

## **Septo-optic Dysplasia**

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Septo-optic dysplasia designates a unique and rare malformation of anterior midline structure of the brain including: (i) agenesis of the septum pellucidum, (ii) primitive optic ventricles, and (iii) hypoplasia of the optic nerves, chiasma and infundibulum(1,2). It is also known as De Morsier syndrome that first described its necropsy findings in 1956 and coined this term. Huseman *et al.* in 1978 considered it a form of holoprosencephaly as it results from abnormal induction of tissue from the forebrain and not a defect in the closure of the neural tube (1).

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Cases of septo-optic dysplasia with classical features or its variants have been described in the Western literature (3-5). We are not aware of any report from Indian workers and this prompted the current description.

### **Case Report**

A four months old male infant was brought with suspected blindness. He was born normally to a 28 years old second gravida mother after a term uncomplicated pregnancy. Birth weight, length and head circumference were 2.9 kg, 50 cm and 34 cm, respectively. The baby had prolonged jaundice in the neonatal period but there was no history suggestive of hypoglycemia, convulsions or any other problem. Feeding and activity were normal. At about 1 month of age the parents noted lack of response to bright objects or light.

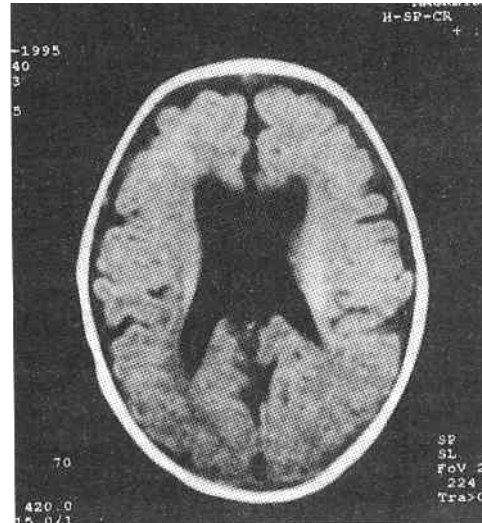
On examination his weight, length and head circumference were 5.7 kg, 60 cm and 37 cm respectively. Anterior fontanel was 4 x 4 cm in size and at level. There was moderate pallor. Ophthalmological examination revealed absence of response to light and small pale optic disc with typical double rim. There were no other abnormal findings. Hemoglobin was 8.2 g/dl. Other routine blood and urine tests were normal. Results of neuro-endocrinal investigations (*Table I*) showed normal thyroid profile and FSH with low levels of basal GH. Prolactin and LH levels were on the higher side of the normal range. CT scan head showed absent septum pellucidum and squared off configuration of the frontal horns. Magnetic resonance imaging (MRI) revealed hypo plastic optic nerves and chiasma along with absent septum pellucidum and box like configuration of the frontal horns of both lateral ventricles (*Fig. 1*). Third ventricle, thalamus and basal ganglia were normal. No other anomaly of the brain was detected.

**TABLE I**—*Neuroendocrinal Investigations*

Investigation	Results	Normal range
Growth hormone (ng/ml)	1.2	1-10
Prolactin (ng/ml)	24.0	0-25
LH (MIU/ml)	1.7	0.5-1.0
FSH (MIU/ml)	2.0	0.5-30
Thyroid profile		
T <sub>3</sub> (ng/ml)	0.8	0.8-1.8
T <sub>4</sub> (ng/dl)	6.7	5.0-12.7
TSH (nU/ml)	4.1	0.5-5.0

**Discussion**

This case had typical features of



*Fig.1. MR (T<sub>1</sub> weighted) axial image showing absence of septum pellucidum.*

septo-optic dysplasia. The syndrome is more frequent in female babies born to young primigravida mothers with uneventful antenatal period. Two major etiological and embryological theories include developmental theory and destructive theory(6). According to the former theory some teratogenic interference at six weeks of gestation disrupts the normal development and differentiation of the retinal ganglion cells, hypothalamus and the septum pellucidum. The second, popularly supported theory suggests an intrauterine destruction of already established retinal ganglion cells, optic nerve, hypothalamus and septum pellucidum. This intrauterine insult can

occur any time during pregnancy before the full development of the visual pathway.

The present case had some variations as compared to classical description of this syndrome(1,2). These include male sex, normal growth and lack of hypoglycemia and seizures. The mother was 28 years old and not a primigravida. Brook *et al.*(4) observed that age of the child may have some effect on the clinical presentation. The youngest (2.7 year) patient in his series appeared to be growing well and had normal endocrinal function but the older children manifested progressively more severe endocrine and growth problems. The reason for the same is not explained. Age of the present case was four months only. Long term follow up of these children is needed in order to assess their future growth.

Prolactin levels were at upper limit of the normal range and only basal GH levels could be estimated in the present case which is not diagnostic of GH deficiency. Normal thyroid functions found in this case are also reported earlier(1). Margalith *et al.*(6) in 1985 reported an extremely variable spectrum of neuroendocrina-

logical findings that ranged from deficiency of growth hormone to hypersecretion of GH, corticotropin and prolactin.

#### REFERENCES

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#### NOTES AND NEWS

##### NATIONAL CONGRESS ON HEALTH AND DIETARY FATS

The Human Nutrition Unit, All India Institute of Medical Sciences in association with IAP subspecialty chapter on Nutrition is organizing this event on 29th October 1996. The objective of this Congress is to orient the academicians (Faculty Members in different Medical Colleges and Colleges of Home Science) in India on Health and Dietary Fats with special reference to Palm oil. Some of the topics deliberated will be: (i) Utility of crude palm oil in prevention of vitamin A deficiency amongst children; (ii) Effects of dietary fats and oils on blood lipids and cholesterol; and (iii) Health implications of trans-fatty acids. Guest speakers from Australia and Malaysia will be delivering the state of art lectures.

To encourage participation of young faculty members from medical and home science colleges, 30 travel fellowships are available on first come first served basis. The fellowship includes registration fee of Rs. 500/- and reimbursement of the actual fare in railway travel by II class. For details please contact: Dr. Umesh Kapil, Organizing Secretary, Addl. Professor, Human Nutrition Unit AIIMS, New Delhi 110 029. Telephones: 661123/Extn 3383 and 4632; 6864851/Extn 3383 and 4632.