Congenital Partial Hemihypertrophy, Low Set Ears, Hypertelorism, and Epicanthi Folds: A Novel Syndromic Association

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Abstract

**Background:** Congenital hemihypertrophy is a very rare condition that can be isolated or occurs in association with other congenital abnormalities and neoplasms, and well-recognized syndromes. Well-known syndromes that are associated with congenital hemihypertrophy include Klippel-Trenaunay-Weber syndrome, Beckwith Wiedemann syndrome, Goldenhar syndrome, Silver-Russell’s syndrome. Partial hemihypertrophy affecting a limb is an extremely rare condition, and was probably first reported in 1949 by web, and very few cases have been reported in the literature.

**Patients and methods:** Ten month old infant with partial hemihypertrophy, delayed development and facial dysmorphism who was observed at the pediatric neuropsychiatry clinic of Baghdad Medical City was studied.

**Results:** The boy was hypotonic with developmental delay and has not been able sit yet. He had low set ears and facial dysmorphism consisting of hypertelorism and epicanthic folds. His right lower limb was obviously larger than the left. Parents were consanguineous, and family history was negative for similar condition. The rest of the examination was normal. Brain CT-scan, echocardiography, abdominal ultrasound, and chromosomal karyotype showed normal finding.

**Conclusion:** Hemihypertrophy has not been reported in Iraq before. A novel syndromic association consisting of congenital partial hemihypertrophy, low set ears, hypertelorism, epicanthi folds, and developmental delay is reported in this paper.

**Key words:** congenital partial hemihypertrophy; low set ears; hypertelorism; epicanthi folds; and developmental delay; new syndrome

Introduction

Congenital hemihypertrophy is a very rare condition that can be isolated or occurs in association with other congenital abnormalities and neoplasms including hepatoblastom (Geiser et al; 1970; Rattan et al; 1995); Wilms tumor (Sauer and Wemmer; 1977; Mohanna and Sallam; 2008); embryonal rhabdomyosarcoma (Samuel; Tsokos; and DeBaun; 1999); epithelioid haemangioendothelioma (Miller et al; 1999); congenital mesoblastic nephroma (Abosoudah et al; 2008) [1-29]; and well-recognized syndromes. Well-known syndromes that are associated with congenital hemihypertrophy include Klippel-Trenaunay-Weber syndrome; Beckwith Wiedemann syndrome; Goldenhar syndrome; Silver-Russell's syndrome [30-33].

Partial hemihypertrophy affecting a limb is an extremely rare condition; and was probably first reported in 1949 by web [34]; and very few cases have been reported in the literature.

Patients and methods

Ten month old infant with partial hemihypertrophy; delayed development and facial dysmorphism who was observed at the pediatric neuropsychiatry clinic of Baghdad Medical City was studied.

Results

The boy was hypotonic with developmental delay and has not been able sit yet. He had low set ears and facial dysmorphism consisting of hypertelorism and epicanthic folds (Figure-1). His right lower limb was obviously larger than the left (Figure-2). Parents were consanguineous; and family history was negative for similar condition. The rest of the examination was normal. Brain CT-scan; echocardiography; abdominal ultrasound; and chromosomal karyotype showed normal finding.

Discussion

Congenital hemihypertrophy is well known to be associated with multiple congenital anomalies (Table-1). Partial hemihypertrophy affecting a limb is an extremely rare condition; and was probably first reported in 1949 by web [34]; and very few cases have been reported in the literature [12; 35; 36; 37]. Kasantikul et al (1994) reported a rare case of adrenocortical adenoma in a 7-month-old female infant with congenital hemihypertrophy of left leg [35]. Gönilü et al (2009) from Turkey report a very rare case of giant melanocytic naevus with lipomatosis; Dandy-Walker malformation occurring in association with hemihypertrophy of the leg [36].
Author(s) | Congenital anomalies | Abnormalities associated with congenital hemihypertrophy
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Walia et al [1971][13] | Congenital heart disease | Mohanna and Sallam (2008) from Yemen reported a case presented with partial hemihypertrophy of the right leg and foot with polydactyly; a right sided abdominal mass caused by an ureteropelvic junction stricture with hydrenephrosis; and absent left kidney [12].
Paré and Elhilali [1972][14] | Hydrometrocolpos and polydactyly | Deyrup et al (2011) from USA reported a very rare association of cutaneous angiosarcomas with congenital hemihypertrophy of the contralateral limb [37].
Indridason; Thomas; and Berkoben (1996)[25] | Abnormalities of the aortic arch; and of the skeletal; cutaneous and ocular systems. |
Kusz et al (2019)[29] | | 
Henry et al [1973][16] | Abnormalities of the aortic arch; and of the skeletal; cutaneous and ocular systems. | 
Temtamy and Rogers [1976][17] | Macrodactyly; and connective tissue nevi | 
Fischer; Strand; and Shapiro [1984][18] | Abnormalities of the cerebral vasculature: giant aneurysm; capillary hemangioma; and arteriovenous malformation | 
Hidano and Arai [1987][19] | Syndactyly; scoliosis; short forth metacarpus; hypoplastic mandible; peroneal exostosis; multiple faint nevi flammei; telangiectasis; nevus anemicus; fibromatous tumors of the tip of the tongue; mitral prolapse; and vascular; cerebral; abnormalities | 
Agarwal et al [1988][20] | Benign nephromegaly and duplex ureter | 
Giani; Lapi; Pezzati [1991][21] | Nevus depigmentosus | 
Dawn et al [1995][22] | Right upper limb triplication; polythelia; congenital hip dislocation; facial dysmorphism; congenital heart disease; and scoliosis | 
Sabry et al [1995][23] | Hemimegalencephaly | 
Calzolari et al [1996][24] | Renal dysplasia and benign nephromegaly | 
Memon et al [2005][26] | Congenital hemihypertrichosis | 
Akarsu et al [2005][27] | Café au lait spots; left-sided cryptorchidism; and albinotic spots of the retinal pigment epithelium | 
White et al [2018][28] | | 

**Conclusion**

Hemihypertrophy has not been reported in Iraq before. A novel syndromic association consisting of congenital partial hemihypertrophy; low set ears; hypertelorism; epicanthi folds; and developmental delay is reported in this paper.
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