CASE NOTE

TAY-SACHS'S DISEASE (AMAUROTIC FAMILY IDIOCY)*

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The following cases are reported to draw attention to the occurrence of this rare condition in India.

Case Reports

Case 1, a male Hindu child, born at full term after a normal pregnancy and confinement history, was first seen at the age of 2 years in the Paediatric Department, V.J. Hospital. He had convulsions shortly after birth and was brought to hospital with a persistent respiratory illness. The mother reported that the baby had never been able to lift his head or sit up, recognize her, or take any interest in the world around him. He was the sixth in birth order. Three siblings were aged 7 and upwards and appeared normal, and two had died, one shortly after birth and the other at 8 months with convulsions before reaching the stage of being able to sit up.

The child was ill with bronchopneumonia, did not follow light, lift his head, or show any interest. He was able to move all four limbs. The deep jerks were brisk, and plantar reflexes flexor. Fundus examination showed clear media, normal discs, and at each macula a greyish-white area about 2 disc diameters in size with a typical "cherry-red" spot at the fovea. Laboratory tests were negative, but x-ray examination showed bilateral cystic disease of the lungs. The parents were not consanguineous.

Case 2, a male Hindu child aged 4, was brought to the same hospital on account of his inability to sit up or talk. He had been seen by doctors since the age of 8 months and pronounced mentally backward but the eyes had not been examined. He was the first and only child of unrelated parents and nothing abnormal was disclosed in the history of the family, the pregnancy, or the birth. Physically he thrived for the first few months. Examination showed that the child was poorly nourished with hypotonia and dystonic spasms of rigidity. Tendon jerks were present, and plantar reflexes flexor. There was no paralysis of any part of the body. He could follow light and recognize his mother but could not sit up or speak. Fundus examination showed a typical picture as in Case 1. All other pathological and radiological investigations were negative.

Discussion

The parents had no history of consanguinity, which is strictly forbidden by Hindu custom, and there was no Jewish element in these two Punjabi families. Both patients were males and the only suggestion of familial incidence was the death of a brother of Case 1 where a fundus examination might have shown the same macular picture.

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