CASE 8

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Clinical Abstract:

This 25 month old boy was normal at birth, with head circumference of 34.0 cm. (50%). His early development was normal. When first seen, at age 9 months, the head circumference was 43.5 cm. (25%), and he was apathetic. The anterior fontanelle was closed. A CT scan disclosed mild cerebral atrophy. At age 11 3/4 months the head circumference was only 44.0 cm. (<2%), and he did not show consistent imitative behavior. Deep tendon reflexes were abnormally brisk, with bilateral ankle clonus and extensor plantar responses. By 18 months of age he displayed no imitative behavior, he was only able to crawl for short distances, and he had minimal receptive language function. He continued to deteriorate, losing the ability to crawl and to sit. The head circumference did not increase further.

The patient's mother was an intravenous drug abuser. At age 9 months the child had desquamative interstitial pneumonitis, diagnosed on open lung biopsy. Biopsy of the thymus at that time revealed involution. A duodenal biopsy at age 19 months revealed acid fast bacilli in the submucosa, and culture grew Mycobacterium avium - intracellulare (MAI). He was treated for MAI with appropriate antibiotics, but a repeat duodenal biopsy at age 23 months revealed persistence of the organism, although fewer bacilli were present on acid fast stain. He died at age 25 months.

Autopsy revealed disseminated MAI. Lymph nodes not involved by MAI were normocellular or hyperplastic. The lungs had foci of desquamative interstitial pneumonitis. There was also calcification of medium and large-sized arteries, involving spleen, lungs, kidneys, thymus, mesentery and coronary arteries. The brain was reduced in size but did not have any other gross abnormalities.

Material submitted: One H+E slide of cerebral cortex and white matter
One H+E slide of basal ganglia

Points for discussion:
1. Diagnosis
2. Pathogenesis