References

1. Virchow R. Zwergenkind. Zeitschrift fur Ethnologie, Berlin, 1882, 14: 215.
2. Seckel HPG. In: Bird-headed Dwarfs: Studies in Developmental Anthropology Including Human Proportions. Eds: Springfield C, Thomas C. S. Karger, Basel-New York, 1960.
3. Black J. Low birth weight dwarfism. Arch Dis Child 1961; 36: 633-644.
4. Harper RG, Orti E, Baker RK. Bird-beaded dwarfs (Seckel's syndrome). A familial pattern of developmental, dental, skeletal, genital, and central nervous system anomalies. J Pediatr. 1967; 70: 799-804.
5. McKusick VA, Mahloudji M, Abbott MH, Lindenberg R, Kepas D. Seckel's bird-headed dwarfism. N Engl J Med. 1967; 277: 279-286.
6. Hayani A, Suarez CR, Molnar Z, LeBeau M, Godwin J. Acute myeloid leukaemia in a patient with Seckel syndrome. J Med Genet. 1994; 31: 148-149.
7. Capovilla G, Lorenzetti ME, Montagnini A, Borgatti R, Piccinelli P, Giordano L, et al. Seckel's syndrome and malformations of cortical development: report of three new cases and review of the literature. J Child Neurol. 2001; 16: 382-386.
8. Rubbini L, Preti P, Sudanese A, Brillante C. Seckel's syndrome: presentation of one case associated with Legg-Calve-Perthes disease. Chir Organi Mov. 1991; 76: 193-195
9. Aslan Y, Aynaci FM, Baki A, Soylu H, Harova G. Seckel's syndrome associated with congenital cystic adenomatoid malformation of the lung. Acta Paediatr. 1995; 84: 839-840.

From : Department of Pediatric Medicine, NRS Medical College, Kolkata, West Bengal, India.

Address for Correspondence: Dr Tryambak Samanta, 15/D, Lenin Sarani, Serampore, Dist Hooghly, WB-712203. Email: ts_ped@rediffmail.com

E-published : February 2010

LETTER TO EDITOR (VIEWERS CHOICE)

HYPOHIDROTIC ECTODERMAL DYSPLASIA

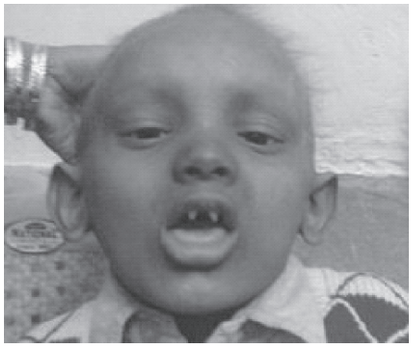
Neerupam Gupta, Shirin Nomani**, Vikram Gupta***, Sanjeev Kumar Digra*****

Key Words: Ectodermal Dysplasia, Hypohydrotic, Anodontia

We report two male sibs one 6 years and another 3 years of age from a nonconsanguineous married Hindu family. Both were admitted in department of Pediatrics, SMGS hospital, Govt Medical College, Jammu with history of high grade fever and cough of short duration with a background history of frequent rise of body temperature since early infancy. Absence of sweating even in hot summer, lack of hair and abnormal dentition were associated complaints. Family history revealed healthy parents with these two sons without history of similar illness in relatives. Clinical examination revealed thin strands of silky hair, sparse eyelash, frontal bossing, depressed nasal bridge and dry skin but normal nails in both of them. Elder brother had only two conical upper incisor teeth (Figure 1) but the younger had one. Systemic examinations revealed no abnormality except for few crackles in lungs. Skin biopsy of both the brothers showed thinned out epidermis, absence of sweat glands and pilosebacious unit in dermis suggestive of hypohidrotic (anhidrotic) Ectodermal dysplasia. Other investigations were nonconclusive. The chest infection in both of these sibs responded to appropriate antibiotics and were discharged after 15 days of hospital stay with advice to avoid high ambient temperatures as far as possible.

Ectodermal dysplasia is a heterogeneous group of disorders characterized by a constellation of findings involving defects of two or more of the following : teeth, skin, and appendageal structures including hair, nails, and eccrine and sebaceous glands. Although 170 ectodermal dysplasias have been described, the majority are rare and only 30 have been genetically defined (1). Hypohidrotic ectodermal dysplasia is also known as anhidrotic ectodermal dysplasia (EDA I) and Christ-Siemens-Touraine syndrome (2). Hypohidrotic ectodermal dysplasia was described as early as 1848 by British physician J. Thurnam (3). The incidence has been reported to be 1 per 10,000 to 1 per 100,000 live births (4). Hypohidrotic ectodermal dysplasia is manifested as a triad of defects: partial or complete absence of sweat glands, anomalous dentition and hypotrichosis. Anodontia or hypodontia with widely spaced, conical teeth is a constant feature (1). Episodes

Figure 1: Sparse hair and eyelashes with only 2 conical incisor teeth



of high fever in warm environment, dry skin, sparse hypopigmented hair, frontal bossing, flattened nasal bridge are some of other features. Poor development of mucous glands in the respiratory tract may result in increased susceptibility to respiratory tract infections. Treatment of these children include protecting them from high ambient temperatures and early dental evaluation so that prostheses can be provided. Artificial tears should be used to prevent the damage to cornea. Wigs may be advised to improve appearance. (1) Both the brothers reported here had the classical triad along with other clinical features of hypohidrotic (anhidrotic) ectodermal dysplasia which was confirmed by skin biopsy. The mode of inheritance of hypohidrotic ectodermal dysplasia is X-linked recessive with full expression only in males; however, an autosomal recessive mode of inheritance may be operative in some families (1,5).

REFERENCES

1. Gray LD. Ectodermal Dysplasia. In: Nelson Textbook of Pediatrics. Behrman RE, Kliegman RM, Jenson HB, editors. 16th edn. W.B.Saunders Company 2000; 1974-1975.
2. Smagrupps Centrum - the Swedish Information Center for Rare Diseases- produced and edited this information material. Date of publication: 2005-05-03 Version: 2.1 Article number: 2002-110-10
3. Priolo M, Lagana C. Ectodermal dysplasias: a new clinical-genetic classification. J Med Genet. 2001; 38: 579-585
4. Sepulveda W, Sandoval R, Carstens E, Gutierrez J, Vasquez P. Hypohidrotic ectodermal dysplasia: prenatal diagnosis by three-dimensional ultrasonography. J Ultrasound Med. 2003; 22: 731-735.
5. Munoz F, Lestringant G, Sybert V, Frydman M, Alswaini A, Frossard PM, Jorgenson R, Zonana J. Definitive evidence for an autosomal recessive form of hypohidrotic ectodermal dysplasia clinically indistinguishable from the more common X-linked disorder. Am J Hum Genet. 1997; 61: 94-100.

From : Sub-Distict Hospital, Ramban, Jammu & Kashmir*, Ayush Hospital, Bhubaneswar, Orissa**, Ascoms, Jammu, Jammu & Kashmir***, Department Of Pediatrics, Government. Medical College, Jammu, Jammu & Kashmir

Address for Correspondence: Dr. Neerupam Gupta, H.No-35,Sector-7, Trikuta Nagar, Jammu. Mob:09419286878

E-published : February 2009

LETTER TO EDITOR (VIEWERS CHOICE)

VENTRICULOPERITONEAL SHUNT CATHETER MIGRATION THROUGH UMBILICUS - A RARE COMPLICATION

Dipankar Sarkar, Shruti Sarkar

A 9 month old baby was brought to our outpatient department with the complaint of a plastic tube protruding out of abdomen through umbilicus for a few days. The baby had undergone Ventriculoperitoneal shunt surgery nearly 4 months back for congenital hydrocephalus in another hospital. He was born at term by caesarean section and was diagnosed to have congenital hydrocephalus soon after birth. The baby had recovered well from surgery, was feeding well on demand and thriving. On examination the baby looked well and the VP shunt catheter was seen protruding out of umbilicus (Figure 1). On pressing the shunt reservoir behind the ear, fluid started dribbling out of the tube protruding through the umbilicus confirming the protruding end to be the distal end of the VP shunt. Systemic examination apart from this did not reveal any other abnormality. The infant along with his parents had come from a distant village. The parents were explained in detail the occurrence of this complication and the need for correction by surgery. They declined corrective surgery and were lost to subsequent follow up.

Migration or protrusion of the ventriculoperitoneal shunt catheter to various sites is not very uncommon and has been reported in several case reports. Cases of shunt migration into stomach (1), bowel (2), liver (3), chest (4), jugular vein (5), anus (6), vagina (7), and scrotum (8) have been reported. In 1973 Adeloye et al reported a 9 month old baby with shunt migration through umbilicus within 2 months after surgery (9). Another case has been reported from Srinagar, India in 2000 where an 18 month old child presented with low grade fever, irritability, abdominal pain along with the distal end of VP shunt coming out through the umbilicus (10) .This particular complication can be explained on anatomical basis. Umbilicus is a centrally situated scar

Figure 1: Distal end of VP shunt protruding from the umbilicus