

Case Report

Rubinstein-Taybi syndrome: a case report.

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Abstract

Rubinstein-Taybi syndrome (RSTS) or Broad Thumb-Hallux syndrome is a very rare autosomal dominant disease of multisystem abnormality including facial dysmorphism, broad thumb and toes, growth retardation, intellectual abnormality, dermatological abnormalities. Here is a case of RSTS with multiple spontaneous keloids and squamous cell carcinoma of a 22 year old man in the department of dermatology and venereology in BSMMU, Dhaka which is the first reported case in Bangladesh.

Keywords: Rubinstein -Taybi syndrome, Spontaneous keloids, Squamous cell carcinoma

Introduction:

Rubinstein and Taybi reported seven cases of this syndrome in 1963 with clinical manifestation of short-broad thumb and great toes, psychomotor retardation, high arched palate and particular facial abnormalities.¹ Rubinstein-Taybi syndrome (RSTS) or Broad Thumb-Hallux syndrome is an autosomal dominant disease with an incidence of 1 in 100000 and mutation in two genes CREBBP and EP300 in 16p13.3 are the genetic basis of this disease and are responsible for cellular proliferation, differentiation, memory and tumour suppression.^{2,3} These two genes encode histone acetyltransferases (HATs) which are transcriptional coactivator of many signaling pathways. Loss of HAT activity produces phenomena of RSTS.⁴ Multisystem abnormality is reported in this syndrome with variable clinical manifestations. Some of the dermatological manifestations include atrophic scars, keloids, hypertrichosis.⁵ Other system involvement includes GIT as reflux, megacolon, hirschprung disease, congenital heart disease, renal anomalies, respiratory problem, skeletal abnormalities, developmental delay, mental retardation and increased chance of tumour formation mainly meningioma and other brain tumours and leukemia.⁴⁻⁶ Diagnosis is based on clinical manifestation and genetic confirmation is found in about

50% of cases. Here, diagnosis is made clinically and genetic test was not done due to unavailability. Herein a case of this very rare syndrome and to the best of our knowledge it is first reported case in Bangladesh.

Case report:

A case of RSTS of a 22 year old man was admitted in dermatology and venereology department of BSMMU, Dhaka, Bangladesh presented with multiple spontaneous keloids in body and verrucous lesions on legs. He is growth retarded and had a history of feeding difficulty in his neonatal period, but his prenatal period was uneventful. At the age of thirteen he noticed spontaneous appearance of keloids in left arm which gradually spreaded to all limbs with increasing in size. Some lesions of both legs became verrucous for last two years. He complained mild occasional itching in the lesions. He had history of developmental delay of milestone with frequent respiratory tract infection in childhood. He also had learning difficulties and discontinued his education at primary level. On examination, he has growth retardation, long eye lashes and thick eyebrows, broad nose, high arched palate, hypodontia, dental caries, telon cusps. He also has broad end of terminal phalanges of thumbs and toes, ulnar deviation of wrists and fingers, multiple atrophic

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scars in face and trunk, keloids of different sizes largest one measuring 10x8cm over right wrist joint and 4 verrucous lesions in legs largest one measuring 15x10cm in right leg and are non tender. Other systemic examination reveals no abnormality. X ray of lumbo sacral region reveals spina bifida at fifth lumbar vertebra and bilateral sacralization. Dental X-ray showed hypodontia (figure-4). Histopathological examination of the keloidal lesion confirms keloid and verrucous growth shows squamous cell carcinoma.

Discussion:

RSTS is a genetic disease of multisystem abnormality with diverse clinical manifestation and occurs sporadically with no sexual predilection. Dermatological manifestation includes hypertrichosis, hirsutism, atrophic scars, keloids, tumour formation, pilomatrixoma, capillary hemangioma, transverse palmar creases, keratosis pilaris, atopic eczema, seborrheic dermatitis, chronic paronychia, abnormal dermatoglyphics, long eyelashes, brown spots in lumber region.⁷⁻¹⁰ Other system involvement includes GIT as reflux, megacolon, hirschprung disease, congenital heart disease, renal anomalies, respiratory problem, skeletal abnormality, developmental delay, mental retardation and increased chance of tumour formation mainly meningioma and other brain tumours and leukemia.¹⁰⁻¹¹ Dental abnormalities are hypodontia, malocclusion of the teeth, talon cusps, dental caries, high arched palate.¹² These signs and symptoms vary among affected individuals. Life expectancy is normal except cases of respiratory infection and cardiac defects and malignancy.

This patient had feeding difficulty in infancy which is a feature of RSTS and found in 76.3% of patients and he is growth retarded which is consistence with other patient of RSTS of 92.7%. Other features of this patient is mental retardation which is 98.5% of RSTS, heavy and highly arched eyebrow which is 69.6%, long eyelashes 57.8%, dental anomalies 67.4%, thumbs and first toes with broad terminal phalanges 99.0%, other fingers with broad terminal phalanges 74.0%, keloid 23% found in this syndrome¹³. He has atrophic scars in face and trunk, deviation of wrist and fingers to ulnar side which are also reported in RSTS patients. Tumours reported so far in RSTS patients are medulloblastoma, neuroblastoma, meningioma, oligodendroglioma, pheochromocytoma, nasopharyngeal rhabdomyosarcoma, leiomyosarcoma, seminoma, embryonal carci-

noma, odontoma, choriostoma, dermoid cyst and pilomatrixoma.^{10,14} In this case squamous cell carcinoma (SCC) in his legs is a new finding in RSTS patient.

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Figure-1 : Multiple kloids



Figur-3: squamous cell carcinoma.

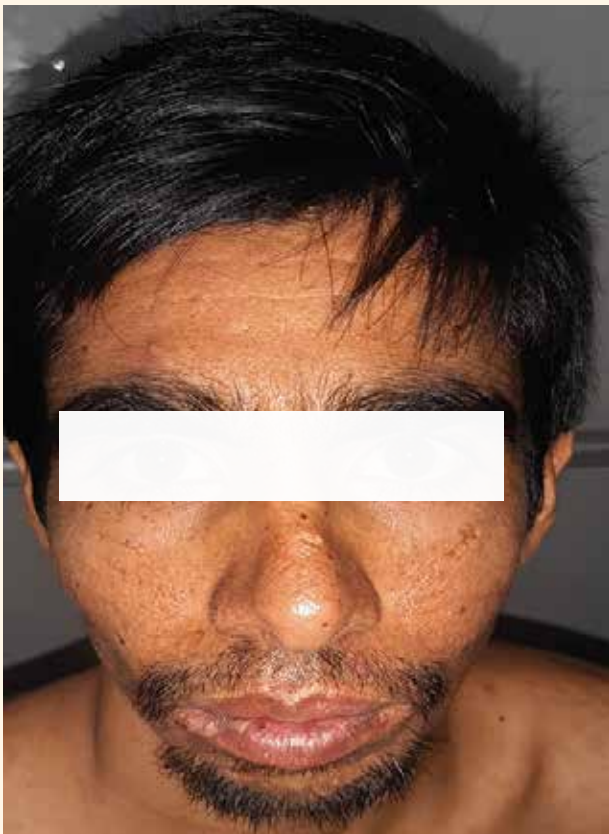


Figure-2: Thick eyebrows, bignose, atrophic scarring



Figure-4: Dental Xray