Urofacial Syndrome

Alternative Names
UFS
Ochoa Syndrome
Hydronephrosis with Peculiar Facial Expression
Inverted Smile and Occult Neuropathic Bladder
Facial Palsy, Partial, with Urinary Abnormalities

WHO International Classification of Diseases
Congenital malformations, deformations and chromosomal abnormalities

OMIM Number
236730

Mode of Inheritance
Autosomal recessive

Gene Map Locus
10q23-q24

Description
The urofacial syndrome, also known as the Ochoa syndrome, is a rare autosomal recessive disorder comprising a congenital obstructive uropathy and abnormal facial expression. Obstructive uropathy originates from impaired neural communication between the bladder and the spinal cord, resulting in incomplete emptying of the bladder. Neurogenic bladder causes involuntary discharge of urine, urinary tract infections, and abnormal accumulation of urine in the kidneys. When affected patients smile, their facial musculature turns upside down or ‘‘inverts’’ so that they appear to be grimacing or crying. Some severely affected patients become hypertensive and progress to end-stage renal disease; others become uremic. In addition to facial and urinary abnormalities, about two-thirds of the patients also have moderate to severe constipation.

Molecular Genetics
Genome-wide scan located the disease gene within a genomic interval of approximately 1 cM (between D10S1433 and D10S603) on chromosome 10q23–q24 using homozygosity mapping and linkage disequilibrium analysis. Physical and genetic mapping studies further refined the disease gene to a region between the markers D10S110 and D10S2494, which are located on one YAC clone of approximately 1410 kb.

Epidemiology in the Arab World

Kuwait
Teebi et al. (1989) described an Arab child of consanguineous parents with the characteristic 'inverted' facial expression, i.e., when smiling or laughing, he appeared to be crying. Investigations at age 6 showed minor changes, indicating the probable importance of detecting the abnormality through the facial expression at an early age. In 1991, Teebi and Hassoon indicated that at age 8 the child still showed the inverted facial expression upon laughing, but also had renal changes as a consequence of a neurogenic bladder in addition to hydrocephalus due to stenosis of the aqueduct of Sylvius. Teebi and Hassoon (1991) suggested that the association is not fortuitous and probably widens the spectrum of urofacial syndrome or represents a distinct entity mimicking the urofacial syndrome.

References
Teebi AS, Farag TI, el-Khalifa MY, Besisso MS, al-Ansari AG. Urofacial syndrome. Am J Med Genet. 1989; 34(4):608.
Teebi AS, Hassoon MM. Urofacial syndrome associated with hydrocephalus due to aqueductal stenosis. Am J Med Genet. 1991; 40(2):199-200.

Contributors
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