Functional medicine

Neurogenic bladder associated with xeroderma pigmentosum type A: A case report and literature review

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Introduction

Xeroderma pigmentosum (XP) is a rare autosomal recessive disease caused by a defect in deoxyribonucleic acid repair. Along with cutaneous symptoms, neurological symptoms are important clinical features of XP. However, information on neurogenic bladder occurrence among XP cases is rare. Herein, we describe a case of neurogenic bladder in a patient with XP type A (XPA). In this case, low bladder compliance, impaired bladder emptying, and urethral sphincter discoordination were significant cystometric findings, and frequent febrile urinary tract infection was a clinical problem. XPA patients often cannot express their symptoms because of cognitive dysfunction. Close follow-up and assessments are necessary.

Case presentation

A male patient was diagnosed with XPA at age 1 year after severe sunburn. Cognitive dysfunction and physical disability had gradually progressed, and brain atrophy was identified by magnetic resonance imaging at age 19. The patient was referred to our department for examination of bladder function after febrile UTI at age 20. Although 50–100 mL of post-void residual urine volume (PVR) was observed, bilateral hydronephrosis did not occur. After initiating treatment with bethanechol chloride [45 mg/day], intermittent catheterization; SPT, suprapubic tube; MCC, maximum cystometric capacity.

Abbreviations: XP, xeroderma pigmentosum; XPA, xeroderma pigmentosum type A; UTI, urinary tract infection; PVR, post-void residual urine volume; CIC, clear intermittent catheterization; SPT, suprapubic tube; MCC, maximum cystometric capacity.

Xeroderma pigmentosum (XP) is a rare autosomal recessive disease caused by a defect in deoxyribonucleic acid repair, and it is classified into types A to G and a variant type. In addition to cutaneous symptoms, neurological symptoms are important clinical features of XP; however, few reports have described the neurogenic bladder, especially its pathophysiology. Herein, we present a case of neurogenic bladder associated with XPA with a focus on its cystometric findings and clinical problems.

ABSTRACT

Xeroderma pigmentosum (XP) is a rare autosomal recessive disease caused by a defect in deoxyribonucleic acid repair. Along with cutaneous symptoms, neurological symptoms are important clinical features of XP. However, information on neurogenic bladder occurrence among XP cases is rare. Herein, we describe a case of neurogenic bladder in a patient with XP type A (XPA). In this case, low bladder compliance, impaired bladder emptying, and urethral sphincter discoordination were significant cystometric findings, and frequent febrile urinary tract infection was a clinical problem. XPA patients often cannot express their symptoms because of cognitive dysfunction. Close follow-up and assessments are necessary.

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A male patient was diagnosed with XPA at age 1 year after severe sunburn. Cognitive dysfunction and physical disability had gradually progressed, and brain atrophy was identified by magnetic resonance imaging at age 19. The patient was referred to our department for examination of bladder function after febrile UTI at age 20. Although 50–100 mL of post-void residual urine volume (PVR) was observed, bilateral hydronephrosis did not occur. After initiating treatment with bethanechol chloride [45 mg/day] and periodic urological evaluation, including urine test and bacterial examination, the frequency of febrile UTI had reduced. His swallowing and respiratory functions were remarkably impaired. At age 21 and 23, gastrostomy and tracheotomy were performed, respectively. At age 27, he developed acute pyelonephritis that required hospitalization, and we re-evaluated his bladder function. PVR increased slightly (approximately 100–150 mL), and bilateral hydronephrosis occurred. Cystoscopy showed severe trabeculation in the bladder, and we suspected a decrease in bladder compliance. In addition to his medications (silodosin [8 mg/day] and bethanechol chloride [45 mg/day]), clean intermittent catheterization (CIC) was initiated by his parents. After 3 years, febrile UTI did not occur, but he developed acute pyelonephritis at age 30, and we performed cystometry (Fig. 1). Detrusor pressure gradually increased from the beginning of the infusion, increased sharply after approximately 200 mL, and eventually rose to 42 cmH2O at 302 mL of maximum cystometric capacity. Voided volume was small, and the urethral sphincter contracted in the voiding phase. Meanwhile, detrusor overactivity was not detected in both filling and voiding phases. From these results, peripheral neuropathy and urethral sphincter discoordination was suspected as the cause of low bladder compliance. Although more frequent CIC was performed (≥3 times a day), the patient developed febrile UTI that required hospitalization about once a year. However, hydronephrosis and decrease of renal function had not been observed until he died of sudden cardiopulmonary arrest at age 34 years (Fig. 2).
Discussion

XP is a rare autosomal recessive disease and classified into types A to G and a variant type based on the locus of the genetic defect or mutation in the gene coding for nucleotide excision repair enzymes. The frequency of XP cases is approximately 1:22,000 in Japan, and XPA is the most frequently occurring type. Hearing loss, intellectual impairment, speech delays, and dysphagia were reported as major symptoms.

Reports about neurogenic bladder have been extremely rare. Six cases, including the present case, of neurogenic bladder associated with XP have been reported (Table 1). All were Japanese patients with XPA, and no case with other subtypes of XP was reported. Neurogenic bladder was diagnosed in the second decade of life in 5 of 6 patients, and febrile UTI and urinary retention were the reasons for urological examinations in four cases. Neurological symptoms are frequent in XPA and XP type D, and neurological symptoms occur in the first decade of life and gradually progress. A peak age of onset was reported to be different for each neurological symptom, and neurogenic bladder might be more likely to develop in the second decade of life. In this case, an increase in detrusor pressure without detrusor overactivity was observed by cystometry. This result indicated neurogenic bladder due to peripheral neuropathy. Although neuropathy can occur throughout the nervous system in XP patients, the influence of pelvic nerve damage on bladder dysfunction was strong. A high-pressure voiding has been reported as in our case; therefore, decreased bladder compliance might be a characteristic finding in XPA patients. Although we used bethanechol in this case, if CIC was initiated, antimuscarinic drugs might be effective in maintaining the low pressure in the bladder.

In this case, CIC was introduced as a treatment for impaired bladder emptying. A problem with CIC was difficult catheter insertion by a single person because of progressive severe contracture of the hip joints. With more significant problems other than the neurogenic bladder such as management of gastrostomy and tracheostomy, CIC would have been an additional burden on the patient’s family. Although the patient’s family did not agree to suprapubic tube (SPT) placement, SPT was thought to be a useful option for urine drainage.

Conclusion

We described the clinical course and examination findings of neurogenic bladder in an XPA patient. Low bladder compliance and increased PVR were important risk factors for frequent febrile UTI. XPA patients often cannot express their symptoms because of cognitive dysfunction; therefore, close follow-up and assessments should be
Consent

Written informed consent was obtained from the patient’s family.

Conflicts of interest

None.

Table 1
Reported cases of the neurogenic bladder in patients with xeroderma pigmentosum type A. *age at neurogenic bladder diagnosis.

| Patient | Sex | Age* (yr) | Reasons for urological examinations | Treatments | Ref. |
|---------|-----|-----------|-------------------------------------|------------|-----|
| 1       | M   | 18        | Febril UTI                          | Distigmine, Prazosin, Cystostomy after CIC | (1) |
| 2       | F   | 13        | Pollakiuria                          | None       | (1) |
| 3       | M   | 16        | Urinary retention, Pyuria            | Distigmine, Prazosin | (1) |
| 4       | M   | 14        | Dysuria, Bladder distension          | unknown    | (5) |
| 5       | M   | 18        | Increase of PVR                      | unknown    | (5) |
| This case | M   | 20        | Febril UTI                          | Bethanechol, Silodosin, CIC |         |

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Appendix A. Supplementary data

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