Volume : 3 | Issue : 5 | May 2014 • ISSN No 2277 - 8179

Hydronephrosis Important For Diagnosis In Schinzel-Giedion Syndrome

Medical Science

KEYWORDS : Schinzel-Giedion syndrome, Hydronephrosis,Midface hypoplasia.

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ABSTRACT

Schinzel-Giedion syndrome is a rare autosomal recessive disorder characterized by coarse facies, midfacer-etraction, hypertrichosis, multiple skeletal anomalies, and cardiac and renal malformations. Craniofacial abnormalities of this syndrome sometimes resemble a storage or metabolic disease. The pathogenesis of the disease remains unknown. The objective of this report was to emphasize the importance of congenital bilateral hydronephrosis for the diagnosisof Schinzel-Giedion syndrome.

SUMMARY

We describe a indo-aryan newborn with typical facies,low set ear,high arched palate,undescended testis and bilateral hydronephrosis confirmed by abdominal ultrasonography.Of the 37 cases already reported in the literature, 33 presented hydronephrosis, which is considered an important cluein diagnosis. If Schinzel-Giedion syndrome were indexed as a cause of congenital hydronephrosis, its identification would be greatly facilitated, since the majority of the other findings in Schinzel-Giedion syndrome are nonspecific and commonto many genetic syndromes.

INTRODUCTION:

Schinzel-Giedion syndrome (SGS),first described in 1978, is a rare syndrome characterized by midfaceretraction,hypertricho sis, multiple skeletal anomalies, and cardiac and renal malformations.Some authors believe that if the syndrome identification were indexed as a cause of congenital hydronephrosis,its diagnosis would be considerably facilitated[1]. In order to reinforce this opinion and the phenotypic spectrum of the syndrome, we resolved toreport another case: a case from india. To date, more than 30cases have been reported in the literature.

CASE REPORT

We report a newborn male infantwith bilateral hydronephrosis;the mother was a 35-year-old woman. There was no parental consanguinity nor family history of congenital abnormalities.

Anthopometric examination showed term baby with a birth weight = 2.740 g (20th centile); length = 49.9 cm (50th centile),O.F.C.= 31 (<2.5 centile); inner canthal distance = 2.2 cm (75th centile); interpupillary distance = 4.2 cm (50thcentile); outer canthal distance = 5.9 cm (25th centile) US/LS ratio-1.7.

The head to toe examination showed prominent forehead, widely patent fontanels and sutures,coarsefacies with short and"squared" nose with anteverted naresand depressed bridge,high arched palate, ears apparently low-set and posteriorly rotated with folded helices, prominent eyes with a deep groove underneath, distended abdomen with bilateral ballotable kidney and undescended testis. (fig-1)

Fig.1-Head to Toe examination of child.

Ultrasound of abdomen detected bilateralhydronephrosis in postnatal life (Fig.2).

Chromosomal analysis was not done.

Fig.2- USG Abdomen

DISCUSSION:

The phenotypic characterization ofSGS includes a coarse midface retraction, a prominent forehead, and an enlargedand protuberant tongue. These craniofacial abnormalities sometimes resemble a storage or metabolic disease,but patients with SGS do nothave a biochemical abnormality[1,2]. Hirsutism disap-

pears and midface retraction becomes less evident with age; In contrast, bitemporal narrowing becomes more evident[3]. In addition, radiological findings with a specific skeletal dysplasia and the presence of bilateral hydronephrosis strengthened the diagnosis of SGS.Hydronephrosis is only occasionally reported as a feature of a malformationsyndrome, such as Johansson-Blizzard syndrome, trisomy 13 and 18,Turner syndrome, triploidy, and Ochoa syndrome[1].Of the 35 SGS cases reviewed by Touge et al. (2001)[4], 31 presented hydronephrosis,which is an importantclue in diagnosis, and Minn et al.(2002)[5] have the same opinion. Kelleyet al. (1982) considered that the syndrome identification would be greatly facilitated if the Schinzel-Giedion syndromewere "indexed" as a cause of congenital hydronephrosis.

We also observed the renal anomaly in our patient and agree with the authors that the majority of the findings of this syndrome except hydronephrosis are nonspecific and common to many genetic syndromes (Table1).Since the gene of the disease has not yet been identified and diagnosis is strictly based on clinical findings,the presence of hydronephrosis assumesan important role for the diagnosis of SGS. Despite the lack of identification of any biochemical abnormality so far, Shah et al. (1999) [6] suggested that the progressive neurodegenerative process in SGS associated with a coarse facial appearance and skeletal abnormalities could be associated with a metabolic defect.

Some authors have described sacral tumors associated with this syndrome,and McPherson et al. (1998)[7] have considered that this finding could help in explaining the pathogenesis and/or identifying candidate genes forthis autosomal recessive condition.Therefore, additional patients should be reported in order to amplify the phenotypic spectrum of SGS.

Table 1 - Features of Schinzel-Giedion Syndrome.

FEATURES	Previous cases	Our patient
Craniofacial findings		
Coarseface	27/27	+
Prominentforehead	26/27	+
Wide anterior fontanels/sutures	22/22	+
Hypertelorism	34/35	–
Midfacialhypoplasia	37/37	+
Low nasal root	23/25	+
Low-set ears	21/23	+
Hypertrichosis	15/24	--
Seizures	23/26	–
Delayed development	26/27	--
Structural abnormalities		
Genital abnormalities	28/29	–
Hydronephrosis	33/37	+
Congenital heart disease	13/29	--
Choanal stenosis	9/28	–

Adapted and updated from Touge et al. (2002); Minn et al. (2002), and Labrune et al.[4](1994) and the following articles: Rodriguez et al. (1994); Antich et al. (1995)[2]; Özkinay etal. (1996)[7]; McPherson et al. (1998); Shah et al.1999)

CONCLUSION: Only few cases are reported from india till now. If Schinzel-Giedion syndrome were indexed as a cause of congenital hydronephrosis, its identification would be greatly facilitated, since the majority of the other findings in Schinzel-Giedion syndrome are nonspecific and common to many genetic syndromes.

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