

Photo Quiz

J Ped. Nephrology 2016;4(1):49-50
<http://journals.sbmu.ac.ir/jpn>

A 3.5 year-old Boy with FTT and Skin Lesions

A 3.5- year old boy presented with failure to thrive. He had a history of congenital hypothyroidism that was under therapy with levothyroxine. There was absent metacarpal of left thumb that was reconstructed by Oscuneiform bone graft (Fig.1) A café-au-lait spot was detect on suprapubic region. The initial lab test revealed mild thrombocytopenia. eGFR was 75 ml/min/1.73m². Renal ultrasound showed that both kidneys were smaller than normal for age with mild hydronephrosis in left pelvic kidney. Tc ^{99m} -DMSA renal scintigraphy confirmed left ectopic kidney (Fig.2)

What is your diagnosis?

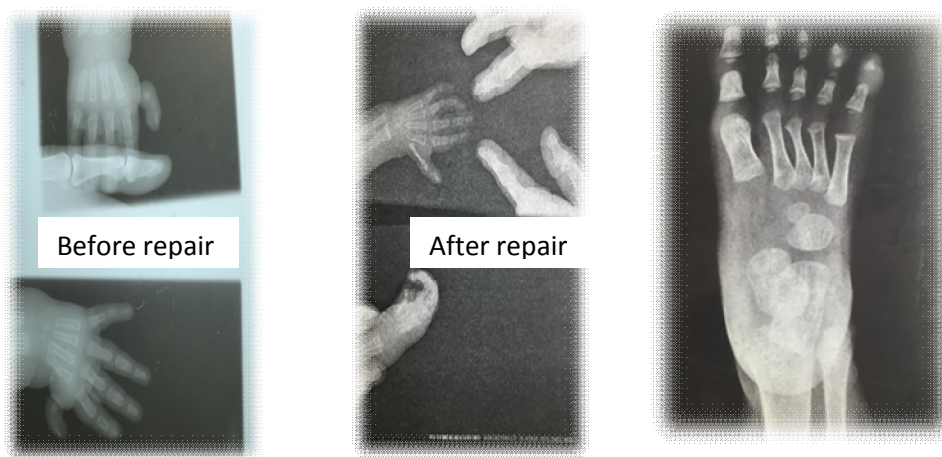


Figure 1. The hand before and after repair

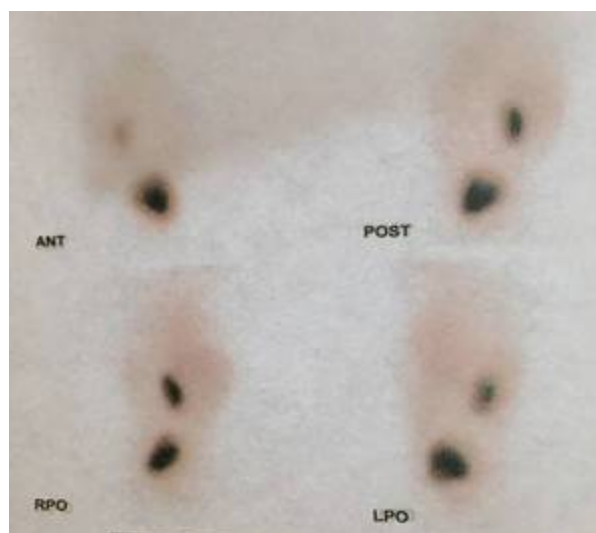


Figure 2. DMSA scan of the patient

Photo Quiz Answer

A 3.5 year-old Boy with FTT and Skin Lesions

Marzieh Dehgan, Nakysa Hooman*

Iran University of Medical Sciences, Tehran, Iran

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*Corresponding Author

Nakysa Hooman, MD.

Professor, Consultant Pediatric Nephrology,

Ali-asghar children hospital,

Iran University of Medical Sciences

Email:hooman.n@iums.ac.ir



Figure 3. The chromosomal study showed 46, XY, with chromosomal breaks compatible with Fanconi anemia

Fanconi anemia is a rare disease associated with hematologic abnormality and congenital anomalies. The majority of patients with Fanconi anemia have birth defects, such as altered skin pigmentation, short stature, thumb or thumb and radial anomalies, abnormal male gonads, microcephaly, eye anomalies, structural renal defects, abnormal ears or hearing, or developmental delay. Asymptomatic siblings need to be screened for occult Fanconi anemia. Pancytopenia can cure by stem cell transplantation. Treatment is recommended for significant cytopenias, such as hemoglobin less than 8 g/dL, platelets fewer than 30,000/ μ L, or neutrophils fewer than 500/ μ L [1].

References

1. Lipton JM, Max J Coppes MJ, Alter BP, et al. Fanconi Anemia. <http://emedicine.medscape.com/article/960401>

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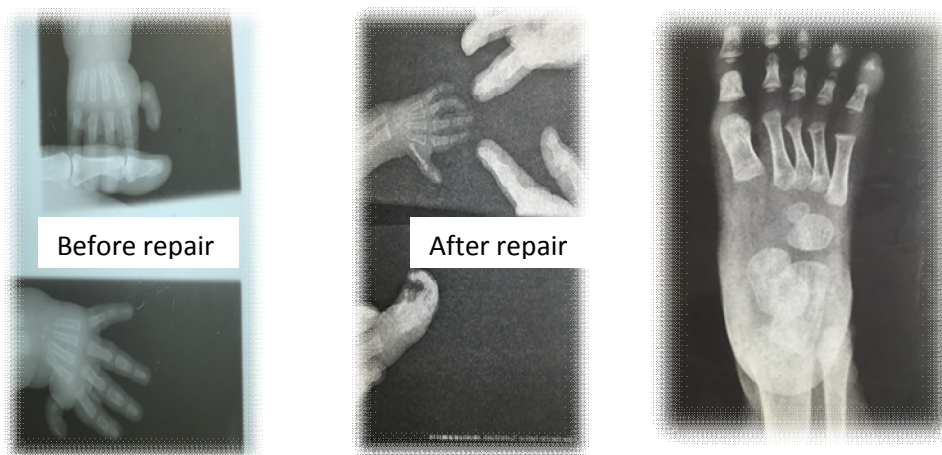


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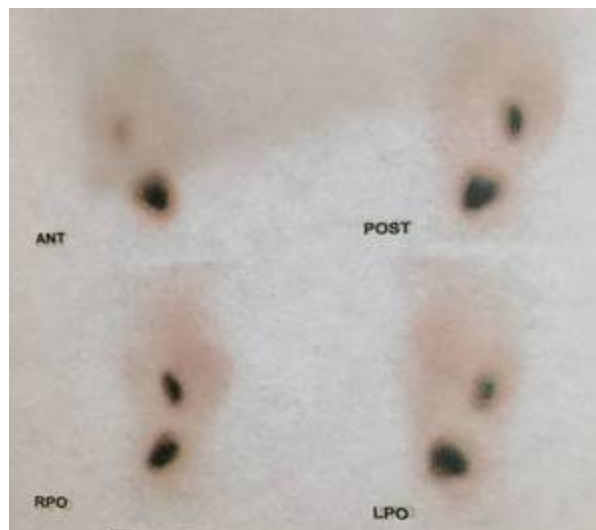


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