Paraplegia **22** (1984) 119–123

CASE REPORT

PARAPLEGIA DUE TO ADRENOMYELONEUROPATHY

By SOFJAN LAMID, M.D.[1] and AHMED Z. EL GHATIT, M.D.[2]

[1] *Assistant Professor of Physical Medicine and Rehabilitation.* [2] *Associate Professor of Neurosurgery and Physical Medicine and Rehabilitation Spinal Cord Injury Service, Wood Veterans Administration Medical Center and the Departments of Physical Medicine and Rehabilitation and Neurosurgery, The Medical College of Wisconsin, Milwaukee, Wisconsin 53193, U.S.A.*

Summary. Only nine cases of adrenomyeloneuropathy have been reported in the literature. It manifested as adrenal insufficiency, spastic paraparesis, loss of bowel, bladder and sexual functions, and peripheral neuropathy. Our paper describes a patient with the diagnosis of adrenomyeloneuropathy who was admitted to our hospital for rehabilitation. Family and neurological history, physical examination and special laboratory studies such as ACTH stimulation test, electrodiagnostic findings, cystometrogram and sexual function evaluation were essential to establish the diagnosis of this disease. His rehabilitation consisted of bowel and bladder training, ambulation with long-leg braces and crutches, wheelchair mobilization and transfers, and independence of all activities of daily living.

Key words: Adrenomyeloneuropathy; Paraplegia.

Introduction

AFTER a literature search, we found only nine cases of adrenomyeloneuropathy that have been reported in the past (Griffin *et al.*, 1977; Schaumburg *et al.*, 1977). The disease manifested as adrenal insufficiency, spastic paraparesis, loss of bowel, bladder and sexual functions, and peripheral neuropathy. As far as we know, rehabilitation of adrenomyeloneuropathy has never been reported before.

Case Report

A 27-year-old male patient with spastic paraparesis and neurogenic bladder dysfunction was admitted to our rehabilitation ward. He was well until he was 18 years old, when he started to have stumbling gate, loss of balance, and urinary incontinence. Lower extremity weakness continued to worsen and he had to use a wheelchair for mobilisation. Prior to his admission to our Hospital, he was hospitalised at another medical centre where he had the ACTH stimulation test. Before the test, the following laboratory report was obtained:

Serum sodium 132 mEq/L (normal 135–155 mEq/L).
Serum potassium 4·6 mEq/L (normal 3·5–5·5 mEq/L).
Plasma glucose 72 mg/100 ml (normal 63–128 mg/100 ml).
Serum cortisol at 8 a.m. 14 μg/dl and at 4 p.m. 11 μg/dl (normal at 8 a.m. 7–27 μg/dl).
Blood pressure was 130/80 mmHg.

Corresponding Author: Sofjan Lamid, M.D., Veterans Administration Medical Center, Wood (Milwaukee), WI 53193, U.S.A.

ACTH stimulation test was performed according to the following technique: 40 units of ACTH (Corticotropin) was infused intravenously in 500 ml of 5 per cent dextrose in water solution over 12-hour period. This infusion of 40 units of ACTH was repeated to a total four consecutive 12-hour periods in four days. Laboratory report after ACTH stimulation test showed the following result:

Day 1: serum ACTH 380 pg/ml (normal 0–125 pg/ml).
Day 1: serum cortisol 17 μg/dl at 8 a.m.
Day 2: serum cortisol 12 μg/dl at 8 a.m.
Day 3: serum cortisol 14 μg/dl at 8 a.m.
Day 4: serum cortisol 14 μg/dl at 8 a.m.

Conclusion

Despite the fact that the basal serum cortisol was normal the ACTH level was elevated indicating an impaired adrenal reserve. This was confirmed by the failure of serum cortisol to rise following exogenous ACTH.

Nerve conduction studies showed peripheral polyneurophathy and a cystometrogram revealed an 'unstable bladder'. Furthermore, his family history (Fig. 1) showed his brother developed adrenomyeloneuropathy at

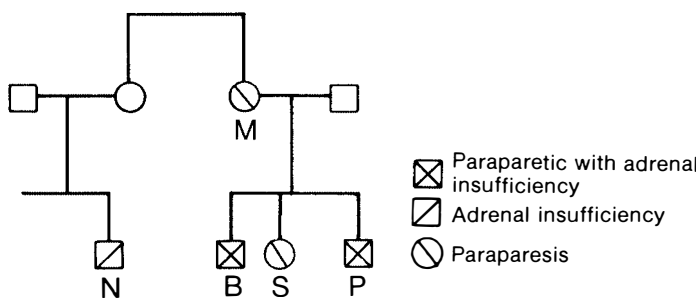

FIG. 1
Family tree of the patient

M = Mother
N = Nephew
B = Brother
S = Sister
P = Patient

age 30, his sister was paraparetic without adrenal insufficiency, and his nephew had adrenal insufficiency without paraparesis. His mother had 'muscle weakness'. The patient's brother had extensive clinical and laboratory investigations at the National Institute of Health in Bethesda (Maryland) and the diagnosis of adrenomyeloneuropathy was established. Also, his sister and nephew had complete physical and laboratory examinations at a local hospital for this hereditary condition. However, at the time of interview his mother was not seen by a physician for her 'muscle weakness'. Therefore, we do not know the nature of her 'muscle weakness'. Probably, it was associated with myelopathic disorder.

Examination revealed that the patient was paraparetic with moderate spasticity of the lower extremities, spinal cord level T10 with incomplete neurological dysfunction. He was alert and there was no evidence of dementia. His skin was bronze pigmented especially in the groin with slight dark palmar crease. Cystometrogram showed voiding pressure of 60 cm H_2O and rectal sphincter electromyography revealed detrusor-sphincter dyssnergia, an asynchronus activity of the bladder detrusor and the striated urethral sphincter muscle (Warwick, 1979). Penile erection capability was evaluated by recording the penile circumference with two strain gauges attached to a Nocturnal Penile Tumescence Monitor (American Medical System, Inc.). Patient had three erectile episodes during the 8-hour period. The average increase of penile circumference was 16 mm and the average duration of erection was 30 minutes. The result was consistent with psychogenic impotence (Kenepp *et al.*, 1979).

Additional laboratory studies were performed. The result was as follows:

Serum FSH 22 mIU/ml (normal 3–17 mIU/ml).
Serum LH 25 mIU/ml (normal 6–30 mIU/ml).
Serum testoterone 543 ng/dl (normal 300–1200 ng/dl).

We do not have any explanation why serum FSH was slightly elevated. Other tests, such as skull X-rays showed no enlargement of the sella turcica.

The patient was seen by our consultant neurologist who concurred with diagnosis of adrenomyeloneuropathy. Initially, the patient was given hydrocortisone orally 20 mg in the morning and 10 mg in the afternoon. After this medication his serum sodium levels remained slightly lower than normal. His serum sodium levels were 127 and 130 mEq/L (normal 135–155 mEq/L) and his serum potassium levels were 4·5 and 5·2 mEq/L (normal 3·5–5·5 mEq/L). Our consultant endocrinologist suggested to place patient on full replacement therapy. Oral fludrocortisone 0·1 mg daily was added to the therapeutic regimen. His serum sodium level rose by 138 mEq/L. His physical rehabilitation (Licht, 1968) included the following objectives:

1. To gain independence in bed and wheelchair activities.
2. To gain maximum use of hands.
3. To gain ability to ambulate and elevate.

Ambulation with long-leg braces and crutches was tried. It was unsuccessful because of high energy expenditure (Fisher *et al.*, 1978) and lack of patient's motivation. Therefore, a patient discontinued using long-leg braces and crutches and he used the wheelchair for mobilisation. He had 'bladder training' with intermittent catheterisation which was successful. He voided well and used external condom drainage. Routine bowel movement was established using digital rectal stimulation on every-other-day basis. At the time of discharge from the hospital he was independent in all activities of daily living (ADL), such as wheelchair transfers, toilet and hygienic activities (washing, brushing teeth, shaving, and combing hair). He was also independent in eating and dressing.

Patient was discharged from our Hospital, however, he was re-admitted to another medical centre because he developed an episode of hypotension (BP 90/60), which did not respond to the usual pressor agents. It was suggested that he had a mild adrenal crisis. He was given hydrocortisone

intravenously and had a prompt response. The patient had multiple admissions to that medical centre. He had significant progression of his disease. The sensory level advanced from T10 to T5. In addition, he had upper limb involvement which manifested as spasticity. Velar speech was suggestive of bulbar involvement. Also, he had increased respiratory effort which was probably due to intercostal muscle weakness. It was felt that he developed the clinical symptomatology of ascending myelopathy that was reaching the bulbar area. Subsequent admissions to the hospital revealed that the patient breathed with abdominal muscles and occasionally had respiratory distress episodes. He was discharged to a nursing home because most of the time he was dependent on others for his daily activities. At the nursing home his upper extremities became weaker, he had episodes of respiratory distress, choking and difficulty of swallowing. He died at the nursing home 2 years after his initial rehabilitation. Prior to his death he had fever due to urinary tract infection which triggered an adrenal crisis. The cause of his death was listed as aspiration pneumonia, respiratory failure (secondary to the progression of his disease) and adrenal crisis.

Discussion

Family and neurological history as in Fig. 1, physical examination, and special laboratory studies such as ACTH stimulation test are essential to establish the diagnosis of adrenomyeloneuropathy. It is a rare disease, males and females are affected, slowly progressive, and sudden death may occur (Griffin *et al.*, 1977). Rehabilitation in patients with adrenomyeloneuropathy is similar to those of other myelopathic conditions, such as traumatic spinal cord injury. Special attention is directed to adrenal insufficiency that should be treated with hormonal replacement therapy. However, the prognosis is uncertain. This hereditary disease is usually progressive. Our patient died with ascending myelopathy 2 years after his initial rehabilitation.

Résumé

Seulement neuf cas d'adrénomyéloneuropathie ont été signalés dans la littérature. Les symptômes de cette maladie sont: insuffisance adrénale; paraparèse spasmodique; perte des fonctions entérique, vésicale et sexuelle; et neuropathie péripherique. Notre communication décrit un patient dont le diagnostic est d'adrénomyéloneuropathie, admis à notre hôpital pour rééducation. Essentiels au diagnostic de cette maladie étaient l'histoire héréditaire et neurologique, la visite médicale, les analyses spéciales de laboratoire telles que l'analyse de stimulation ACTH, l'électrodiagnostic, le cystométrogramme, et l'évaluation de la fonction sexuelle. La rééducation du patient a consisté à lui réapprendre le contrôle de la fonction urinaire et de la fonction fécale et à lui apprendre à déambuler avec appareils orthopédiques et béquilles et par fauteuil roulant. Cette rééducation a visé à le rendre indépendant dans toutes les activités de la vie quotidienne.

Zusammenfassung

Nur neun Fälle von Adrenomyeloneuropathie sind in der Literatur berichtet worden. Charakteristisch ist die Unzulänglichkeit der Nebennieren, spastische Paraparese, Verlust von Damm-Blase und sexualer Funktion, sowie peripherische Neuropathie. Unsere Abhandlung beschreibt einen Patienten mit Diagnose von Adrenomyeloneuropathie der in unser Krankenhaus zur Rehabilitierung eingeliefert wurde. Familien und Nerven Anamäse, physische und spezielle Laboratorium Untersuchungen wie ACTH Reizungsprobe, elektro diagnostische Befunde, Cystometogram und Sexuale Funktions Abschatzung waren er-

forderlich um die Diagnose zu bestimmen. Seine Rehabilitation bestand von Darm und Blasen Trainierung, bewegliche lange Beinschienen und Krücken. Der Patient hatte die Möglichkeit sich selbst vom Bett zum Rollstuhl zu begeben sowie selbstständig im Alltag wirkend zu sein.

REFERENCES

FISHER, S. D. & GULLICKSON, G. (1978). Energy cost of ambulation in health and disability. *Arch. Phys. Med. Rehab.*, **59**, 124–133.

GRIFFIN, J. W., GOREN, E. H., SCHAUMBURG, H., ENGEL, W. K. & LORAUX, L. (1977). Adrenomyeloneuropathy: a probable variant of adrenoleukodystrophy. Clinical and endocrinological aspects. *Neurology*, **27**, 1107–1113.

KENEPP, D. & GONICK, P. (1979). Home monitoring of penile tumescence for erectile dysfunction. *Urology*, **14**, 261–264.

LICHT, S. (1968). Rehabilitation in Medicine, pp. 3–4, E. Licht Publishers, New Haven.

SCHAUMBURG, H. H., POWERS, J. M., RANE, C. S., SPENCER, P., GRIFFIN, J. W., PRINEAS, J. W. & BOEHME, D. M. (1977). Adrenomyeloneuropathy, a probable variant of adrenoleukodystrophy. General pathologic, neuropathologic, and biochemical aspects. *Neurology*, **27**, 1114–1119.

WARWICK, R. I. (1979). Observation on the function and dysfunction of the sphincter and the detrusor mechanism. *Urol. Clin. North Am.*, **6**, 13–30.