CASE REPORT

Smith – Lemli – Opitz Syndrome

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Abstract

This is a very rare autosomal recessive disorder characterised by short stature, microcephaly, low set ears, hypospadias (in males), pyramidal signs in addition to several other features as described.

Key Words

Microcephaly, Short stature, Hypospadias

Introduction

This is very rare disorder of autosomal recessive inheritance, characterized by short stature, microcephaly, hypospadias and several abnormalities of cerebrum, cerebellum and spinal cord (1,2). To our knowledge, our case is the first reported from India so for.

Case Report

A 16 year old boy was admitted with one day history of altered level of consciousness, frothing at mouth and incontinence of urine. There was no history of fever, convulsions, trauma to head or drug intake, nor was any history suggestive of such illness in the past or any illness requiring prolonged treatment or hospitalization. On examination he was stupurous, irritable, moving all limbs on painful stimuli. His pulse rate was 62 per minute, regular, synchronous without any special character and all the peripheral pulses were felt. Blood pressure was normal (110/70 mmHg) and respiratory rate was 18/minute. His general physical examination revealed microcephaly, small chin, low-set ears, hypospadias and short stature (Fig. 1). Pupils were pinpoint and fasciculations were present. Chest examination revealed coarse crepitations all over. On noticing the signs of organophosphorus poisoning, immediate attention was paid to this illness, and was aggressively treated with atropine and pralidoxime besides the usual supportive treatment and nursing care. Within 12-14 hours his level of consciousness improved, and he developed signs of atropine toxicity. Finally he recovered from organophosphorus poisoning and the subsequent history elicited from the attendants revealed that organophosphorus compound was applied to his scalp for treatment of lice, one day prior to the present illness. He was third in birth order, the first two were a boy and a girl respectively with no apparent illness, and they belonged to consanguineous parents. The patient had been growing well upto fourth year of life after which his mental functions were found slower compared to those of the children of same age from neighbourhood. An overall examination revealed the patient having mental retardation, short stature, microcephaly (with head circumference of 58 cms), low set ears, small chin, high arched palate, and hypospadias. His subsequent chest examinations were

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within normal limits. Neurological examination showed him mentally retarded, and was able to speak only few meaningful words. Fundus examination was normal and there was no cranial nerve dysfunction. Motor system didn't show any remarkable abnormality other than bilaterally positive Babinski signs. Sensations couldn't be tested in detail because of his mental retardation. On investigation his hemogram, serum biochemistry including immunoglobulin assay were within normal limits. Radiological survey revealed small skull and mandible (Fig. 2). CT scan revealed small skull and brain matter with widening of sulci and normal ventricular system (Fig. 3). Electro–and echocardiography didn't show any evidence of cardiac disease.

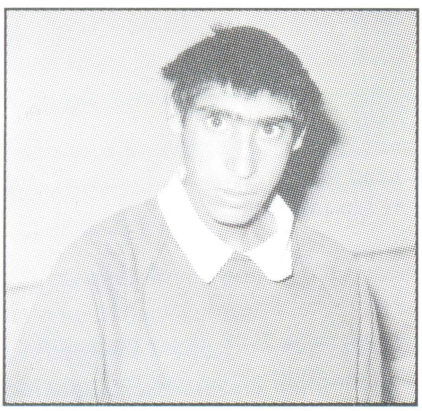

Fig. 1. Photograph showing facial features with low set ears.

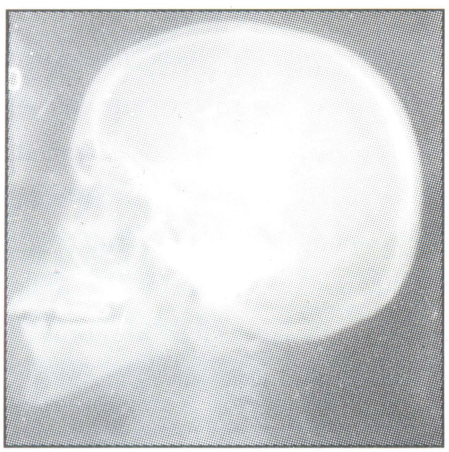

Fig. 2. Photograph showing radiograph revealing small skull.

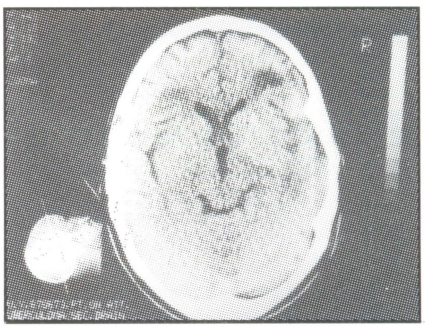

Fig. 3. CT scan head showing small skull normal ventricular system and widening of sulci.

Discussion

This is a very rare disorder, comprising mental retardation, low birth weight, failure to thrive, microcephaly, epicanthic folds, broad nasal tip and anterverled nares, ptosis, broad maxillary alveolar ridges, abnormally rotated auricles, micrognathia, cleft palate, cutaneous syndactyly of toes, other abnormalities of feet and toes and cryptorchidism and hypospadias in males. These features occur with variable severity and, many additional features have been described, making the disorder sometimes difficult to diagnose. In addition there are multiple anomalies of cerebrum, cerebellum and spinal cord. Older survivors are bereft of language and paraparetic with hyperreflexia and Babinski signs. Dislocation of hips is also found (1-3). Electroencephalo graphic abnormalities have been observed in some cases (3). Usually there are normal aminoacids and immunoglobulins, however, the accumulation of 7–dehydrocholestrol in the blood, is considered the specific marker (2). The diagnosis of the present case was solely made on the basis of clinical features, and was similarly differentiated from closely related conditions of dwarfism like nanocephalic dwarfism, Russel - silver syndrome, Rubinstein - Tabi syndrome, Pierre Robin - and DE Lange syndromes (2,3). Although little literature is available about the disorder, our patient needs regular follow up for future course.

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