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De Gennaro M[1], Silveri M[2], Gatti C[1], Cappa M[3], Bizzarri C[3], Mosiello G[1]
1. Urodynamic Unit - Ospedale Pediatrico Bambino Gesù - Roma, 2. Department of Pediatric Surgery - Ospedale Pediatrico Bambino Gesù - Palidoro, 3. Department of Pediatrics - Ospedale Pediatrico Bambino Gesù - Palidoro

URODYNAMIC PATTERN AND CONTINENCE IN A SERIES OF ADRENOLEUKODYSTROPHY PATIENTS

Aims of Study

Adrenoleukodystrophy is an X-linked (X-ALD) genetic disorder characterized by the breakdown of the myelin sheath surrounding nerve cells in the brain and progressive dysfunction of the adrenal gland. There are six recognized clinical subtypes of ALD according to the Moser classification. Lower urinary tract symptoms (LUTS) affect the quality of life of these patients and the rehabilitation efforts. This is the first report on a series of patients with ALD whose LUTS were explored by conventional urodynamic studies. Aims of our study were to screen patients with ambulatory urodynamic studies, to plan a reasonable flow-chart considering disability and life expectancy and to try rehabilitation/treatment.

Methods

A total of 14 patients, both children and adults, aged 8-53 years (average: 30 years) with several different phenotypes of the disease, were referred to our evaluation. Six patients were affected by adrenomyeloneuropathy (AMN) without cerebral involvement (C-); three by adult cerebral form (AC); three by childhood cerebral form (CCER); one by AMN with cerebral involvement (C+) and the last patient resulted asymptomatic. All patients were submitted to a screening protocol consisting of a confidential questionnaire, a not official version acquired from the International Conference on Incontinence Questionnaire-Short Form (ICIQ-SF) and a bladder diary. Patients without symptoms and with an ICIQ-SF score equal to 0 received no further investigation (group A- 3 patients: 2 AMN C- and 1 asymptomatic); those with daytime frequency and urgency and an ICIQ-SF score from 1 to 5 were submitted to uroflowmetry and post void residual evaluation (group B- 4 patients: 3 AC and 1 AMN C-); patients with LUTS and incontinence and an ICIQ-SF score upper than 5 underwent a conventional urodynamic study (group C-7 patients: 3 CCER, 3 AMN C- and 1 AMN C+).

Results

In 4 patients (one in Group B and 3 in Group C) a history of acute retention of urine was detected. In Group B uroflowmetry presented a fluctuating pattern with post void residual upper than 50% in 1 patient, a post void residual upper than 60% in 1 case and a normal flow rate in 2 cases. In group C, a neurogenic detrusor overactivity was the prevalent finding (6/7 patients), despite a clear detrusor underactivity was found in one case.

Conclusions

The vesico-sphincteric dysfunction can be explained both by spinal and/or cerebral involvement in the different neurological forms of ALD. Attempts of treatment with pharmacological treatment (anticholinergics) and electrical stimulation
(Stoller Afferent Nerve Stimulation) in 4 patients gave some temporary benefit which complies with the needs of patients of ameliorate their quality of life. In CCER patients the only possibility consists in imprinting the use of clean intermittent catheterisation by parents to avoid episodes of acute retention of urine.