselet (1997) reported a Palestinian girl in Lebanon with a progeroid appearance, with prominent occiput, blepharophimosis, cataract, arthrogryposis of the upper limbs, and severe pulmonary stenosis. The clinical manifestation of the girl was compared with other four progeroid syndromes. All are characterized by early aging craniofacial appearance, but the case of that girl suggested a new type of Neonatal Progeroid Syndrome. She died aged less than six months. An older sister was born three years before with the same appearance and underwent the same fatal evolution. Their parents were first cousins living in a poor society. The consanguinity of the parents and the appearance of two affected girls point to an autosomal recessive inheritance. However, autosomal dominant or X-linked dominant inheritance is still possible.

References

Contributors
Abeer Fareed: 20.5.2006
Ghazi O. Tadmouri: 8.5.2005