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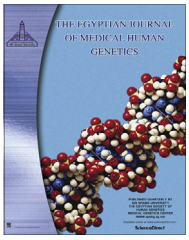

CASE REPORT

Baraitser–Winter syndrome: An additional Egyptian patient with skeletal anomalies, bilateral iris and choroid colobomas, retinal hypoplasia and hypoplastic scrotum

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KEYWORDS

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Choroid coloboma;
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Hypoplastic scrotum;
Mental retardation

Abstract We report a 3.5 year old male child, second in order of birth of non consanguineous Egyptian parents with Baraitser–Winter syndrome (BRWS). The patient had bilateral colobomas of the iris and choroid. Our patient had also retinal hypoplasia, which was not reported previously in this syndrome, bilateral congenital ptosis, hypertelorism, moderate mental retardation, short stature, short neck, hyperextensibility of the joints of the hands, talipes equinovarus, kyphoscoliosis and unilateral hypoplastic scrotum and testis.

1. Introduction

Baraitser–Winter syndrome (BRWS) is a rare but well-defined developmental disorder recognized by the combination of congenital ptosis, high-arched eyebrows, hypertelorism, ocular coloboma and a brain malformation consisting of anterior predominant lissencephaly. Other typical features include postnatal short stature and microcephaly, intellectual disability, seizures and hearing loss [1–4]. BRWS may be considered another example of syndromic neuronal migration defect [5].

We report a case with the typical features of BRWS which has in addition some unreported features after taking consent of the parents.

2. Case report

A 3.5 year old male child, second in order of birth of healthy non consanguineous Egyptian parents. The patient was delivered at full term by cesarean section. His birth weight was 3 kg. No problems were noted by the mother during pregnancy. The patient was referred to the Genetics Clinic, Pediatric Hospital, Ain Shams University complaining of developmental delay and abnormal features.

At the age of 3 days, the mother noticed that her son had poor sucking and difficulty in breathing which necessitated admission to neonatal intensive care unit (NICU) for 3 days. At the age of 1 week he developed convulsions. He was admitted to NICU again and started epanutin and sominaletta for 40 days. The convulsions stopped after 1 month and the patient stopped anticonvulsant drugs. The mother also noticed

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abnormal features in the form of squinted eyes and foot deformity. He had developmental delay as he can only stand with support and can say 3 words only.

Family history was unremarkable. He had two healthy sibs. Both parents were normal.

On examination, the patient had moderate mental retardation, his weight was 12 kg (5th percentile), his length was 86 cm (below 3rd percentile), and his skull circumference was 49.5 cm (50th percentile).

The patient had high forehead, high arched eyebrows, prominent glabella, depressed nasal bridge, downward slanting palpebral fissures with epicanthus inversus, slight hypertelorism, bilateral iris colobomas, wide palpebral fissures, bilateral convergent squint and bilateral congenital partial ptosis, broad bulbous nose, with broad nasal tip, hypoplasia of malar regions, full cheeks, long philtrum, thin upper lip, macrostomia, high arched palate and pointed chin (Fig. 1). The ears were small and low set. The neck was short with mild webbing. The patient had also low posterior hair line and hyperextensibility of the joints of the hands (Fig. 2).

He also had dystrophic nails, broad end of big toes, wide space between big toes and second toes, deviation of other toes medially, overriding of 4th and 2nd toes over 3rd toe and talipus equinovarus more marked on the left side (Figs. 3 and 4).

Also there were narrow shoulders, pectus excavatum and kyphoscoliosis of the back (Figs. 1 and 2).

Abdominal examination revealed small umbilical hernia. Cardiac examination was normal. Genital examination revealed hypoplastic right scrotum and testis (Fig. 5). Neurologic examination demonstrated mild hypotonia in lower limbs.

Abdomino-pelvic ultrasonography and ECHO cardiography were normal. Extended metabolic screen, serum lactate and serum ammonium were normal. Karyotype was also normal. Fundus examination revealed bilateral choroidal colobomas with retinal hypoplasia over the colobomas defects of the choroid. X-ray spine demonstrated kyphoscoliosis, however vertebrae were normal. MRI brain (axial T1WI) revealed cortical thickening in the right occipitotemporal region (arrows) keeping with pachygyria (Fig. 6). Audiometry was normal. X-ray of the feet revealed adduction and varus deformity of the left fore foot (Fig. 4).

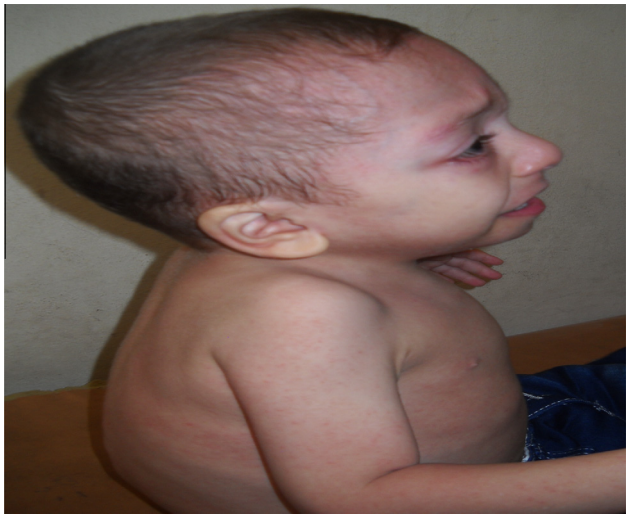

Figure 2 Short neck, small low set ears and kyphoscoliosis.

3. Discussion

We report a 3.5 year old male child with BRWS with bilateral colobomas of the iris and choroid, bilateral congenital ptosis, hypertelorism, moderate mental retardation, short stature, broad nasal bridge, prominent cheeks which slope down to a pointed chin, long philtrum, large mouth, thin upper lip, short

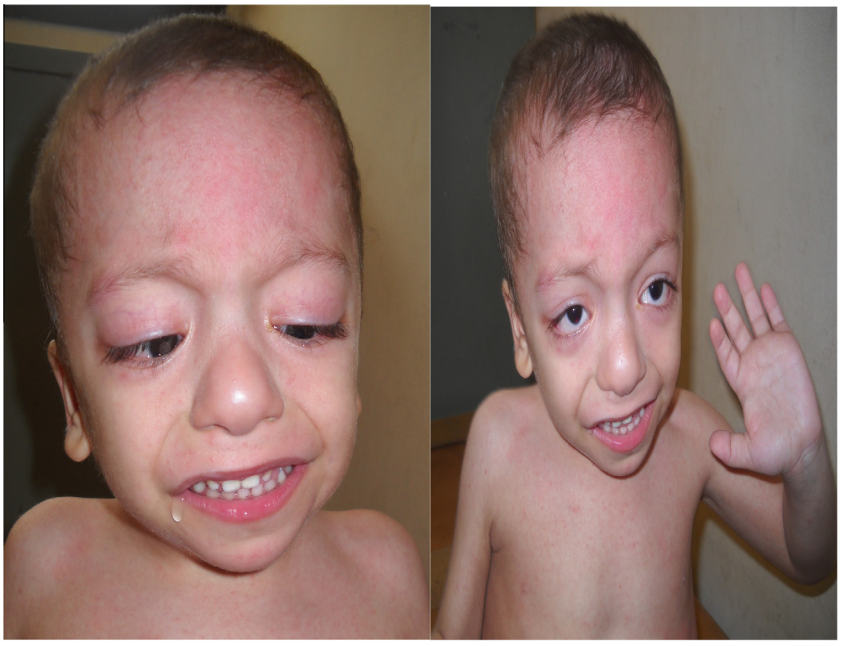

Figure 1 Facial features with short neck and narrow shoulders.