Megalocornea in a Case with Rubinstein-Taybi Syndrome

Rubinstein-Taybi Sendromlu Bir Olguda Megalokornea

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ABSTRACT

Rubinstein-Taybi syndrome (RTS) is a rare congenital disorder with distinctive findings including skeletal abnormalities, postnatal growth deficiency, intellectual disability, various dysmorphic features, and ocular abnormalities such as corneal pathologies, glaucoma, strabismus, optic atrophy, and refractive errors. Herein, a girl diagnosed with RTS and concomitant bilateral simple megalocornea is presented.

Keywords: Glaucoma, megalocornea, Rubinstein-Taybi syndrome.

ÖZ

Rubinstein-Taybi sendromu (RTS), iskelet anomalileri, postnatal gelişme geriliği, zeka geriliği, çeşitli dismorfik bulgular ve korneal patolojiler, glokom, şaşılık ve kırma kusurları gibi oküler anormallikleri içeren karakteristik bulguları olan nadir bir konjenital hastalıktır. Bu yazıda, RTS ile birlikte eşlik eden bilateral basit megalokornea tanılı bir kız olgu sunulmaktadır.

Anahtar kelimeler: Glokom, megalokornea, Rubinstein-Taybi sendromu.

INTRODUCTION

Rubinstein-Taybi syndrome (RTS) is a rare congenital neurodevelopmental syndrome, first described in 1957 by Michail et al.[1], but re-explained as a syndrome by Rubinstein and Taybi in 1963.[2] Short stature, moderate learning difficulties, broad first toes and broad thumbs are characteristic features of RTS, and the incidence at birth is approximately 1/125000. Congenital heart defects, skin abnormalities as well as mental and motor retardation may also be seen in such cases. The major craniofacial features are mild micrognathia, downslanting palpebral fissures, highly arched eyebrows, beaked nose with broad nasal bridge.[2-4] Ophthalmologic findings including nasolacrimal duct obstruction, ptosis of eyelids, corneal abnormalities, congenital glaucoma, and refractive errors can be seen in cases with RTS.[3-7] Herein, the ocular findings of a Turkish girl diagnosed as RTS and concomitant bilateral megalocornea without glaucoma is presented.

CASE REPORT

Presented case is a 4-month-old girl and the first child of healthy unrelated parents. Her mother's pregnancy was resulted in cesarean section because of placenta previa at a gestational age of 39 weeks. Although the double and triple tests weren't performed, obstetric ultrasonography were ordinary in every visit. She was born with a birth weight of 3050 grams and her vital signs were normal as a newborn. Owing to the presence of some dysmorphic features at birth, she was subsequently referred to a children's hospital for further investigation.

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Her initial physical examination revealed cleft lip, mild micrognathia, hypertelorism, mongoloid slant of the eyes, highly arched eyebrows, beaked nose with broad nasal bridge, short neck, sacrococcygeal dermal sinus, broadening of distal phalanges of hands and malalignment of foot fingers (Figures 1 and 2). Her blood and urine tests were normal, and cranial ultrasonography was ordinary. Abdominal ultrasonography was irrelevant except for left hydroureteropyonephrosis. Her vertebral and long bone x-ray images were uneventful except slightly narrowing of iliac wings. Sacral ultrasonography revealed a fistula tract between skin and spinal canal with a diameter of 1.2 mm at its largest point, however surgical correction was not recommended as no purulent discharge was evident. For her dysmorphic body features she was consulted to Genetics Department where she had the diagnosis of RTS. Since no microbiological agent isolated from her conjunctival, blood, urine and umbilical discharge cultures, her general situation was good and she started to gain weight, she was discharged from hospital with follow-up instructions.

Figure 1. *Image demonstrating facial features including mild micrognathia, highly arched eyebrows, hypertelorism, mongoloid slant of the eyes, and beaked nose with broad nasal bridge.*

Figure 2. *Image of right hand demonstrating the broadening of distal phalanges.*

At the age of 4 months, her parents noted slightly increased size of her eyes and applied to an ophthalmology clinic where she was diagnosed as bilateral buphthalmos and congenital glaucoma and referred to our center. On our initial examination, visual acuity could be tested with bilateral fixation pattern according to the younger age, and she had central, steady and maintained fixation in either eye. She had megalocornea in both eyes and slit-lamp biomicroscopy revealed no other pathological findings. Intraocular pressure (IOP) was normal with palpation. Dilated fundoscopy revealed a normal macula and peripheral retina with no cupping in both eyes. A detailed ophthalmologic examination was also performed under general anesthesia in an operating theatre. Short after sedation with midazolam, IOP scores were measured as 8 mmHg and 7 mmHg with Perkins tonometry in the right and left eyes, respectively and 10 mmHg with Reichert TonoPen in both eyes. Corneal diameter was found as 12.5 mm bilaterally and central corneal thickness of right and left eyes were measured as 673 mm and 680 mm, respectively. Axial length of the right eye was 18.90 mm, while it was measured as 18.75 mm in the fellow eye. Fundoscopy revealed no cupping with hypopigmented retina in both eyes. The case was diagnosed as bilateral simple megalocornea since no signs of glaucoma was present in ocular examination (Figure 3 a and b). The follow-up visits scheduled on six-month intervals have been ongoing for two years, and any signs of glaucoma including IOP rise as well as glaucomatous cupping have not been established yet. In the last follow-up visit performed at the second year of age, visual acuities evaluated with Teller visual acuity cards were 13.0 cyc/cm in each eye; anterior segment examination was unremarkable except with megalocornea (corneal diameter

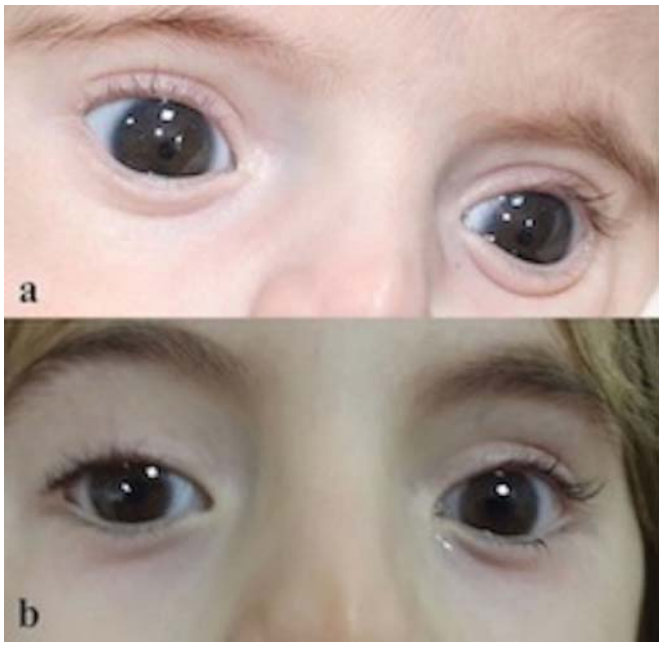

Figure 3. a. *Image of eyes at referral* / **b.** *Image of eyes at the last follow-up visit.*

was 13.0 mm bilaterally and IOP scores were 14 and 15 mmHg with Reichert TonoPen), and dilated fundoscopy revealed that cup/disc ratio was 0.1 in each eye. Cycloplegic refraction was +1.50 (+0.50 X 90°) in both eyes; and orthoptic examination including alternating cover test revealed orthophoria without any ocular motility deficiency.

DISCUSSION

Rubinstein-Taybi syndrome is a neurodevelopmental disorder with diverse systemic findings. There are several case reports and reviews about ocular findings of RTS.[1-7] Rubinstein himself also collected 571 patients with RTS from world literature and communications with other clinicians and families of patients, as well as from his personal observations. He reported ocular pathologies as follows: strabismus in 71%, refractive errors in 56%, ptosis in 29%, and lacrimal duct diseases in 37% of the cases.[5] vanGenderen et al.[6] performed a literature search for reports describing ocular features in 24 patients with RTS and defined the most common ocular abnormalities seen in such cases. Retinal dysfunction, nasolacrimal duct diseases and cataracts were reported as the most frequently found eye anomalies. Their results were consistent with other studies about ocular findings in RTS so far; fundoscopic examination, visual evoked potentials (VEP) and electroretinography (ERG) were also performed and macular abnormalities such as retinal pigment epithelium changes, abnormal waveforms in VEP and retinal dysfunction in ERG were found.[6] Congenital glaucoma is also defined as a rare finding of RTS in the literature.[7-9] On the other hand corneal lesions, optic nerve anomalies, large cup-disc ratio as well as megalocornea that may mimic glaucoma were also reported in nonglaucomatous eyes of cases with RTS.[7-9]

Megalocornea is characterized by symmetric clear corneas larger than 12 mm in diameter at birth with normal endothelial cell density. Congenital form of megalocornea can be associated with trabecular and/or iris dysgenesis and therefore can cause predisposition to glaucoma.[9,10] Brei at al.[9] reviewed 614 cases of RTS and glaucoma was reported in only 32 patients of which 14 had megalocornea. In the same study, authors also published that megalocornea was diagnosed in 4 RTS patients without glaucoma.[9] Presented case was also referred our center with a misdiagnosis of bilateral buphthalmos, but glaucoma was ruled out after a detailed ophthalmologic examination.

Both glaucoma and signs stimulating glaucoma like megalocornea, optic nerve cupping and corneal haziness can be observed together or separately in cases with RTS. In order to rule out the patients who were misdiagnosed as glaucoma, performing a meticulous ophthalmologic examination is mandatory in RTS cases with various ocular abnormalities.

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