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This patient was the product of an unremarkable full term pregnancy in a gravida 3 mother. She appeared entirely normal at birth. Early developmental milestones were within normal limits. She sat alone at 8 months. From then on she progressed somewhat more slowly, crawling at 14 months, and walking with support at 18 months. She never walked alone, and she developed no speech. After age 18 months there was gradual regression in motor performance. By age 28 months she no longer stood or crawled, and she could sit only with support. She had lost the ability to feed hereself. There had been no seizures.

Physical examination at age 28 months showed a well-nourished, fair-skinned child, who was in the 25th percentile for height and weight. Head circumference was 45.8 cm. Visual fixation and following were present, and the pupils reacted briskly to light. A right convergent strabismus was noted. A constant, fine pendular nystagmus was present at rest, and was replaced by a jerk nystagmus on lateral gaze. Fundoscopic examination showed a small cataract on the right, and questionable optic atrophy bilaterally. The child reached for objects presented to her, but grasping movements were clumsy. She appeared generally hypotonic. Little spontaneous movement was seen in the feet. Tendon reflexes were normally active in the upper extremities, brisk at knees, and diminished at ankles. Plantar responses were extensor. There was little response to pin prick over the extremities or trunk, but a normal response was noted over the face.

Re-examination at age 31 months showed absence of ankle jerks, but no other obvious change. The child was subsequently admitted to a school for the retarded.

At age 5-1/2 years the child showed further evidence of worsening in neurologic state. She now was emaciated, weighing only 12 kg., height being 107.5 cm. The head circumference still was only 45.8 cm. Liver and spleen were not palpable. There was a blink response to light, but no visual following, and the pupils responded only sluggishly to light. No movements of the eyes could be elicited on doll's head maneuver. Fundoscopic examination showed pallor of optic discs bilaterally. Corneal reflexes were sluggish. Facial movements were weak bilaterally, and eye closure was incomplete during sleep. The child turned toward loud noise. When undisturbed, she was lying quiently, with elbows and wrists flexed, and legs extended. There were no voluntary movements of the legs. Tendon reflexes were hyperactive in the upper extremities, but knee and ankle jerks were absent. Plantar responses were extensor.

The subsequent course was one of gradual deterioration. She became unable to swallow and required tube feedings. Death occurred at age 9 years.

At autopsy, the visceral pathology was not contributory. Widespread griseal involvement was present.

Diagnosis: Cortical deposits in infantile neuroaxonal dystrophy.

Main Point for Discussion: Locus of the extraneuronal small bodies.

1 H and E slide submitted.