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Case Report Open Access

Unilateral Congenital Blepharoptosis Associated with Ipsilateral Multiple Congenital Anomalies in 2 Cases

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Keywords: Congenital blepharoptosis; Ipsilateral multiple congenital anomalies

Introduction

Ptosis, which was formerly called congenital ptosis, is actually the result of an isolated dystrophy of the levator muscle affecting both contraction and relaxation of the fibers. In 25% of the cases, the superior rectus muscle shares the same dystrophic changes as the levator, resulting in weakness of upgaze [1]. The extraocular muscles were derived from the mesoderm since the fifth embryonic week, while the levator palpebrae superioris muscle was differentiated from the superior rectus at the seventh embryonic week [2]. One large-scale epidemiological data from the Section of Ophthalmic Genetics in China reported the prevalence of congenital ptosis in population was 0.18% (1:552) [3]. The patients with congenital ptosis come to the doctors mainly for appearance and vision-development reason. Although earlier intervention may be required in the case of significant visual impairment (amblyopia), surgery is generally performed during the pre-school years (age 3-5). But in congenital ptosis, there may be some abnormalities of other organs during the embryonic development. Two cases of ptosis with special clinical presentations reported here seem to support this point of view.

Case Report

Case one

A 15 years old boy suffered from ptosis of the right upper eyelid after birth. His intelligence is normal but his voice sounds too high as for a teenage boy. His figure was small as a teenage boy and no obvious secondary sexual characteristics developed (Figure 1). The right upper eyelid covered cornea about two thirds and the strength was about 3 mm. No obvious abnormalities were found in anterior and posterior ocular segment. The left eye is normal. There were no abnormal findings by B ultrasonic-scans in both eyes. The right urinary system (kidney, ureter) sonographic image was normal, whereas the size of left kidney was about 11 × 5.5 cm. No abnormal echo was found in the renal parenchyma and collecting system. Significant low echoic separation, high echoic granule and abnormality of the left urethra, bladder and prostate were not found. A well-defined hypo echoic avascular mass of 16 mm × 6 mm × 11 mm in left inguinal region was detected (Figure 2). No testis echo was observed in the right inguinal region and in both scrotums. Abdominal CT demonstrated the absence of right kidney, ureter, testicles, the right indirect inguinal hernia and left cryptorchidism (Figure 3 and 4). Serum sex hormones level test showed the testosterone was 0.04 ng/ml (normal range 0.28-11.1) and the dehydroepiandrosterone sulfate was 1.02 umol / L (normal range 0.66-6.70).

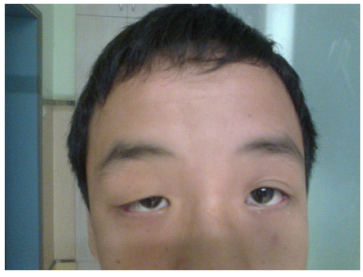

Figure 1: Outlook of the boy.

Case two

A 3 years girl old was admitted to cardiac department after the finding of cardiac murmur for 1 month. The child was found to have ptosis of the right upper eyelid soon after the birth. She was the first child in her family and had a natural labor. During her mother's pregnancy, there was no history of exact toxins, radiation exposure. No similar presentation was detected in her family members. Her intelligence is

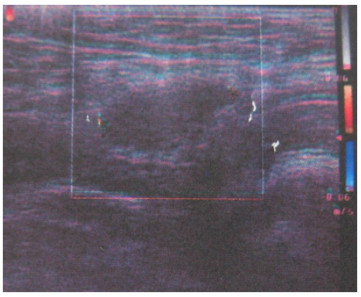

Figure 2: Ultrasonographic scan shows left inguinal cryptorchidism.

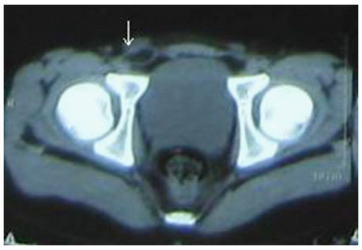

Figure 3: Abdominal CT scan shows right inguinal hernia (arrow head).

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normal. She was too young to pronounce the "E" chart but could speak out the animal pictures from 1 meter's distance. The left upper eyelid ptosis covered the cornea in about half and the strength was about 4 mm (Figure 5). No obvious abnormalities were found in anterior and posterior ocular segment. Doppler ultrasound showed, she had patent ductus arteriosus (PDA) and ventricular septa defect (VSD) and abdominal CT scan revealed left kidney dysplasia and the size was significantly shrinked (Figure 6). There were two renal pelicalyceal systems arranged up and down. The unilateral ectopic ureter opened abnormally in the vagina. Pelvic B ultrasonic-scan was not available for her. No gene abnormality was found during autosomal screening.

Discussion

The neonatal screening program in our country does not cover the ultrasonographic and the color Doppler ultrasonographic screening of the pelvic abnormalities because of Health Economics reason in our country. But china is the second richest country around the world and our party is so "great". But ptosis is easy to be noticed by their parents, which wound be used as a clue to further tests. The authors believe the little boy of the first case would have a better life if the cryptorchidism had been found before age of 6 and 12 months [4]. Therefore, the authors advocate that the abdominal ultrasonography and the color Doppler ultrasonography should be performed to detect other organic abnormalities if congenital blepharoptosis presents. And the noninvasive, easy-preformed and less expensive examination could make a better life to a child.

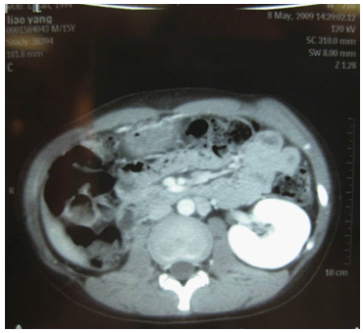

Figure 4: Abdominal CT scan reveals right renal agenesis.

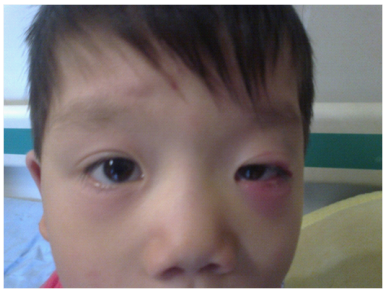

Figure 5: Outlook of the case 2.

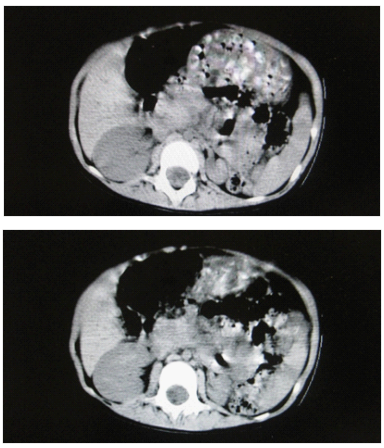

Figure 6: Abdominal CT shows left kidney dysplasia and significant shrink of its size.

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