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Wolfram Syndrome and Urodynamics: What do we know?

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Introduction

Wolfram syndrome is a neurodegenerative disorder affecting both the central and peripheral nervous systems. It is classified a rare disease, with an estimated prevalence ranging between 1/770 000 and 1/60,000. The syndrome is characterized by diabetes insipidus, optic atrophy, diabetes mellitus, brainstem damage, and bowel and bladder dysfunction. Mortality is approximately 65% before the age of 35 due to infections, respiratoryfailure, and renal failure.

Urinary tract disorders are heterogenous and can significantly impact quality of life. Highlighting the importance of early evaluation and close monitoring.

Methods

We report the case of two young girls with Wolfram Syndrome referred to the Physical Medicine and Rehabilitation Department for evaluation.

Results

CASE 1

A 16-year-old female patient presented with chronic urinary retention and urinay incontinence. Ultrasound examination revealed bladder diverticula, bilateral uretero hydronephrosis, and significant post-void residual (PVR). The patient had an indwelling catheter.

Cystometry showed detrusor overactivity associated with leak points (Figure 1). During the voiding phase, detrusor acontractility and detrusor sphincter dysynergia were observed (Figure 2).

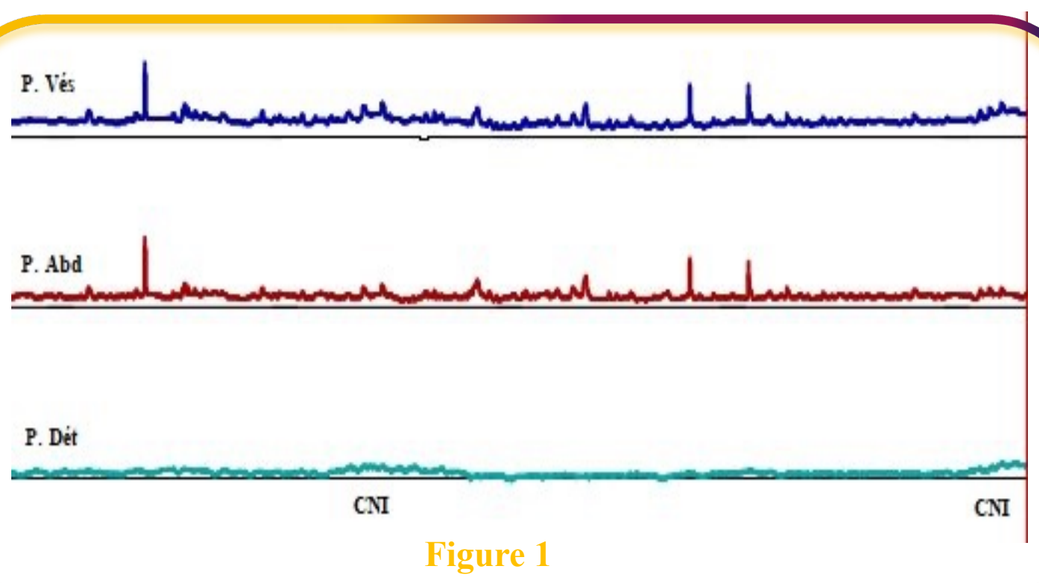

Figure 1

CASE 2

A 20-year-old female patient presented with a sensation of incomplete bladder emptying. She exhibited bilateral ureteropyelo-calyceal dilation and regular bladder wall thickness. The maximum urinary flow rate was reduced to 11ml/s with a significant PVR. The detrusor remained stable during the filling phase, but bladder compliance was decreased (Figure 3). During voiding, the patient used abdominal thrusts as the detrusor pressure was insufficient (Figure 4).

There both have a history of urinary infections.

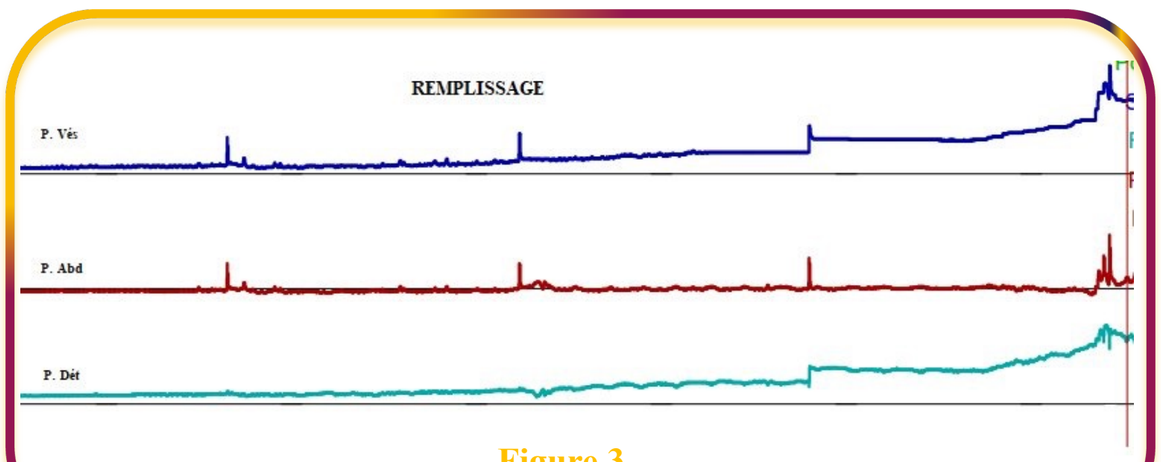

Figure 3

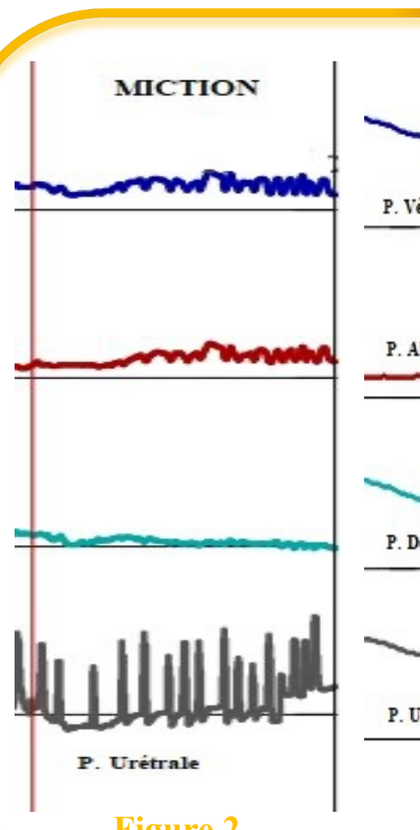

Figure 2

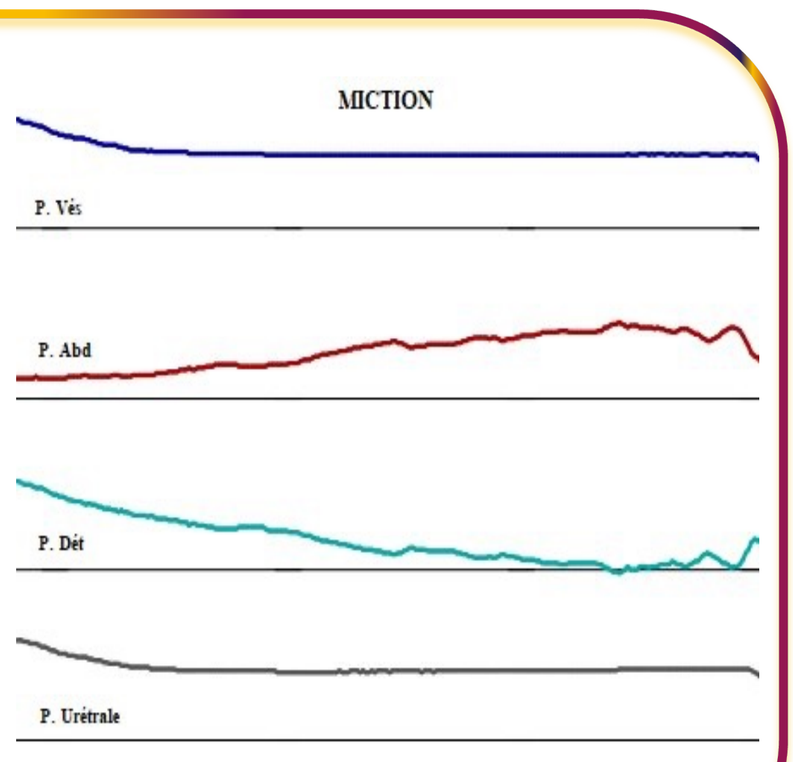

Figure 4

Discussion

Wolfram Syndrome is associated with a wide range of urinary disorders, clinically manifesting as bladder overactivity and voiding dysfunction. The litterature describes diverse urodynamic profiles, ranging from detrusor overactivity to bladder acontractility, as well as compliance disorders and sphincter detrusor dyssynergia.

Central neurogenic bladder is linked to progressive central neurodegeneration, particularly affecting the brainstem, explaining the presence of vesicosphincter dyssynergia. Detrusor hypo-contractility may resukt from neurogenic dysfunction related to diabetes. Denervation of the upper and lower urinary tracts, increased urinary flow, and polyuria associated with diabetes insipidus can lead to recurrent bladder stretch injury and ureteral dilation. Furthermore, functional obstruction to urine evacuation may contribute to ureterohydronephrosis.

The literature also reports a progressive deterioration of urodynamic parameters during the course of Wolfram Syndrome. This evolution is attributed to both myogenic and neuropathic changes in the bladder wall, underlining the need for long-term follow-up due to the risks of infectious and nephrological complications that may compromise prognosis. Treatment options for bladder dysfunction include anticholinergics and clean intermittent catheterization.

Conclusion

Wolfram Syndrome is a neurodegenrative disease involving urinary tract disorders. Early detection and urodynamic assessement are mandatory for diagnosis and follow-up, allowing for tailored therapeutic interventions to optimize management and improve clinical outcomes.

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