CASE 6

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Ref. No. A67-261

The patient was a 9 month-old Negro male at the time of his death in March, 1967. The mother was 21-years old at the time of delivery, gestation had lasted forty weeks, there had been no maternal illnesses, and labor was uncomplicated. The baby's birth weight was 2,630 gms.; length 44.0 cms.; head circumference 29.0 cms. (N.: 36 cms.); and chest circumference 30.5 cms. The infant appeared well-developed, but had a markedly small head, with a sharp angulation at the occipital-cervical region. At twelve hours of age, left facial and right hand seizures were observed. Muscle tone was increased and Moro reflex was present; baby was nursing normally. At ninety-six hours he had lost 130 gms. and was still convulsing. Phenobarbital was administered. Seizures stopped temporarly at six days of age. FBS was 32 mg.%.

At three months of age he was seen in the clinic after being maintained on 20 mg. a day of phenobarbital without complete relief of the seizure activity. Head circumference 32.0 cms. (N.: 40 cms.), chest circumference 27.0 cms., body length 54.0 cms. There was spasticity of the extremities with fisting of the hands, but no other abnormalities.

The patient was readmitted to the hospital at the age of eight months (body weight 6 lbs. 2 oz.). He had been maintained on phenobarbital 15 mg. a day, but had continued to have "occasional twitching". At this time the seizure activity had increased and the patient could be fed only with a medicine dropper. Lethargy had developed. The infant cried weakly and there was noticeable spastic quadriparesis. Spinal fluid: protein 17 mg.%, glucose 81 mg.% (FBS 115 mg.%). Occasional twitching of the face and jerking of the upper extremities were observed at the time of his discharge on the twentieth hospital day. Patient had lost his sucking reflex.

At the time of his last admission (one week later) the child was in deep lethargy, convulsing continuously, and markedly dehydrated. Cyanosis, diarrhea, and vomiting were additional clinical problems. He expired 24 hours later.

Autopsy Findings: Marked malnutrition (body weight: 3450 gms.), lipid pneumonitis, and ulcerative colitis (probably of bacterial origin).

Brain: (weight: 130 gms.; normal for this age: 750 gms.). There was a remarkable degree of atrophy of the convolutions with exposure of the insula, and increased consistency. No structural malformations were noted. Ventricular system was dilated.

Sections submitted are from cerebral cortex, H & E and two blanks. Two gross Kodachromes are also submitted.

Diagnoses: (1) Leukodystrophy (?), (2) Hypoglycemia (?)