

addition to alopecia and milia. A hamartomatous mass on the ventral surface of the tongue is seen in 70% of the cases(7). Our patients' clinical findings were not in accord with the other types of OFD. However, patients without a pathogenic OFD 1 mutation have low frequently lingual hamartomas (7). We would like to emphasize that any patient with tongue masses and cleft palate associated with feeding problems should be evaluated for OFD 1.

Contributors: All authors contributed to case management, review of literature and drafting of the article.

Funding: None.

Competing interests: None.

REFERENCES

1. Papillon-Léage (Mme), Psaume J. Une malformation héréditaire de la muqueuse buccale: brides et freins anormaux. *Rev Stomatol (Paris)* 1954; 55: 209-227.
2. Wettke-Schafer R, Kantner G. X-linked dominant inherited disease with lethality in hemizygous males. *Hum Genet* 1983; 64: 1-23.
3. Feather SA, Woolf AS, Donnai D, Malcolm S, Winter RM. The oral-facial-digital syndrome type 1 (OFD1), a cause of polycystic kidney disease and associated malformation, maps to Xp22.2-Xp22.3. *Hum Mol Genet* 1997; 6: 1163-1167.
4. Gedeon AK, Oley C, Nelson J, Turner G, Mulley JC. Gene localization for oral-facial-digital syndrome type 1 (OFD 1: MIM311200) proximal to DXS85. *Am J Med Genet* 1999; 82: 352-354.
5. Ferrante MI, Giorgio G, Feather SA, Bulfone A, Wright V, Ghiani M, *et al.* Identification of the gene for oral-facial-digital type I syndrome. *Am J Hum Genet* 2001; 68: 569-576.
6. Majewski F, Lenz W, Pfeiffer RA, Tunte W, Muller H. Das oro-facio-digitale Syndrome: Symptome und Prognose. *Z Kinderheilkd* 1972; 112: 89-112.
7. Thauvin-Robinet C, Cossee M, Cormier-Daire V, Maldergem LV, Toutain A, Alembik Y, *et al.* Clinical, molecular, and genotype-phenotype correlation studies from 25 cases of oral-facial-digital syndrome type 1: A French and Belgian collaborative study. *J Med Genet* 2006; 43: 54-61.

Intracranial Calcification in β -thalassemia Major

S Verma

A K Saxena*

R K Marwaha

A 13-year-old boy with thalassemia major presented with generalized seizures. He was non-chelated and had hypoparathyroidism with extensive intracranial calcification, an association that is rarely reported.

Key words: Hypoparathyroidism, Intracranial calcification, Seizure, Thalassemia.

Unchelated transfusion dependent β -thalassemia major children are at risk to develop endocrinopathies(1). Hypoparathyroidism, leading on to hypocalcemia, seizures and tetany is one such well-

recognized endocrine disturbance(2). However, metastatic calcification in the central nervous system as a consequence of hypoparathyroidism is an infrequent occurrence in these individuals(3,4). The rarity of the condition prompted us to report this case.

Case Report

AK, a 13-year-old boy with transfusion dependent thalassemia major presented with

From the Department of Pediatrics and Radio-diagnosis, Advanced Pediatric Center, Postgraduate Institute of Medical Education and Research (PGIMER), Chandigarh, India 160 012.*

Correspondence to: Dr. Sumit Verma, Advanced Pediatric Center, Postgraduate Institute of Medical Education and Research (PGIMER), Chandigarh, India 160 012. E-mail: drsumitverma@yahoo.com

Manuscript received: May 14, 2007;

Initial review completed: May 31, 2007;

Revision accepted: June 5, 2007.

generalized seizures. The diagnosis of thalassemia was established in early infancy. He received red cell transfusion at intervals of 2-3 weeks to maintain a pre-transfusion hemoglobin level of 8-9 g/dL. Financial constraints denied him access to iron chelation therapy. Seizure episode was managed with anti-convulsants by the local pediatrician.

A review of history at our center revealed a developmentally normal child with no past history of seizures. On examination he had pallor, hemolytic facies (prominent frontal, parietal, malar eminences and depressed nasal bridge), greenish brown complexion secondary to pigmentary skin changes and stunted growth with height and weight <5th centile. Spleen and liver were palpable 6 and 4 cm below the right costal margin, respectively. The cardio-vascular examination was normal and there was no neurological deficit. Chovstek and Trousseau's signs could not be elicited.

Investigations confirmed hemoglobin of 8 g/dL, hypocalcemia 7 mg/dL hyperphosphatemia 7.6 mg/dL and alkaline phosphate of 19 KAU. Parathyroid hormone level was 30.30 pg/mL (normal 7-53 pg/mL); it was lower than expected for the degree of hypocalcemia. Computed tomography scan (CT scan) of the head showed multifocal calcification in the gray white junction of both cerebral hemispheres and basal ganglia (*Fig. 1*). In addition, calcification was seen in bilateral cerebellum including dentate nuclei. EEG had no epileptiform discharges.

Child was put on oral calcium carbonate (1500 mg elemental calcium) and 1,25 dihydroxycholecalciferol (0.25 mg). On subsequent follow-up of 10 months there has been no seizure recurrence.

Discussion

Hypoparathyroidism in thalassemia major patients has been proposed to be a consequence of iron deposition in parathyroid gland or due to suppression of parathyroid hormone secretion induced by resorption secondary to chronic anemia. Hypocalcemia leading on to tetany, hyperreflexia and seizures is the most common manifestation of hypoparathyroidism. Other biochemical abnormalities include raised phosphate level, near normal alkaline phosphate and normal or low parathyroid

levels(1,2). The propositus had all these abnormalities. He had not received any iron chelation therapy whilst receiving over 200 red cell transfusions that resulted in iron overload state.

Basal ganglia calcification in hypoparathyroidism has been well documented; however, anecdotal experience of intracranial calcification in individuals with thalassemia is available(3). Karimi, *et al.* reported 40% (8 out of 20) of their thalassemia patients with hypoparathyroidism had intracranial calcification. All cases were in their second decade of life and had no association between the severity of hypoparathyroidism and extent of intracranial calcification(4).

The proposed pathogenesis for metastatic calcification in hypoparathyroidism is from decreased reservoir function of the bone for the absorbed calcium and phosphate from the intestine leading on to extra osseous calcification(5). In the basal ganglia this possibly results from a degenerative vascular process in extra-pyramidal system initiated by the deposition of calcium crystals(6).

The problems related to calcium metabolism are uncommon in well-chelated transfusion dependent

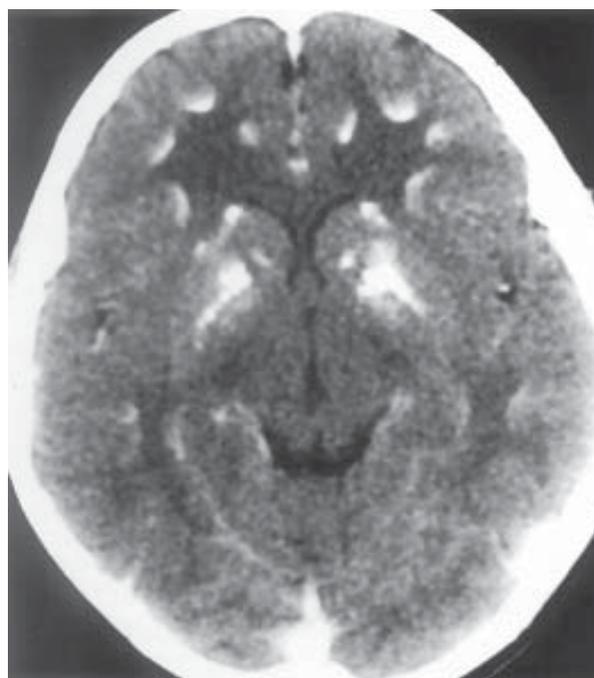


Fig. 1. CT scan showing multifocal calcification

thalassemia patients; however, it would be desirable that unchelated children older than 12 years should have their serum calcium and phosphate levels estimated at regular intervals of 3-6 months. Supplemental therapy with oral calcium and active form of vitamin D would preclude the occurrence of hypocalcaemia related complications. Although routine neuroimaging (CT scan) cannot be recommended, it should be done in children with neurological dysfunction.

Contributors: SV and RKM managed the patient; AKS reported the CT scan, SV reviewed the literature and RKM supervised the same.

Funding: None.

Competing interests: None.

REFERENCES

1. Costin G, Kogut MD, Hyman CB, Ortega JA. Endocrine abnormality in thalassemia major. *Am J Dis Child* 1979; 133: 497-502.
2. De Sanctis V, Vullo C, Bagni B, Chiccoli L. Hypoparathyroidism in β -thalassemia major. Clinical and laboratory observation in 24 patients. *Acta Hematol* 1992; 88: 105-108.
3. Zafeiriou D, Athanasiou M, Katzos G, Economou M, Kontopoulos E. Hypoparathyroidism and intracranial calcification in b-thalassemia major. *J Pediatr* 2001; 138: 411.
4. Karimi M, Habibzadeh F, De Sanctis V. Hypoparathyroidism with extensive intracranial calcification in patients with b-thalassemia major. *J Pediatr Endocrinol Metab* 2003; 16: 883-886.
5. Kurz P, Monier-Faugere MC, Bognar B, Werner E, Roth P, Vlachoianis J, *et al* Evidence for abnormal calcium homeostasis in patients with adynamic bone disease. *Kidney Int* 1994; 46: 855-861.
6. Goel A, Bhatnagar MK, V ashisha A, Verma NPS. Hypoparathyroidism with extensive intracranial calcification: a case report. *Postgrad Med J* 1994; 70: 913-915.

Suprasellar Arachnoid Cyst with Delayed Puberty

**Raj Kumar
Namit Singhal**

A 15-year-old female presented with primary amenorrhea and delayed onset of secondary sexual characteristics. Earlier she was operated for endoscopic third ventriculostomy (ETV) for a tense suprasellar arachnoid cyst with obstructive hydrocephalus. MRI revealed recurrence of hydrocephalus. Hormonal levels were suggestive of hypogonadism and deficiency of growth hormone. She was operated for fenestration of cyst. In this case, suprasellar arachnoid cyst presented with delayed puberty, which is unusual.

Key words: *Delayed puberty, Suprasellar arachnoid cyst.*

Arachnoid cysts constitute approximately 1% of intracranial masses, with 50-60% occurring in middle cranial fossa. Other locations include suprasellar/chiasmatic cistern, cerebello pontine

angle (11%), quadrigeminal plate cistern (10%), in relationship to vermis (9%) and prepontine/interpeduncular cistern (3%). Most arise as developmental anomalies. Arachnoid cysts often are an incidental finding on imaging. The most common associated clinical features include headache, calvarial bulging, intracranial hypertension, cranio-megaly, developmental delay, visual loss and seizures. Arachnoid cysts may cause a wide spectrum of endocrinological disorders, most common being precocious puberty and GH deficiency. We report an interesting case of suprasellar arachnoid cyst presenting with delayed puberty and physical developmental delay.

From the Department of Neurosurgery, Sanjay Gandhi Postgraduate Institute of Medical Science, Lucknow 226 014, U.P., India.

Correspondence to: Dr. Raj Kumar, Associate Professor, Department of Neurosurgery, Sanjay Gandhi Postgraduate Institute of Medical Science, Lucknow 226 014, U.P., India. E-mail: rajkumar@sgpgi.ac.in

Manuscript received: March 2, 2007;

Initial review completed: June 1, 2007;

Revision accepted: June 28, 2007.