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ADVANCES IN PEDIATRIC RESEARCH

High myopia, V-pattern esotropia and bilateral nasolacrimal duct obstruction in a child with Rubinstein–Taybi syndrome

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Abstract

We report a female child of Indian origin with Rubinstein–Taybi syndrome with ocular features of convergent strabismus (V-pattern esotropia), bilateral high myopia and congenital nasolacrimal duct obstruction. We present the details of the management of this case and the final outcome. This case report not only highlights the variability of the ocular features, but also the importance of an ocular examination in patients with Rubinstein–Taybi syndrome.

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Introduction

Rubinstein–Taybi syndrome (RTS) is a genetic multisystem disorder characterized by distinct facial dysmorphism, broad thumbs and toes, growth retardation and learning difficulties. The most frequent RTS-associated eye anomalies reported in the literature are strabismus, refractive errors, lacrimal duct obstruction, corneal abnormalities, congenital glaucoma, congenital cataract, and colobomata [1,2].

Case Report

An 11 year-old female child born of a non-consanguineous marriage presented with complaints of deviation of the eyes with watering and discharge since birth. She had undergone cardiac surgery for patent ductus arteriosus (PDA) at an early age. Her parents gave a history of delay in her global milestones. She had never been prescribed spectacles.

Clinical evaluation revealed poor understanding and speech with gross developmental delay. Her height, weight and head circumference were below the third percentile for her age. Head-to-toe examination revealed broad and short terminal phalanges of her thumbs (Fig. 1A) as well as the big toes (Fig. 1B), an antimongoloid slant of the palpebral fissues with long eyelashes, beaked nose and an overhanging columnella (Fig. 1C), a small mouth with dental crowding and talon cusps on the premolars, and generalized hirsutism, especially on the back (Fig. 1D). She had undergone cardiac surgery for ligation of patent ductus arteriosus (PDA) at an early age. Analysis of chromosomes taken from a blood sample revealed a normal female karyotype.

Visual testing revealed a good ability to fix on and follow a light source with either eye, but she was inattentive to Teller acuity card (TAC) testing. Ophthalmic examination by retinoscopy revealed refractive errors of -22.50DS in the right eye and -15.50DS/+1.50DC at 90 in the left eye. The child's head posture showed chin depression. Further orthoptic evaluation revealed an esotropia (Fig. 2A) with good alternation. The Hirschberg and Krimsky test revealed 45 prism diopters of esotropia in the

primary position, which decreased in upgaze, and there was bilateral inferior oblique muscle overaction suggestive of a V-pattern. Mucopurulent regurgitation was observed when pressure was applied over the sac region in both eyes; the anterior segment was normal in both eyes. Examination under anesthesia (EUA) revealed myopic discs with large cups and annular crescents, and myopic degeneration. No treatable lesion was found in the periphery.

The child first underwent simultaneous probing with laser dacryocystorhinostomy (DCR) in both eyes, followed by extraocular muscle surgery. An 11mm recession of the medial rectus muscle from the limbus, with symmetrical inferior oblique myectomy, was performed in each eye with an acceptable postoperative alignment at 1.5 years follow-up (Fig. 2B). At that visit, the patient's best-corrected visual acuity in each eye was 20/250 with 80% reliability, as checked with TAC.

Written consent was obtained from the parents for this study.

Discussion

In 1963, Dr. Jack Rubinstein and Dr Hooshang Taybi first described the syndrome causing broad thumbs and toes, and facial abnormalities, which bears their names today. This is a genetic, multi-system disorder associated with mental retardation, postnatal growth retardation, characteristic facies of a beaked nose with overhanging nasal columnella, broad thumbs and toes, and other congenital anomalies. Western literature estimated prevalence rate at birth to be 1:100000 to 1:125000 [3].

Genetic studies of RTS have revealed either rearrangements or submicroscopic deletions on chromosome 16 (16p13.3), detectable by fluorescence in situ hybridization (FISH) [4], or mutations in CREB-binding protein (CBP) or E1A-binding protein (p300) in approximately 50% of cases [5]. However, in the remaining cases the diagnosis has not been confirmed by molecular genetic tests, indicating that there is genetic heterogeneity.

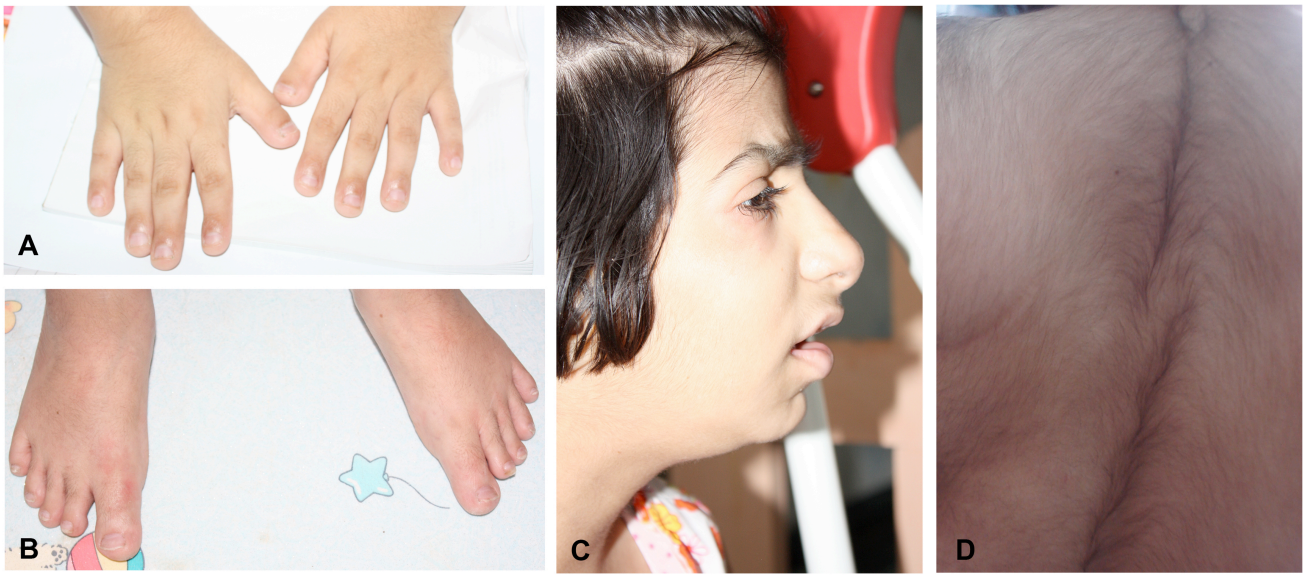

Figure 1. Clinical photographs of an RTS patient showing characteristic broad terminal phalanges of thumbs and big toes (A and B), beaked nose (C) and hirsutism (D)

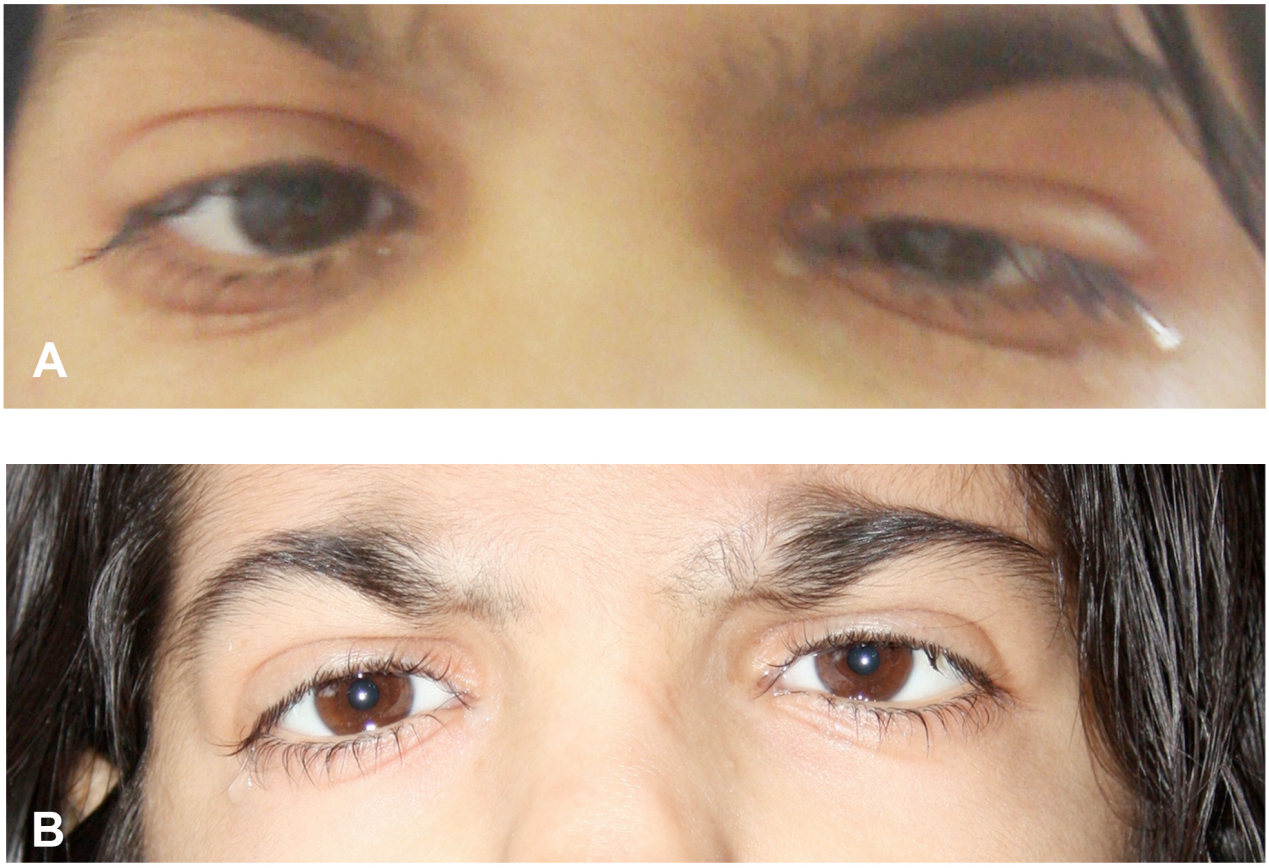

Figure 2. Appearance of the eyes before squint showing inward squint (A)
Appearance of the eyes after squint surgery showing straight eyes (B)

A diagnosis of RTS is usually made based on presentation of clinical features. The characteristic clinical features include growth retardation, generalized hirsutism, typical facial and dental anomalies, broad and short terminal phalanges of the thumbs and big toes, and mental retardation [6]. Systemically, congenital cardiac defects are also known to occur in one third of cases [2]. The most common congenital heart defects described in this condition are PDA and ventricular septal defects (VSD). Our patient presented with all of the features suggestive of RTS, and she had PDA for which she had undergone surgery in the past.

Rubinstein–Taybi syndrome is associated with multiple ocular abnormalities involving all the segments of the eye. The most frequently observed eye anomalies are strabismus [2], nasolacrimal duct problems, cataracts, retinal abnormalities and refractive errors. Other findings include congenital glaucoma, ptosis, and nystagmus [1]. After searching PubMed, we can confirm this is the first case report from India describing these ocular features.

Strabismus is a frequent finding in Rubinstein–Taybi syndrome. In a study of 571 affected individuals in 1990, Rubinstein reported that strabismus was present in 71% of cases, followed by refractive errors in 56% [6]. Similarly, Van Genderen et al. studied 24 Dutch patients and found 17 patients to have strabismus. Of these, only two patients had convergent squint. In addition, high myopia of more than -6 diopters was seen in six of the 24 cases; three of these cases had strabismus and all were divergent [1]. Our patient had a V-pattern esotropia. Myopia with convergent squint has not been reported in RTS before.

Our patient had myopia of -22.50 DS in the right eye and -17.00DS/-1.50DC at 180 in the left eye. Following a report of retinal detachment in RTS patients, a complete retinal examination and regular fundoscopy is mandatory in patients with RTS and high myopia [7].

Patients with Rubinstein–Taybi syndrome tend to have lacrimal duct abnormalities; in 25% of cases, these take the form of nasolacrimal duct obstruction [1] and duct anomalies in 37% [6] of these patients. Recurrent attacks of acute dacryocystitis and watering have been described in previous cases. Our case had nasolacrimal duct obstruction with mucopurulent regurgitation but no acute attacks.

The association of high myopia with convergent strabismus and congenital nasolacrimal duct obstruction contributes to the evidence that there is a high degree of variability of ocular features in RTS patients. It also highlights the need for a complete ocular examination so as to identify these findings and provide early intervention if needed.

This case is reported to highlight variable ocular presentation, and to present our findings following strabismus surgery in a clinically diagnosed case of Rubinstein–Taybi syndrome. The occurrence of these features has hitherto been unreported in an Indian family.

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