SPOT DIAGNOSIS (IMAGE GALLERY)

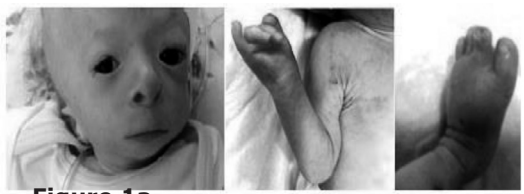

Figure 1a.

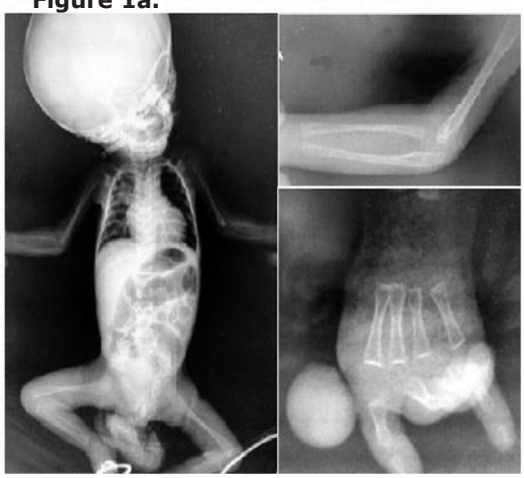

Figure 1b.

WHAT IS YOUR DIAGNOSIS?

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A twenty-month-old boy with weight of 1.9 kg (<3% percentile) and length of 45 cm (< 3% percentile) was brought to our pediatric emergency room with dyspnea, tachypnea, and hypoxemia. He had hyponatremia (110 mEq/L), hypochloremia (79 mEq/L), hypocalcemia (total 7.2 mg/dL,ionised calcium 3.8 mg/dL), hypokalemia (3.5 mEq/L), elevated transaminases (AST 152 U/L, ALT 54 U/L), high CRP (16.2 mg/L) and high creatinine (0.54 mg/dL). Venous blood gas was normal. He had multiple congenital anomalies including cleft palate, congenital bone defects (radial ray anomalies including right absent thumb and left partially formed thumb without bone formation, syndactyly in the left foot, thin extremities, bilaterally pes equinovarus, short stature, saddle nose, and frontal bossing), low-set ears, micrognathia, truncal hypotonia, macrocephalus with hydrocephalus, hypotrichosis including scarce eyelashes, eyebrows, and hairs, hypodontia, erythematous skin changes and telangiectasias on extremities (Figure 1a and 1b), thin and dry skin, micropenis and small testes. He was second born out of a consanguineous marriage (second degree cousins), and was born by emergency cesarean section at 35 weeks of gestation. Birth weight was 1300 gram. He was operated for anal atresia at 13 day of life. He had recurrent vomiting and diarrhea without a specific diagnosis to date. His sister who was 4.5 years old had the same congenital bone defects plus dermal findings including hyperpigmentation and multiple telangiectasias on her face and extremities.

Figure 1a. Hypotrichosis, saddle nose, low-set ears, micrognathia, absent right thumb, syndactyly, erythematous skin changes, atrophy and telangiectasias.

Figure 1b. Radiological findings showing absence of epiphyses, malformed radius, short metacarpal and phalangeal bones, and absence of first metacarpus and phalanx.

What is the diagnosis?

Rothmund–Thomson syndrome (RTS) or poikiloderma congenitale. It is a rare congenital autosomal recessive disorder that is attributed to mutations of the RECQL4 helicase gene on 8q24. (1) Poikilodermatous skin changes and photosensitivity, skeletal, dental and nail abnormalities, juvenile cataracts, sparse hair, eyelashes, and, or eyebrows, small stature and predisposition to skin cancer and osteosarcoma are some key features of this syndrome. (2,3) Other features in individual cases include gastrointestinal problems in infancy, such as chronic diarrhea and vomiting; hematologic abnormalities including isolated anemia and neutropenia, aplastic anemia, myelodysplasia and leukemia; immune dysfunction; premature aging; sexual abnormalities; cleft palate; micrognathia; anal atresia and sensorineural deafness. (4) RTS has been described in all races and many nationalities and there was no clear gender predilection. (5) No population appears to be at higher or lower risk for the disorder and there are approximately 300 cases described in the literature. (5) The most consistent feature of the syndrome is skin findings. The cheeks are usually first involved with red patches or edematous plaques, sometimes with blistering. Over months to years, the rash enters a chronic stage characterized by atrophy, telangiectasias, pigmentary changes (poikiloderma) and spread to other areas of the face, the extremities, and the buttocks. Skeletal abnormalities of this syndrome are short stature, radial ray anomalies such as absent thumbs, ulnar defects, absent or hypoplastic patella and osteopenia. (5) RTS should be kept in mind with patients who have a small stature and skeletal-dental abnormalities with poikilodermatous skin changes.

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