

## Recurrent UTI – Make the Child Smile!

An 8-year-old girl presented with history of recurrent urinary tract infection (UTI) along with episodes of urinary and fecal incontinence. Investigations revealed elevated creatinine (1.2 mg/dL, estimated glomerular-filtration rate (eGFR) = 42 mL/min/1.73 m<sup>2</sup>), bilateral hydronephrotic scarred kidneys with grade IV dilating vesico-ureteric reflux (VUR), and thickened urinary bladder wall. Urodynamic study confirmed a low capacity, high pressure urinary bladder with detrusor over activity. Neurological examination and magnetic resonance imaging of spine was un-remarkable. The diagnosis was clinched on seeing her typical facial expression on being asked to smile (**Fig. 1** and **Web Video 1**).

Ochoa syndrome or Urofacial syndrome (UFS) is characterized by urinary bladder or/and bowel dysfunction along with a characteristic facial expression that is most obvious during smiling or laughing wherein one gets an appearance of a 'grimace' despite an attempt at smiling (resulting from abnormal co-contraction of the corners of the mouth and eyes). It is inherited as autosomal recessive disorder with abnormalities in either of two genes – *HPSE2* localized on chromosome 10q23-10q24 or *LRIG* localized on chromosome 1p13. A heterozygous nonsense variation in exon 4 of the *LRIG2* gene (chr1:113636129; C>C/G; Depth: 121x) that results in a stop codon and premature truncation of the protein at codon 153 (p.Ser153Ter; ENST00000361127) was detected in the child by Next Generation Sequencing.



**FIG. 1** Characteristic facial expression on asking to smile (See video at website).

Apart from early identification and prompt treatment of urinary tract infection, the cornerstone of management of Ochoa syndrome includes reducing the bladder pressure and ensuring proper bladder drainage. Anticholinergics and α-adrenergic blockers along with clean intermittent catheterization are usually the initial steps. Bladder augmentation (augmentation cystoplasty) along with Mitrofanoff (bladder drainage conduit) is often chosen as a long-term management plan.

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