Subacute Combined Degeneration*  

A CASE REPORT

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SUMMARY

It is well known that subacute combined degeneration and other neurological syndromes related to pernicious anaemia, may present before the anaemia becomes evident. A case is presented illustrating this fact, and attention is drawn to the importance or early diagnosis of this eminently treatable condition.


CASE REPORT

History

A 72-year-old White woman was well until 8 months before admission in April 1972, when she noted the gradual onset of paraesthesia in fingers and toes. This progressed to clumsiness of the hands, with inability to fasten buttons. Three months before admission she noticed that her gait was unsteady, with a tendency to stagger from side to side. By the time of admission she was unable to walk more than 3 paces without aid.

She gave no history of headache, neck pain, or limitation of movement, and there had been no trauma to head or spine. There was no associated visual upset, no vertigo, no preceding respiratory illness or ‘flu-like illness, and at no time did she feel out of sorts. There had been no exposure to toxins, she took no drugs, no remedies of any sort, nor any alcohol.

Hers was a balanced diet and her appetite good, and she had never undergone abdominal surgery. The only therapy she had received for this illness before admission was in the form of a small bottle of ‘tonic’, the constituents of which are unknown.

Clinical Features

The patient was a fit-looking, elderly woman without anaemia or jaundice. The skin texture and hair distribution were normal and the thyroid gland was impalpable. There was no lymphadenopathy, no hepatosplenomegaly and no lumps detectable in the breasts.

The pulse was 65 per minute, and regular. All peripheral pulses were palpable. The blood pressure was 110/75 mmHg, the lungs were clear. There were no abnormal masses in the abdomen. Rectal examination and a stool were normal.

Nervous System

She was fully alert, with normal intellectual function and emotional responses. The speech, cranial nerves, and optic fundi, were normal. There was full and pain-free movement of her cervical spine.

Upper limbs: the motor power, co-ordination, and vibration sense were normal. However, pinprick sensation was diminished on the palms, and also light touch sensation below the left elbow and below the right wrist. All the upper limb reflexes were normal. No motor, sensory, or reflex abnormality was detected in the trunk.

Lower limbs: the motor power was normal; vibration sense was absent from both lower limbs and this extended to the pelvis. There was defective position sense in the toes and feet. The heel-knee test on each side was ataxic. Pinprick sensation was patchily diminished over the lower third of legs and feet. Light touch sensation was similarly diminished. Deep pressure pain and the lower limb reflexes were all normal. The plantar responses were flexor. Her gait was markedly ataxic and Romberg’s sign was present.

Investigations

The urine analysis was normal; the erythrocyte sedimentation rate (ESR) 85 mm in the first hour; haemoglobin, 13.6 g/100 ml, with a packed cell volume (PCV) of 38%; mean corpuscular volume (MCV), 109 μm³; mean corpuscular haemoglobin concentration (MCHC), 34%; total white cell count 5 700/mm³; differential count: 54% polymorphs, 38% lymphocytes, 5% monocytes and 3% eosinophils; platelet count 195 000/mm³; and reticulocyte count 2.2%.

Examination of the peripheral smear showed the red cells to be normochromic and normocytic. There were no hypersegmented neutrophils. Examination of the bone marrow showed an essentially normal marrow with some very early megaloblastoid changes in the erythroid series.

The serum iron was 100 μg/100 ml; total iron-binding capacity 340 μg/100 ml, and 29% saturation; serum vitamin B₁₂, 49 pg/ml (normal 300 - 1 100 pg/ml); blood urea 30 mg/100 ml; serum creatinine, 0.7 mg/100 ml; protein electrophoresis, a normal pattern; and protein-bound iodine (PBI), 6.4 μg/100 ml.

The test for thyroid antibodies was negative; the serum VDRL and fluorescent treponemal antibody (FTA) tests were negative; the cerebrospinal fluid was normal and the VDRL and FTA tests on the cerebrospinal fluid.
were negative. The augmented gastrin test revealed a total achlorhydria; the Schilling test showed a 0.5% excretion of the isotope in the urine over 24 hours, which rose to 20% excretion with the addition of the intrinsic factor.

The chest X-ray film, barium meal and follow-through were all normal; and screening tests for porphyrins in urine and stool were negative.

Course and Management

The neurological deficit with low vitamin B₁₂, abnormal Schilling test corrected with intrinsic factor, and total achlorhydria, strongly suggested the diagnosis of subacute combined degeneration complicating pernicious anaemia. However, the high ESR was unexplained. No primary neoplasm could be found in thyroid, breast, chest, or upper gastro-intestinal tract; at follow-up, 6 months later, the ESR was normal.

Therapy was started with intramuscular vitamin B₁₂, 1000 μg/day for 5 days. Within 7 days of therapy there was subjective improvement in the paraesthesia, and a week later she could walk unaided, though unsteadily, and could fasten buttons on her blouse. There was no change in her haemoglobin or reticulocyte count. Six months later she was well, her gait almost normal, and she no longer had paraesthesia.

DISCUSSION

There is little doubt, in the light of the above description, that this patient had neuropathy related to her vitamin B₁₂ deficiency and that she had pernicious anaemia. The neurological syndromes of pernicious anaemia may precede the anaemia, sometimes by many years. The neurological syndrome may take the form of a psychiatric disturbance with personality change, visual disturbance including optic atrophy, or more commonly, a neuropathy with the chief stress on paraesthesia with loss of vibration sense which is out of proportion to other modalities, particularly in the lower limbs. Ankle jerks may be absent and extensor plantars present, but reflexes may be normal. The serum B₁₂ is always very low—less than 80 pg/ml. Delay in diagnosis affects prognosis and the earlier specific therapy is begun, the better the chance of recovery. Neurological change, if left too long, can become permanent.

This case is presented to draw attention to the appearance of subacute combined degeneration prior to that of the anaemia. In any neuropathy where the accent is on sensory loss, with vibration sense particularly affected, and with defective position sense, vitamin B₁₂ deficiency should be considered, as the results of early therapy are very rewarding.

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REFERENCES