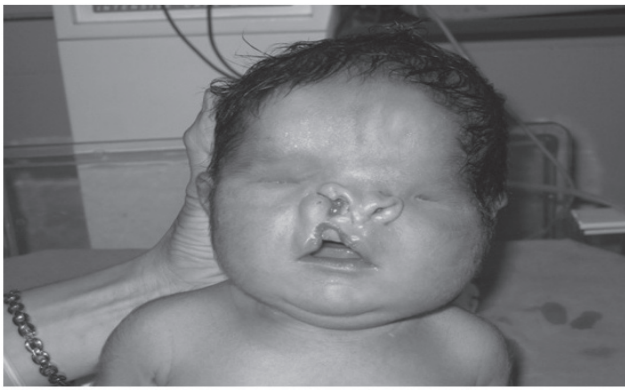
SPOT DIAGNOSIS (IMAGE GALLERY)



FACIAL ABNORMALITY

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A 22 years old primigravida mother delivered a male term baby with normal Apgar score. Birth weight was 2.9 kg. There is no history of teratogen exposure. Soon after birth he developed respiratory distress. Detailed examination revealed wide anterior fontanel, hypertelorism, depressed nasal bridge, midline clefting of nose, bilateral choanal atresia, right cleft lip, retrognathia and single umbilical artery. Ophthalmological examination suggested bilateral severe blephrophimosis, anopthalmos and hypertelorism. X ray skull showed aplastic left frontal bone. Transorbital sonography suggested empty orbital fossa and transfontanelle sonography showed semilobar holoprosencephaly with left frontal encephalocele. Echocardiography was normal. Other systems were normal.

What is the diagnosis?

Median cleft face syndrome. It was described by De Myer. (1) It is an unknown primary defect in mid facial development with incomplete anterior appositional alignment of eyes. (2) This rare disorder is characterized by abnormalities affecting mid-face that includes lower portion of the forehead in the midline, nose, tissue between orbits, and region of upper lip. Most common findings are hypertelorism, notched to completely divided nasal tip, low set ears and defect in midline frontal bone (cranium bifidum occultum). (3) Less common abnormalities include accessory nasal tags, anomalies of optic disc, optic nerve, retina, or eye (colobomas, cataracts), preauricular tags, conductive deafness, frontal cutaneous lipoma or lipoma of corpus callosum, agenesis of corpus callosum, anterior basal encephalocele and Tetralogy of Fallot. (2) Inheritance is sporadic although there are rare reports of familial recurrence. (2) However in families with an affected child, generally malformation tend to occur a little more frequently. (4) Development of the midline facial structures is closely associated with the development of forebrain. (5) The outcome depends on the severity of the defect and neurologic involvement. Most children are of normal intelligence and life span. Risk of severe mental retardation is 8 percent. (2) The risk of neurologic impairment is higher in the presence of a frontal anterior cephalocele. Prenatal diagnosis is difficult and majority of cases are not diagnosed until birth. Major cosmetic surgery is usually needed. Psychosocial therapy will help the affected person adjust to their appearance. (6)

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