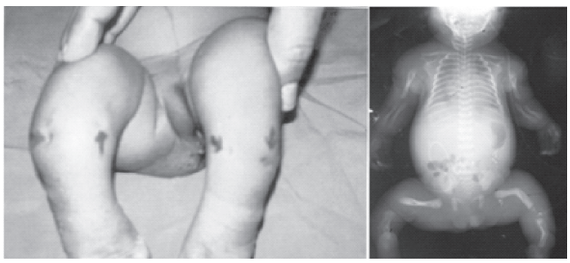
SPOT DIAGNOSIS (IMAGE GALLERY)



A rare skeletal dysplasia

Prashant Patil, Pallavi Saple
Department of Pediatrics, Cama and Albless Hospital andGrant Medical College, Mumbai.

Address for Correspondence: Dr Prashant Patil, NICU, Department of Pediatrics, Cama and Albless Hospital, Mumbai -400001. Email: drprashant1981@gmail.com

A term female neonate was delivered by cesarean section with birth weight of 2.6 kg with APGAR at 1min 3 and at 5min 7. Examination revealed short bowed legs, bell shaped chest, soft cleft palate, distinctive flattened facies, bilateral talipus equinovarus, pretibial pits and multiple café au lait spots and hepatosplenomegaly. Infantogram showed telephone handle appearance of long bones, flat vertebral bodies. Abdominal ultrasound showed hepatosplenomegaly. Karyotyping revealed 46XX. Patient died on day 6 of respiratory insufficiency.

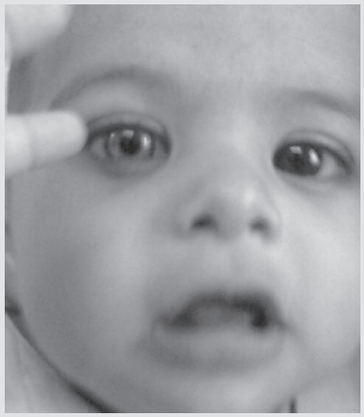
What is the diagnosis?

Campomelic dysplasia. It is a rare autosomal dominant congenital short limbed dwarfism characterized by congenital bowing of long bones associated with skeletal and extra skeletal features like pretibial pits, hypoplastic lungs, malformations of cervical spine, heart, and kidneys. Characteristic feature is male to female sex reversal. It is caused by mutations in the SRY related gene SOX9. It has almost fatal outcome.

Funding: - none

Contribution of each author: Both authors were involved in diagnosis and management of patient. PP was involved in preparation of manuscript. PS will act as guarantor of the paper.

E-published: January 2010



EYE DISEASE

N D Vaswani
Department of Pediatrics, PGIMS, Rohtak, India

Address for Correspondence: N D Vaswani, 55, 9J Medical Campus, P.G.I.M.S, ROHTAK, India. Email: dr_vaswani@yahoo.co.in

Question: Spot Diagnosis

Answer: Heterochromia {also known as heterochromia iridis} is a condition of eye in which color of one iris is different from other {complete heterochromia}, or where part of one iris has a different color from the remainder {partial heterochromia or sectoral heterochromia}. The color of the irises is determined primarily by the concentration and distribution of melanin pigment within the iris tissues.{1} It is a result of the relative excess or lack of pigment within an iris or part of an iris, which may be genetically inherited or due to mosaicism, or acquired by disease or injury.{2} The affected eye may be hyperpigmented {hyperchromic} or hypopigmented {hypochromic}.{3} Partial or sectoral heterochromia is much less common than complete heterochromia and is typically found in autosomally inherited disorders such as Hirschsprung`s disease and Waardenburg syndrome. Congenital heterochromia is inherited as an autosomal dominant. Abnormal darker iris is seen in Lisch nodules, Ocular melanosis, Pigment dispersion syndrome , Sturge-Weber syndrome. Abnormal lighter iris is seen in Fuchs` heterochromic iridocyclitis, Acquired Horner`s syndrome, neoplasm. Acquired Heterochromia is acquired usually due to injury, inflammation, the use of certain eyedrops, or tumors. {4} We are reporting a 9 month old boy who was having simple heterochromia.

References

1. Wielgus AR, Sarna T. Melanin in human irides of different color and age of donors. Pigment Cell Res. 2005; 18: 454-464
2. Imesch PD, Wallow IH, Albert DM. The color of the human eye: a review of morphologic correlates and of some conditions that affect iridial pigmentation. Surv Ophthalmol. 1997; 41 Suppl 2: S117-123
3. Loewenstein J, Scott L. Ophthalmology: Just the Facts. New York: McGraw-Hill. 2004.
4. Van Emelen C, Goethals M, Dralands L, Casteels I. Treatment of glaucoma in children with Sturge-Weber syndrome. J Pediatr Ophthalmol Strabismus. 2000; 37: 29-34

E-published: March 2010.