Rubinstein-Taybi Syndrome: A Rare Case Report

Sir,

The Rubinstein-Taybi syndrome (RTS) is a rare neurodevelopmental disorder characterised by mental retardation, microcephaly, specific facial characteristics, broad thumbs and big toes. Diagnosis is often difficult due to its rarity and non-familiarity with the classical features of this syndrome. Cutaneous findings such as capillary malformations, hirsutism, keloid formation, and pilomatrixomas have been described previously in association with RTS. We report a case of RTS with multiple keloids along with classical features of the disorder in a 30-year male.

A 30-year-old male born out of non-consanguineous marriage presented to our department with multiple dark flat shiny lesions over chest and both feet since 10 years. The lesions were associated with occasional itching. There was no obvious history of preceding trauma, injections or burns on the site of the lesions. He also had a history of difficulty in speech and poor learning abilities. No similar history was present in any of the family members.

Thorough physical examinations including anthropometric measurements were within normal limits. Rest of the systemic examination including detailed neurological, genitourinary, audiological and skeletal examination was unremarkable.

On cutaneous examination, patient had dysmorphic face with micrognathism, microstomia, microtia and low set ears, thick eyebrows and prominent beaked nose [Figure 1]. Multiple well-defined hyperpigmented shiny keloidal scars were present over chest and bilateral feet [Figure 2a, b]. Other abnormalities included the presence of broad thumbs and halluces [Figure 3a, b]. Oral examination showed the presence of high-arched palate and malpositioned teeth [Figure 4a, b].

Psychiatric evaluation was suggestive of delay in linguistic, social and intellectual milestones which was evident as difficulty in reading, writing, calculation and poor conversation skills.

Based on history and clinical examination, differential diagnoses of RTS, Goemanni syndrome, Dubowitz syndrome, Cornelia de Lange syndrome and Floating harbour syndrome were considered [Table 1].

Routine haematological investigations were normal. Chest X-ray and electrocardiography revealed no abnormality. X-ray of feet was suggestive of abnormal soft tissue swelling with radial deviation of hallux [Figure 5a, b], while X-ray of wrist revealed no significant abnormality. Orthopentogram was suggestive of generalised microdontia and absence of lingual fossa (Talon Cusps). Ophthalmological examination showed

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aphakia. Skin biopsy from lesion over chest revealed increased number of thick collagen fibres arranged in bundles in the reticular dermis, which was consistent with keloid. Genetic analysis could not be done in this patient due to lack of financial resources.

Based on the history, clinical features and investigations a diagnosis of RTS was made. Patient was referred to psychiatrist for behavioural therapy and ophthalmologist for correction of aphakia.

RTS also known as Broad Thumb-Hallux syndrome was first described by Michail et al. in 1957 and then a case series of seven cases was reported by Rubinstein and Taybi in 1963.[1] It is caused by a mutation in either CREB-binding protein (CBP) or E1A-binding protein present in the chromosome 16p13.3. The occurrence of disease is mostly sporadic and its incidence has been reported to be 1:100,000-125,000 at birth. Additional features can include eye abnormalities, heart and kidney defects, dental problems and obesity.[3] Dermatological manifestations which have been most frequently described include hirsutism and capillary malformations. Other unusual manifestations encountered in RTS includes keloids, thick and high-arched eyebrows with long eyelashes, brown spots in the lumbar region, transverse

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| Characteristic feature | Rubinstein-Taybi syndrome | Cornelia de-Lange syndrome[7] | Floating-Harbor syndrome[8] | Dubowitz syndrome[9] | Goeminne syndrome[10] |
|-----------------------|--------------------------|-------------------------------|----------------------------|----------------------|-----------------------|
| Inheritance           | AD/Sporadic              | Sporadic/AD                   | AD                         | AR                   | XLinked               |
| Growth retardation    | +/-                      | +                             | +                          |                      |                      |
| Mental retardation    | +                        | +                             | +                          |                      |                      |
| Facial dysmorphism    | +                        | +                             | +                          |                      |                      |
| Skeletal abnormalities| +/-                      | +                             | +                          |                      |                      |
| Behavioural problems  | +/-                      | +                             | +                          |                      |                      |
| Broad thumb-halluces  | +                        | -                             | -                          |                      |                      |
| Spontaneous keloids   | +                        | -                             | -                          |                      |                      |
| Other dermatological manifestations | Hirsutism, capillary malformation, abnormal dermatoglyphics, spatulate nails, chronic paronychia, supernumerary nipples, keratosis pilaris and atopic eczema | Hirsutism, cutis marmorata, bluish discolouration of skin | Atopic eczema, sparse hair, multiple nevi | Multiple pigmented nevi |
palmar creases, abnormal dermatoglyphics, spatulate nails, chronic paronychia, supernumerary nipples, keratosis pilaris and atopic eczema.[4] Keloids in RTS were first described by Kurwa.[5] Diagnosis mainly depends upon the presence of classical clinical features of the syndrome. Treatment requires a multidisciplinary approach. Treatment of extensive keloids is often disappointing. These patients are also at increased risk of developing meningioma, other brain tumours and leukaemia, thus early diagnosis and recognition of malignancy can aid in successful life saving interventions.[6]

We report a case of 30-year-old male with RTS with presence of multiple extensive keloids without any systemic manifestation. There is paucity of Indian literature describing this entity. Patients presenting with multiple spontaneous keloids, facial dysmorphism, broad thumbs or halluces and history of mental retardation should be investigated in detail, keeping a possibility of RTS in mind. The patients are prone to systemic malignancies and metastases which should be screened for and detected at the earliest so that prompt management can be initiated.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

References

1. Hennekam RC, Lommen EJ, Strengers JL, Van Spijker HG, Jansen-Kokx TM. Rubinstein-Taybi syndrome in a mother and son. Eur J Pediatr 1989;148:439-41.
2. Bansal S, Relhan V, Garg VK. Rubinstein-Taybi syndrome: A report of two siblings with unreported cutaneous stigmata. Indian J Dermatol Venereol Leprol 2013;79:714-7.
3. Hennekam RC. Rubinstein-Taybi syndrome. Eur J Hum Genet 2006;14:981-5.
4. Selmanowitz VJ, Stiller MJ. Rubinstein-Taybi syndrome. Cutaneous manifestations and colossal keloids. Arch Dermatol 1981;117:504-6.
5. Kurwa AR. Rubinstein-Taybi syndrome and spontaneous keloids. Clin Exp Dermatol 1979;4:251-4.
6. Stevens CA, Carey JC, Blackburn BL. Rubinstein-Taybi syndrome: A natural history study. Am J Med Genet Suppl 1990;6:30-7.
7. Chawla C, Rao PK, Kini R, Shetty D. Cornelia de-Lange syndrome - A case report. J Indian Acad Oral Med Radiol 2018:30-92-5.
8. Robinson PL, Shohat M, Winter RM, Conte WJ, Gordon-Nesbitt D, Feingold M, et al. A unique association of short stature, dysmorphic features, and speech impairment (Floating-Harbor syndrome). J Pediatr 1988;113:703-6.
9. Stewart DR, Pernov A, Johnston JJ, Sapp JC, Yeager M, He J, et al. Dubowitz syndrome is a complex comprised of multiple, genetically distinct and phenotypically overlapping disorders. PLoS One 2014;9.
10. Fryns JP, Gevers D. Goeminne syndrome (OMIM 314300): Another male patient 30 years later. Genet Couns 2003;14:109-11.