

Submitted by: Drs. William C. Schoene and Patrick Lynch
Brigham and Women's Hospital
Boston, Massachusetts 02115
and
Royal Infirmary, Preston, England

Reference No.: NSP 35/79-XP BBH-387

Clinical Abstract:

The patient, a 42 year old mentally-retarded female, was referred for examination at the age of 34 years because of progressive lethargy, loss of appetite, urinary incontinence and a tendency to fall to the right when walking. She was born to unrelated parents at the term of a first pregnancy. Delivery was effected by forceps after a labor lasting 6 hours. Her birth weight was 8½ lbs. She held her head up at 4 months, sat at 8 months, said her first words at 10 months and crawled normally until the age of 18 months. At 7 months she had the first of prolonged bouts of unconsciousness each lasting 5 to 6 hours. These attacks occurred from time to time until she was 6½ years old. As a child she had measles and whooping-cough. She attended special schools, workshops, and ultimately an Adult Training Center. At the age of 37 she was found to have an I.Q. of 30.

Physical examination revealed a somewhat infantile female weighing 94 lbs. She was mentally-retarded and exhibited a plastic type of rigidity in the upper and lower limbs on both sides. All laboratory investigations, including serum caeruloplasmin and thyroid studies, were normal.

Progress and Treatment: The patient was seen again at the age of 38 years. By then she was demented, smiling occasionally but not understanding what was said to her.

Physical examination revealed normal optic fundi. There was bilateral footdrop, paraplegia on extension in both lower limbs and inversion of the left foot. The plantar responses were extensor. She was treated initially with Sine-met and Anafranil syrup, but because of intolerance to Sinemet this was replaced by Amantidine. The patient was lost to follow-up until September, 1978, when she was admitted to hospital because of severe bed sores and dental caries. She died 48 hours after admission.

Necropsy Findings:

Principal abnormalities outside the central nervous system were severe wasting, dental caries, decubitus ulcers and bronchopneumonia.

The formalin-fixed brain weighs 790 gm. (the cerebellum and brainstem together weighed 130 gm.). There was generalized gyral atrophy in both cerebral hemispheres especially affecting the frontal lobes. There was moderate symmetrical dilatation of the lateral ventricles. The cerebral cortex and white matter appeared normal. The pallida were light brown and the substantia nigrae contained a broad band of rust-colored pigmentation. The cerebellum appeared normal.

MATERIAL SUBMITTED: One H & E slide; one slide stained by Bielschowsky method

Points for Discussion:

1. Nature of pathological changes
2. Relationship, if any, of various lesions to each other