235

Mori K[1], Sumino Y[1], Akita Y[1], Sato F[1], Mimata H[1]
1. *Oita University Faculty of Medicine*

NEUROGENIC BLADDER ASSOCIATED WITH XERODERMA PIGMENTOSUM A IN JAPAN

Hypothesis / aims of study

Xeroderma pigmentosum (XP) is a rare autosomal recessive disease with cutaneous, ocular and neurological symptoms. XP is classified into a nucleotide excision repair-deficient type A–G and variant type (XP-V). XP is more prevalent in Japan (1:22000) than in the Europe and United States (1:250000) [1]. In Japan, about 500 patients with XP were existed. Approximately 50% of all Japanese patients with XP are diagnosed with XP-A and 25% are diagnosed with XP-V. It has been known that patients have high risk of skin cancer. However, it has not been well known that patients with XP-A develop neurological and cognitive dysfunction in childhood. The neurological disease advances in an orderly fashion through its successive stages, finally affecting the entire nervous system. Patients with XP-A develop voiding dysfunction because of neurological dysfunction. However, there is no detailed report about neurogenic bladder with XP-A. Therefore, we analyze four patients of neurogenic bladder associated with XP-A in Japan.

Study design, materials and methods

In this report, four Japanese patients with XP-A were obtained. The patients with XP-A were three boys aged 18, 16 and 15 years and a girl aged 13 years who complained of febrile urinary tract infection, voiding dysfunction, febrile urinary tract infection, and pollakiuria respectively. We investigated the voiding function in four patients with XP-A in Japan. Patients' informed consent was obtained.

Results

Case1: The 18-year-old boy had febrile urinary tract infection because of a large amount of residual urine, and he had bilateral hydronephrosis. Cystometry was performed to investigation his voiding function. The compliance was low such as 2ml/cmH_2O, and bladder capacity was very small such as 120ml (Figure). Some medical treatment could not improve the voiding dysfunction. Therefore, performing clean-intermittent-catheterization (CIC) by his mother was introduced to decrease residual urine. However, it was very difficult to perform CIC by mother because lower-extremity contracture was very developed. Finally, cystostomy was performed, and febrile urinary tract infection could become prevented. Case 2: The 16-year-old boy had voiding dysfunction. Residual urine was more than 100ml, and asymptomatic pyuria was recognized. Treatment with distigmine bromide and prazosin hydrochloride was performed, and the boy made urination possible and improved asymptomatic pyuria. Case 3: The 13-year-old girl had pollakisuria. The girl was sister of case 1. Residual urine, pyuria in urine analysis, and hydronephrosis were not recognized. She could not receive treatment with an anticholinergic drugmedical treatment because of astriction. Therefore, periodical follow-up was performed for the girl. Case 4: The 15-year-old boy had febrile urinary tract infection. Cystometry was performed, and indicated low compliance bladder and detrusor sphincter dyssynergia [2]. These all four patients with XP-A had already neurological dysfunction in their early childhood, and they were condemned to a wheelchair.

Interpretation of results

The onset of neurological dysfunction in patients with XP-A was recognized in their early childhood. However, detection of voiding dysfunction in patients with XP-A was usually beyond the onset of neurological dysfunction (table). Furthermore, the type of voiding dysfunction in patients with XP-A might become low compliance bladder and detrusor sphincter dyssynergia.

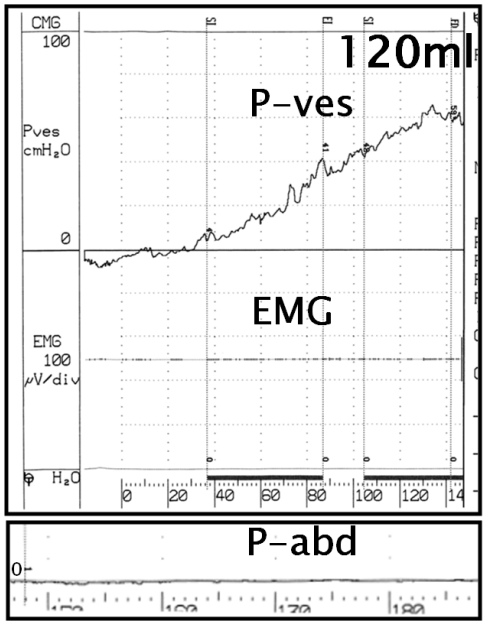
Concluding message

Since present day patients that are diagnosed early and well protected from sunlight suffer minimal skin problems and are living longer than in the past, neurological problems associated with XP becoming of relatively greater importance in XP families and focus of future research. Patients with low compliance bladder have high risk of febrile urinary tract infection and renal dysfunction. Therefore, periodic evaluations of voiding function in patients with XP-A should be initiated in their early childhood. Furthermore, the edification is very important that patients with XP-A could become neurogeic bladder.

Table: The age of onset in neurological dysfunction and that of urological consultation.

Case	XP type	Neurological dysfunction	Urological consultation
1	XP-A	5 (years old)	18 (years old)
2	XP-A	4	16
3	XP-A	5	13
4	XP-A	5	15

Figure: Cystometry in the 18-year-old boy with XP-A. The compliance was low such as 2ml/cmH_2O.



References
1. Anttinen A et al. Brain. 2008;131(8):1979-89.
2. Hamano A et al. Hinyokika Kiyo. 2009;55(7):461 (abstract)

Specify source of funding or grant	**None**
Is this a clinical trial?	**No**
What were the subjects in the study?	**HUMAN**
Was this study approved by an ethics committee?	**Yes**
Specify Name of Ethics Committee	**Ethics Committee of Nishibeppu National Hospital. Oita. Japan**
Was the Declaration of Helsinki followed?	**Yes**
Was informed consent obtained from the patients?	**Yes**