usefulness of computed tomography. Eur Respir J. 1990; 3: 728-731.
9. Megran DW, Scheifele DW, Chow AW. Odontogenic infections. Pediatr Infect Dis. 1984; 3: 257-265
10. Gridley J, Franaszek J. Ludwig's angina and pneumococcal sepsis. J Emerg Med. 1986; 4: 201-204.
11. Quinn P, Guernsey LH. The presentation and complications of odontogenic septic shock. Report of a case. Oral Surg Oral Med Oral Pathol. 1985; 59: 336-339.
12. Standring S. Gray's Anatomy Ch 29, Face & Scalp, 39th edn Elsevier, Churchill Livingstone, Edinburgh, 2005: 497-517.
13. Kissig B, Zierz S. Isolated pareses of the terminal branches of the facial nerve: clinical findings, etiology and differential diagnosis. Fortschr Neurol Psychiatr. 2006; 74: 371-376.

From : Department of Pediatrics*, Department of Anatomy**, Department of Oral and Maxillofacial Surgery***, Peoples College of Medical Science & Research Center, Bhopal, India.

Address for Correspondence: Dr. Dipankar Sarkar, Assistant Professor, Department of Pediatrics, Peoples College of Medical Science & Research Center, Bypass Road, Bhopal ,MP 462037. India. Email: dipankarshruti@gmail.com

E-published : February 2010

CASE REPORT

PROTEUS SYNDROME

Rahul Sinha, Chandrashekhar, Kirandeep Sodhi, Y K Kiran

Abstract

Proteus syndrome is a rare hamartomatous condition with multisystem involvement and great clinical variability. A 7yr old girl presented with recurrent falls with progressive maculopapular lesions over face, trunk and limbs, asymmetry of face and calf, scoliosis, lower limb discrepancy and lipoma over lower back suggestive of Proteus syndrome. She is planned for lipoma excision and laser treatment.

Introduction

Proteus syndrome is a rare hamartomatous condition with multisystem involvement and great clinical variability with prevalence of less than 1 per 1,000,000 live births. (1) First reported in 1979 and named Proteus syndrome after Greek god Proteus who could change his shape at will to avoid capture. (2)

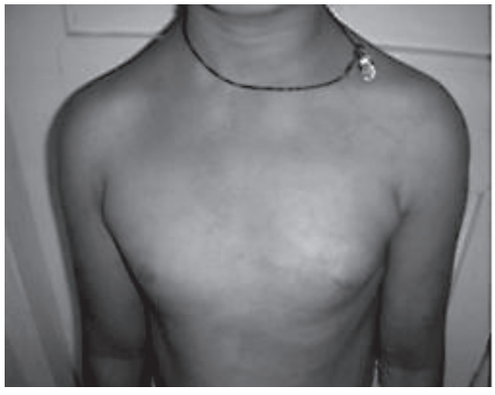
Case Report

A term appropriate for gestational age female baby weighing 3.2 kgs was born to a booked immunized multigravida by normal vaginal delivery. The mother and father were non consanguineous and were aged 23 and 25 years respectively. The baby was detected to have a lump on right lower back with an approximate size 2x2cm with no associated tuft of hair or watery discharge. Mother also noticed there was facial asymmetry with a deviation of angle of mouth to left side with no impaired closure of eyes or drooling of saliva from angles of mouth. There were multiple hyperpigmented lesions over face, trunk and upper limbs. On D2 of life baby developed neonatal jaundice and was managed by double surface phototherapy for 24 hrs. Developmentally child had achieved all milestones in time.

Now at the age of 7yrs the child is brought to our hospital with history of recurrent falls while running and progressive nature of cutaneous lesions. The examination revealed height, weight and head circumference appropriate for age and sex. The cardiopulmonary status was stable. There were multiple hyperpigmented maculopapular lesions of various sizes over face, trunk and limbs which increase in size on crying (picture1). There was asymmetry of face with deviation of angle of mouth to left. There was lipoma on thoracolumbar region measuring 12cm x 10cm .There was asymmetry of calf muscles (left>right), lower limb length discrepancy (left>right by 3.5 cm) and macrodactaly. There was scoliosis to left; however other neurological examination was essentially normal.

X-ray spine revealed scoliosis to left and soft tissue swelling at thoracolumbar region, CECT (chest) was normal; MRI (brain) did not reveal any abnormality. Presently child is under follow up in orthopedic and skin OPD, LASER treatment is planned for cutaneous lesion later on. The lipoma on back is planned for excision.

Picture 1: Multiple maculopapular lesion over trunk and limbs



Discussion

The name, Proteus syndrome, was coined by Wiedemann in 1983 after the Greek sea God, Proteus [1]. As the son of Poseidon, Proteus possessed the ability to transform himself into any shape to avoid prophesying the futures of the mortals who hounded him. Like the Greek God, this syndrome manifests itself in many ways. The most famous patient afflicted with Proteus syndrome was Joseph Merrick, better known as the "Elephant Man" [2]. Proteus syndrome is an uncommon clinical entity characterized by abnormalities in growth (asymmetric overgrowth, increased stature, macrodactyly, soft tissue hypertrophy, elongated neck, macrocephaly), skin (plantar and palmar skin thickening, epidermal nevi, lipomas, lymphangiomas, hemangiomas, cafe au lait spots, varicosities, dermal hypoplasia), musculoskeletal (hemihypertrophy, bony prominences, ankle ankylosis, craniosynostosis, mandibular prognathia, scoliosis, pectus excavatum, thinning of the cortical layer of long bones), eye (ptosis, strabismus, nystagmus, myopia, colobomas, cataracts, epibulbar dermoids, blue sclera), central nervous system (seizures, mental retardation), venous (varicosities, hemorrhoids, dilated superior mesenteric vein [3,4,5,6,7].

Patients afflicted with Proteus syndrome frequently appear normal at birth. Features begin to appear during the first year of life with subsequent progression. The etiology of Proteus syndrome remains unknown. The majority of patients have a normal chromosomal complement. However, it has been suggested that there may be a mosaic somatic mutation affecting the regulation of tissue growth factors, leading to the associated polymorphic characteristics of this disease. The main differential diagnoses are Klippel-Trenaunay Weber syndrome, Bannayan- Riley syndrome (autosomal dominant, macrocephaly, capillary malformation, polyposis coli etc.) [8], encephalocraniocutaneous lipomatosis (characteristic nevus psiloliparus consisted of large, slightly protuberant usually unilateral soft masses on scalp with complete alopecia, skin colored papular eruptions on face with some bony and eye and neurological changes), hemihyperplasia syndrome (multiple lipomas, cutaneous vascular overgrowth). The morbidity and mortality risk of pulmonary cysts in Proteus syndrome is unknown. The mainstays of treatment are the following: Identify serious medical problems early and employ prophylactic and symptomatic treatment. Medical approaches are limited and should be considered in the context of functional improvement. Leg length discrepancy can create a host of secondary morbidities and needs to be addressed by an experienced orthopedist. Macrodactyly can make it difficult for the patient to write, hold objects, dress, eat, or find comfortable footwear. Early recognition of scoliosis may permit nonsurgical attempts to halt progression. - Periodic evaluation is essential for cutaneous and subcutaneous lesions since lipomas and vascular malformations may have local or even systemic effects. Laser treatment is useful for removing cutaneous vascular markings and malformations such as port wine stains and capillary hemangiomas. It is not yet effective for permanently removing café au lait spots or melanin-related hyperpigmentation.

References

1. Wiedemann HR, Burgio GR, Aldenhoff P, Kunze J, Kaufmann HJ, Schirg E. The proteus syndrome Partial gigantism of the hands and/or feet, nevi, hemihypertrophy, subcutaneous tumors, macrocephaly or other skull anomalies and possible accelerated growth and visceral affections. Eur J Pediatr. 1983; 140: 5-12.
2. Tibbles JA, Cohen MM Jr. The Proteus syndrome: the Elephant Man diagnosed. Br Med J (Clin Res Ed). 1986; 293: 683-685.
3. Fay JT, Schow SR. A possible case of Maffucci's syndrome: report of case. J Oral Surg. 1968; 26: 739-744.
4. Happle R, Steijlen PM, Theile U, Karitzky D, Tinschert S, Albrecht-Nebe H, Küster W. Patchy dermal hypoplasia as a characteristic feature of Proteus syndrome. Arch Dermatol. 1997; 133: 77-80.
5. Newman B, Urbach AH, Orenstein D, Dickman PS. Proteus syndrome: emphasis on the pulmonary manifestations. Pediatr Radiol. 1994; 24: 189-193.
6. Nowaczyk MJ, Mernagh JR, Bourgeois JM, Thompson PJ, Jurriaans E. Antenatal and postnatal findings in encephalocraniocutaneous lipomatosis. Am J Med Genet. 2000; 91: 261-266.
7. Proteus Syndrome. In: Jones KL (eds). Smith's Recognizable Patterns of Human Malformation. 4th edition. Philadelphia, PA, W.B. Saunders. 1988, pp. 458-459.
8. Biesecker LG, Happle R, Mulliken JB, Weksberg R, Graham JM Jr, Viljoen DL, Cohen MM Jr. Proteus syndrome: diagnostic criteria, differential diagnosis, and patient evaluation. Am J Med Genet. 1999; 84: 389-395

From : Department of Pediatrics, Command Hospital Air Force, Bangalore

Address for Correspondence: Dr Rahul Sinha, Department of Pediatrics, Command Hospital Air Force, Agaram Post, Old Airport Rd, Bangalore 560007. Email: drrahul_2000@yahoo.com

E-published : March 2010