

# Multiple subcutaneous folds in oculocerebrorenal syndrome of Lowe

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## Introduction

Oculocerebrorenal syndrome of Lowe is a very rare, X-linked disease with estimated prevalence of 1:500,000. It is characterised by congenital cataracts, mental retardation, and renal Fanconi syndrome. Reports of associated dermatological manifestations are very few and do not include unusual multiple subcutaneous folds.

## Case report

A 3-months old infant born to non-consanguineous parents was evaluated for hypotonia and developmental delay. He had a cataract in left eye, congenital glaucoma and megalocornea in right eye with no other dysmorphism. Anterior fontanelle was widely open. He was hypotonic with diminished tendon reflexes. Hypermobility was noted around both elbows and knees. There were unusual multiple skin folds with generalised increase in subcutaneous tissue noted in the limbs (Figure 1). His weight was between mean and -1SD.

Investigations revealed serum sodium 134 mmol/l, potassium 3.8 mmol/l and chloride 101 mmol/l. Serum bicarbonate was 20.8 mmol/l and blood pH was 7.4. Serum alkaline phosphatase was 1864 U/l (normal < 480). Serum inorganic phosphorus was 0.91 mmol/l (1.30 - 1.75 mmol/l) and serum calcium was 2.2 mmol/l (2.1-2.6 mmol/l). Blood urea and serum creatinine were normal. Urine was positive for glucose and proteins. X ray of wrist showed features of active rickets. Ultrasound scans of brain and kidneys and 2D echocardiogram were normal. Deep skin biopsy showed exocytosis of lymphocytes into the epidermis focally. Dermis was unremarkable and there was no evidence of cyst formation.

Evidence of ocular, central nervous system (CNS) and proximal renal tubular involvement confirmed the clinical diagnosis of oculocerebrorenal syndrome of Lowe. He was commenced on phosphate buffer, calcitriol and sodium bicarbonate.



Figure 1. Photograph (taken at the age of 6 months) showing multiple skin folds and increase in subcutaneous fat.

## Discussion

Lowe syndrome is a very rare disease in childhood, characterised by eye, renal and CNS involvement. Causative gene, oculocerebrorenal-Lowe 1 (OCRL 1) is mapped to Xp24-26 [1]. Skin manifestations are part of this protean syndrome but remain significantly under-reported. Those reported are mainly related to cyst formation and include eruptive vellus hair cysts, multiple eruption cysts in oral cavity and benign cystic skin tumour

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known as tricoepithiloma [2-4]. The pathophysiology of this cyst formation is suggested to be due to a localised reaction intended to wall off excess extracellular lysosomal enzymes found in tissues in these patients. The dermatological manifestation in our baby was presence of excess skin folds and is distinct from those previously reported.

## References

1. Tricot L, Yahiaoui Y, Teixeira L, *et al.* End-stage renal failure

in Lowe syndrome. *Nephrology, Dialysis, Transplantation* 2003; **18**: 1923-5.

2. Nandedkar MA, Minus H. Eruptive vellus hair cysts in a patient with Lowe syndrome. *Pediatric Dermatology* 2004; **21**: 54-7.

3. Batirbaygil Y, Turgut M. Lowe syndrome: case report. *The Journal of Clinical Pediatric Dentistry* 1999; **23**: 357-9.

4. Erdogan F, Ismailogullari S, Soyuer I, *et al.* Different seizure types and skin lesions in oculocerebrorenal syndrome of Lowe. *Journal of Child Neurology* 2007; **22**: 427-31.

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## Double inferior vena cava: beware!

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### Introduction

Double inferior vena cava (DIVC) is a congenital anomaly arising due to abnormal development of embryonic venous channels. It is rare with an incidence of 0.2% to 3% percent [1,2]. Its non recognition may result in diagnostic and surgical errors. Modern imaging with CT and MR has simplified its detection [3]. We report one such case to illustrate how it can be useful to know of its presence.

### Case report

A 27-year old male was found to have infrarenal DIVC during evaluation for living kidney donation. The IVC was seen on either side of the abdominal aorta. He had normal IVC on right side and an abnormal IVC on the left side of aorta. The left renal vein was draining into left IVC (Figures 1 and 2). Left kidney was harvested. Patient

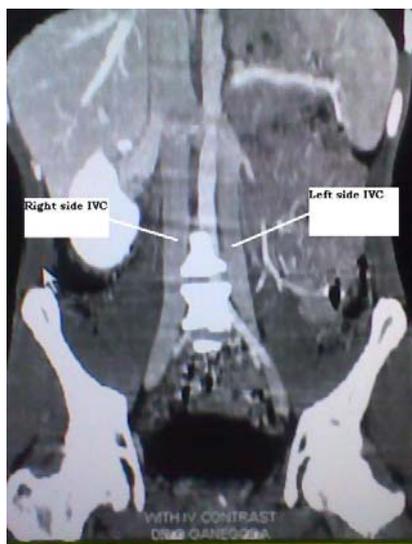


Figure 1.

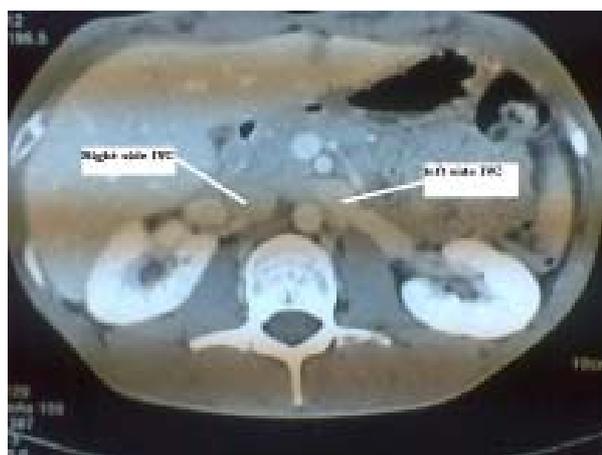


Figure 2.

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