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Schinzel-Giedion Syndrome

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Schinzel-Giedion syndrome Is a rare disorder characterized by midface retraction, hypertrichosis, multiple skeletal anomalies, cardiac and renal anomalies and severe developmental retardation(l). This was first described by Schinzel and Giedion in 1978(2). A total of 11 cases have been so far reported(3) and here we report the first case from India.

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Case Report

The patient was a 1-year-old boy, fifth issue of a third degree consanguinous marriage, of a thirty year old gravida five, para three mother and thirty seven year old father. The pregnancy was full term by dates and was uncomplicated by illness, irradiation or teratogenic drug exposure. The three elder siblings were normal, but the fourth issue was a boy who was born at term and had dysmorphic features with club feet like the patient and died at 3 months of age. There was no history of any major illness during infancy. Motor, language and intellectual milestones were markedly delayed.

On physical examination, he was a markedly dysmorphic child with coarse face, wide open anterior fontanelle, frontal bossing, anteverted nostril, midface retraction, ocular hypertelorism and bilateral talipes equinovarus. External genitalia showed bilateral undescended testes with hypoplastic scrotum *(Fig; 1).* Other anomalies are listed in *Table I.* Laboratory investigations revealed normal serum concentration of blood urea, creatinine, thyroid profile, urinary mucopolysaccharides, aminoacids and

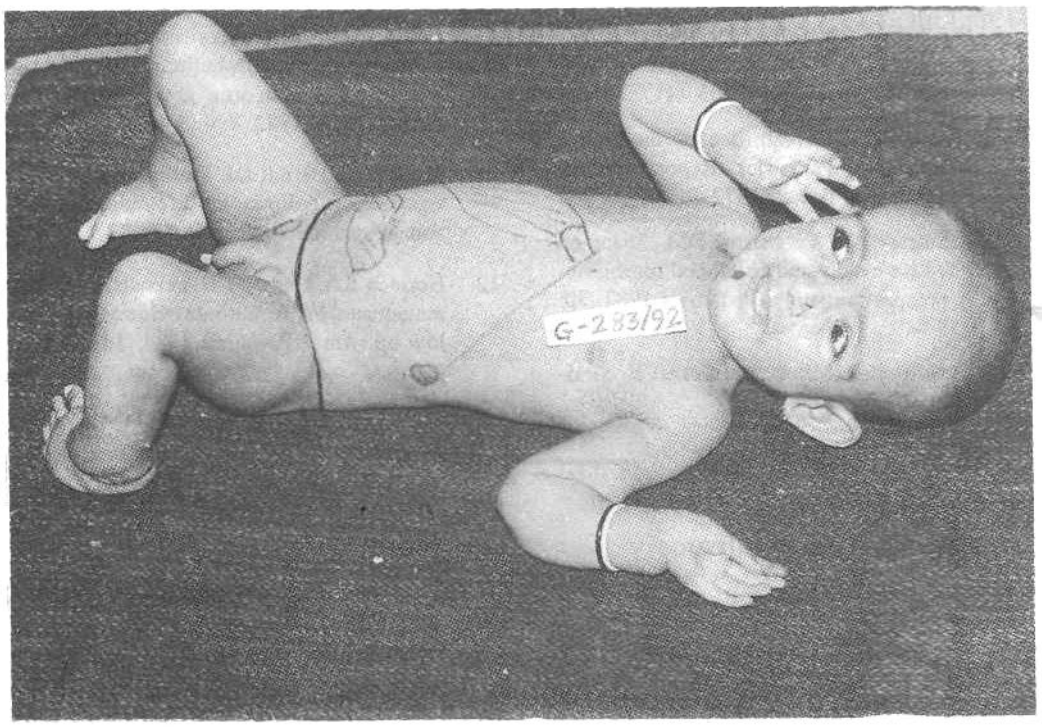

Fig. 1. The distinctive appearance of Schinzel-Giedion syndrome with wide open fontanelle, lowset ears, midface hypoplasia, protruding forehead, ocular hypertelorism, anteverted nostrils, bilateral talipes, equinovarus and undescended testes.

organic acids. Leucocyte enzymes were normal. Optic fundi and BERA were normal. Serum levels of TORCH infections were normal and Giemsa banded karyotype was 46XY. Ultrasound examination of abdomen showed normal kidneys and intravenous pyelogram and micturating cystourethrogram were normal. EMG and nerve conduction study was normal. 2D Echocardiogram of heart revealed moderate subaortic ventricular septal defect. CT scan of brain showed symmetrically dilated ventricular system. The radiologic findings are summarized in *Table I.*

Discussion

The concordance of the clinical and radiologic findings of our patient with those previously reported(2) in the Schinzel-Giedion syndrome is diagnostic *(Table I).* The characteristic features of our patient, midface retraction, wide open fontanelle, short anteverted nose, hypertelorism, lowset ears and CTEV are all salient features of this unique syndrome(4). The diagnosis was further confirmed by the distinctive radiological anomalies, especially of the skull *(Fig.* 2). The most important systemic involvements other than skeletal system are cardiac defects (as seen in our patient), renal defects in the form of hydronephrosis and cerebral ventricular system dilatation (also seen in our patient). Hydronephrosis is a very useful association helping in the

TABLE I-*Major Clinical and Radiological Findings in Schinzel-Giedion Syndrome*

Major features	Schinzel & Gideion		Donnai &	Present
	Case I	Case 2	Harris(4)	case
Craniofacial dysmorphic features				
Wide open anterior fontanelle	+	+	+	+
Frontal bossing	+	+	+	+
Midface retraction	+	+	+	+
Ocular hypertelorism	+	+	+	+
Short upturned nose	+	+	+	+
Hemangioma	-	-	+	+
Lowset ears	+	+	-	+
Major structural defects				
Genital anomalies	+	+	+	+
Congenital heart defect	+	+		+
Severe mental retardation	+	+	+	+
Hydronephrosis	+	+	+	-
Ventricular dilatation	?	?	+	+
Talipes equino varus	-	-	+	+
Radiologic anomalies				
Sclerotic base of skull	+	+		+
Multiple wormian bones	+	+	+	+
Wide open synchondrosis	+	+	+	+
Broad ribs	?	+	+	
Widening of distal femora	-		+	+
Short thumb metacarpal	+	+		+
Hypoplasia of distal phalanges	+	+		+

antenatal diagnosis of the syndrome(5).

In the differential diagnosis, the coarse facial appearance of these children resemble cretinism, mucopolysaccharidosis, gangliosidosis and other metabolic storage disorders(l). These conditions were excluded in this patient by appropriate screening tests as well as by the absence of specific skeletal features. Chromosomal anomalies like partial trisomy l0q, trisomy 9 mosaic, trisomy 20p- and 9p-syndrome phenotypically have resemblance with his syndrome, but are excluded by karyotyping.

Schinzel-Giedion syndrome is a single gene, autosomal recessive disorder. Prenatal diagnosis could be offered by means of abdominal ultrasonography looking for hydronephrosis and severe skeletal anomalies(l). The natural history of this condition is severe growth retardation and profound

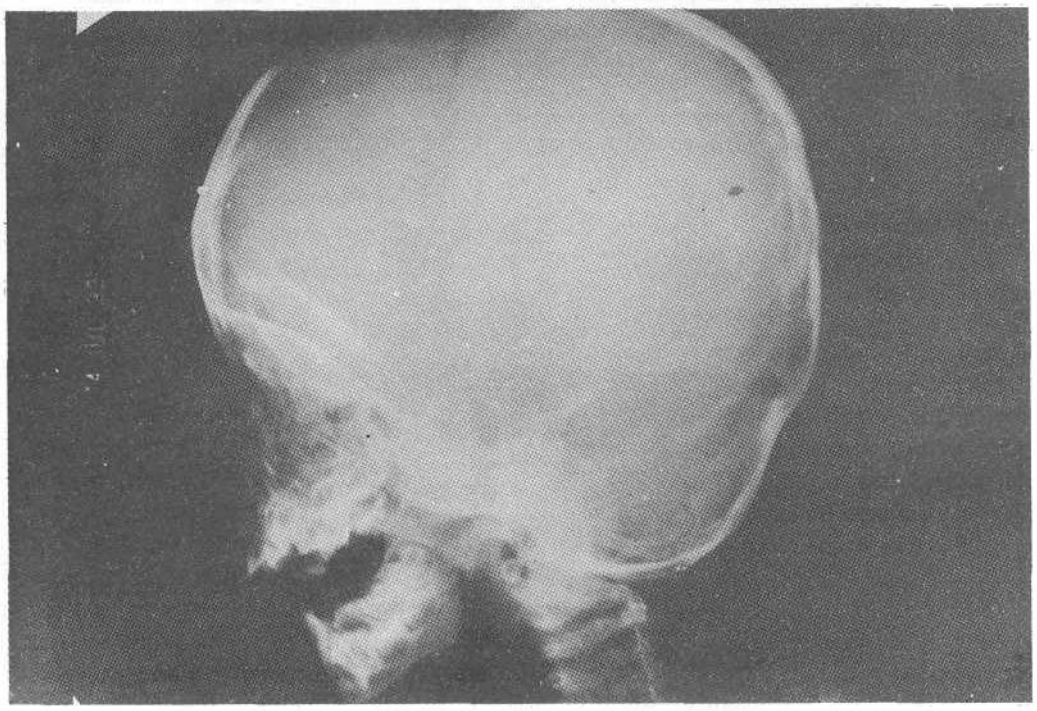

Fig. 2. Radiograph of lateral view of skull showing sclerotic base of skull.

mental retardation with visual and hearing defects. All the previously reported children died before the age of 2 years. Genetic counselling to parents and proper antenatal diagnosis are very important(6).

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