

STUDY REPORTS

1. Title : Early Juvenile Cerebral Lipoidosis and Its Occurrence in a Thai Family

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Objective : This study was designed to diagnose a progressive generalized neurological disease in an eight year old child and to evaluate its occurrence in the child's kinship.

Description : The child is an eight year old Thai female who was admitted to the hospital because of a progressive dementia, mutism, rigidity and a movement disorder.

The patient was the product of a normal gestation and birth. Developmental milestones were all delayed (the parents state that this is normal in all their children.) The child sat alone at 8 to 9 months, crawled at 1½ years, began walking unassisted at three and was toilet trained at four. At two the child received both diphtheria antitoxin and a small-pox vaccination without apparent ill-effects. At three, when she began walking she was unsteady. This unsteadiness became progressively worse over the next year. Speech did not progress beyond simple monosyllable phrases. At four the child could say only occasional simple words and at five was completely mute. Increased resistance to passive movement appeared at five. At seven the child was unable to crawl or walk. Forced grimacing, dystonic postures, and choreo-athetotic movements started at five and became progressively more severe. The parents state that the child's vision has always been adequate and during the course of illness has not gotten worse. They have not observed any seizures or myoclonic jerking.

The patient has five siblings. A male died at age twelve of a progressive neurological disease which apparently began at two with ataxic gait, progressive dementia, mutism, rigidity and choreo-athetotic movements. Prior to death the child had a generalized seizure. No blindness is reported in this child. Another child, a six year old male began having increased tone and a mild ataxia in the lower extremities at age three. His speech has not progressed beyond the three year level. A four year old male is beginning to demonstrate a mild ataxic gait. The ten and two year old males are healthy.

Examination of the patient revealed an emaciated, mute child with numerous bed sores. She was incontinent of urine and feces and drooled intermittently. At times she could carry out simple instructions but most often disregarded all directions. Head circumference was normal. Liver and spleen were not palpably enlarged.

Grimacing, choreo-athetotic movements, and body contorsions were present and were aggravated by tactile stimulation. There was a plastic resistance to passive movement in all extremities.

Examination of the fundi revealed bilaterally strophic nerve heads. The retinal vessels were sparse and attenuated and there were spider-like areas of degeneration in the periphery of both fundi. The macular areas were brick-red and were surrounded by a grayish halo. Both pupils reacted directly and consensually to light. The child could often follow dangling objects.

All laboratory and X-ray examinations were normal. Some of the following results are of interest:

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Spinal fluid: The fluid was clear, no cells were seen on microscopic examination and the total protein was 17.5 mgms%. The colloidal gold curve was flat.

Liver function: BSP was 1.9% in 45 minutes. Amino acid distribution in urine was normal. No metachromatic granules were found in the urine. The EEG showed bursts of non-focal high voltage slow waves and spikes.

In this case the principal diagnoses can be placed in two large groups of diseases: the cerebral lipoidosis and the leukodystrophies. The presence of a macular lesion makes the diagnosis of cerebral lipoidosis clinically more likely. The age of onset and progression of symptoms further places the illness in the group of early juvenile cerebral lipoidosis. This illness has not been previously reported in Thailand.

To help confirm the clinical impression a brain biopsy was done at Sirriraj Hospital, 4 August 1965. A small bone flap was turned in the right anterior frontal area and a plug of cortex and white matter removed. The biopsy specimen was examined at Sirriraj Hospital, at the SEATO Medical Research Laboratory and at the Neuropathology Branch, AFIP. The biopsy tissue contained no definite changes characteristic of either a cerebral lipoidosis or a leukodystrophy.

Thirty-four patients in this child's kinship were examined. Except for a paternal aunt with bifid thumbs, no congenital abnormalities and no neurological disease were found. Refer to the generalogical chart for a listing of the entire kinship.

The three living affected children will continue to be followed. If a death should occur in these children a complete necropsy will be requested.

Summary: A Thai family has been uncovered in which three children have a progressive, generalized neurological disease; a fourth child recently died of the illness. Clinically, the disease appears to be an early juvenile cerebral lipoidosis; unfortunately, the recent examination of a cortical brain biopsy taken from the most severely affected child has not confirmed this impression. Thirty-four living members of the kinship of this family have been examined and no similar or related illness has been found.

KINSHIP OF A THAI CHILD WITH A GENERALIZED PROGRESSIVE NEUROLOGICAL DISEASE FATHER'S FAMILY

