

Case 7

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Clinical Abstract: This young girl suffered severe mental impairment and died at the Kansas Neurological Institute in Topeka, KS, at the age of 10.

Past History - The patient was the product of an uneventful pregnancy and delivery, and appeared to be developing normally up to the age of one year, when the mother noticed that other children appeared to be getting ahead of her in developmental milestones. She had started to take steps without support but had progressively more difficulty walking. About that time it was noted that she became increasingly hard of hearing.

Family History - Negative for neurologic or developmental disease among parents, uncles and aunts.

Course - At age of 27 months the patient was admitted to Kansas University Medical Center. She was well nourished and friendly but with obviously short stature and disproportionately large head: 48 cm. in circumference (greater than 25th percentile). She had a prominent forehead and depressed broad bridge of nose. No cherry red spot was found in the fundi. Her white blood cells showed an abnormality of granulation. Gradually the patient became profoundly impaired and from the age of 3 years was given institutional care at KNI. She died of an intercurrent infection.

Autopsy Findings: At the time of death her head circumference was 51 cm. The brain was small, weighed 570 gm. The leptomeninges were thickened. There was marked hydrocephalus involving all ventricles and the aqueduct, and there was massive distention of the cisterna magna. Because of the hydrocephalus the cerebral hemispheres felt fluctuant, but the brain stem and cerebellum had a firm consistency.

Points for Discussion:

1. Diagnosis?
2. Is the fact that certain abnormalities involve the grey as well as the white matter in this case helpful in suggesting the diagnosis?