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ASSOCIATION BETWEEN DELETION 18Q AND TERMINAL DUPLICATION 1P IN PATIENT WITH BILATERAL VESICO-URETERAL REFLUX: CASE REPORT AND LITERATURE REVISION

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Introduction
Vesico-ureteral reflux (VUR) is a dynamic event in which is present a retrograde flow of urine into the upper tracts. VUR may occur isolated or in association with other congenital abnormalities or as part of syndromic entities.
We present a patient with a bilateral primary VUR, plurisyndromic disease caused by a large deletion of 18q (18q21.3-qter) and terminal duplication of 1p (1p36.32-p36.33).

Case Report
The patient was 8 years old female with a plurisyndromic disease including moderate growth retardation, psychomotor retardation, facial dysmorphism, single umbelical artery, umbelical hernia, urachal residue, bilateral congenital clubfeet and renal-urinary disease.
Chromosomal analysis and Array-CGH revealed two heterozigous chromosomal rearrangements: 1p terminal duplication and de novo 18q terminal deletion.
She referred to our clinic to further evaluation of hidronephrosis and right hypoplastic kidney which had been discovered after renal ultrasonography execution.
Voiding cystourethrography (VCUG) demonstrated bilateral grade IV VUR.
The patient underwent ureterovescical reimplantation after failure of 3 endoscopic submeatal dextranomer hyaluronic acid copolymer (Deflux®) injection with VUR resolution.

Discussion
Several studies were undertaken in order to explore genes involved in VUR.
There is not reported cases in the literature regarding a correlation between VUR and 18q21.3-qter deletion syndrome or 1p36.32-p36.33 chromosomal duplication.
Also this is the first report involving a patient with deletion of chromosome 18q and contemporary presence of 1p chromosomal terminal duplication.
It is appropriate others studies to recognized chromosomal abnormality correlated with VUR.

Key words: Vesico-ureteral reflux; deletion of 18q; duplication of 1p.

RECTAL DUPLICATION CYST IN PREVIOUS ANORECTAL MALFORMATION AND DOWN SYNDROME

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Introduction
Gastrointestinal (GI) tract duplications are rare congenital malformations. Most of them occur in the ileum and only 1-5%, of all duplication, were in the rectum. Different clinical features including chronic constipation, rectal prolapsed or polips. We report on a 4-years-old girl with Down syndrome and anorectal malformation (ARM) who was found to have a rectal duplication cyst.

Materials and Methods
A 4- years-old female presented with chronic constipation in our clinic. She has undergone surgery to colostomy and transanal proctoplasty for imperforate anus at birth. Rectal and abdomen examination, abdomen X-ray, pelvic ultrasound, barium enema and magnetic resonance (MRI) were very useful to define the presence of the lesion which was located posteriorly to the bladder and have raised the suspicion of a cystic duplication of the rectum.

Results
The cyst was excised completely and histological examination confirmed a rectal duplication cyst with colonic mucosa. Complications are not documented and currently the patient is well.

Conclusions
Rectal duplications are uncommon and often located in the retro-rectal space. Duplication cyst are even rarer located anteriorly to the rectum (only five previous cases reports are available from the Literature) and more difficult to diagnose in children with ARM that may have chronic constipation because of their underlying disease. Early complete excision is desirable soon after diagnosis to minimize risk for malignant change, even if asymptomatic. Morbidity associated with complete excision is minimal and the outcome is excellent.

Keywords: Rectal duplication cyst, ano-rectal malformations (ARM).