RUBALCABA SYNDROME : A CASE REPORT

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SUMMARY : This article describes a 2-year-old boy with growth, motor and mental retardation and several clinical findings particularly suggestive of the features of Rubalcaba syndrome.

***Key Words :** Growth Deficiency - Rubalcaba Syndrome - Unusual Face.*

INTRODUCTION

In 1971, Rubalcaba et al. reported a new familial dysmorphic syndrome characterized by mental retardation, short stature, unusual face, microcephaaly, small hands and feet, skeletal dysplasias and hypoplastic genitalia (Rubalcaba et al.1971). Up-to present time there have been about 18 cases reported in the literature (Bialer et al. 1989; Bianchi et al. 1984; Hunter et al. 1977; Rubalcaba et al. 1971). In this article a 2-year-old male patient with Rubalcaba syndrome is presented and pertinent literature is reviewed.

CASE REPORT

The male, 2-year-old patient was the first child of healty unrelated parents (mother 28, father 32 years old). The birth weight was 2450 g. The neonatal period was referred normal. His early development was delayed. He was not able to sit without support or speak yet. He was having stiffening episodes during his fecal discharge or micturation.

On physical examination, he weighed 8200 g. with a height of 74 cm, his OFC was 46,5 cm (all of them were below the 3rd percentile) and anterior fontanel measured as 2x2 cm open. His personal-social development was extremely retarded. He had sparse-grey hair. The face was peculiar: metopic prominence, arched eyebrows, downslanted palpebral fissures, a long nasal septum and hypoplastic nasal alae, small downturned mouth and thin lips, long philtrum, high arched palate, irregular and crowded mandibular teeth, slightly low set and anteverted ears. The neck was short and the nipples were wide spaced (see fig. 1, 2, 3). He had inguinal hernia and sacral dimple bilaterally. His wrists showed limited flexion, the thumbs were deviated radially and elbows revealed incomplet extantion. The fingers were thin. Examination of the external genitalia revealed the cryptorchidism bilaterally and the ultrasonographic examination showed both testicles in canalis inguinalis. Although the EEG was normal, the stiffening episodes were thought to be seizures and they were controlled with phenobarbital. The CT scan revealed a slight cerebral atrophy. Ophtalmologic examination showed pale optic discs. The vessels showed distention and convolution and displacement nasally. Laboratory data revealed iron deficiency anemia. Routine urine analysis was normal and the two urinary bacteriolo-

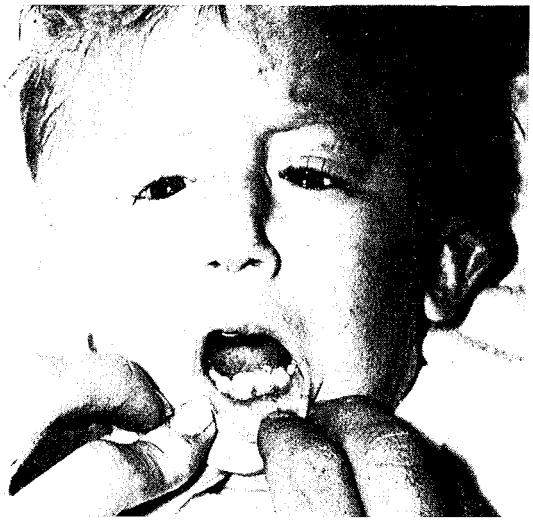

Fig - 1 : Metopic prominence, arched eyebrows, downslanted palpebral fissures, crowded mandibular teeth, slightly low set and anteverted ears.

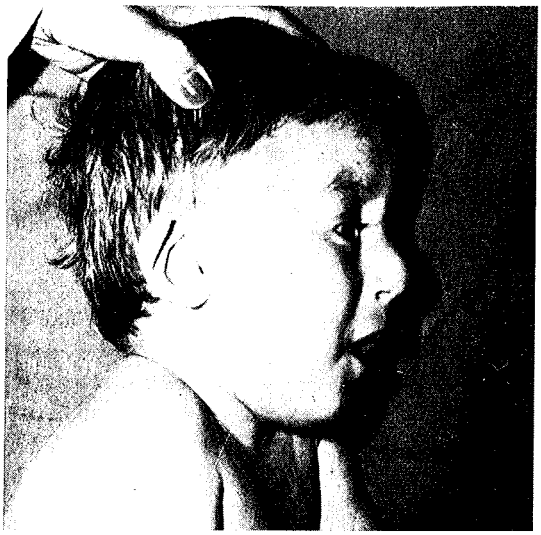

Fig - 2 : Abnormal (beaked) nose, hypoplastic nasal alae, small downturned mouth.

gic cultures were negative. The chromosome analysis disclosed 46 x pattern. LH-FSH and testosteron tests showed normal prepubertal values. Induced by human chorionic gonadotropin (hCG), testesteron response was positive. At the end of (hCG) course, one of the tests descended, the other became retractile. The radiological examination revealed spina bifida occulta at C_1-7 and S_1There was slight thoracal scoliosis. The bone age was 6 months. Intravenous pyelography, Technetium-labeled dietylenetriaminepenta - acetic acid (DTPA), and Technetium-labeled dimecaptosuccinic acid (DMSA) data were normal.

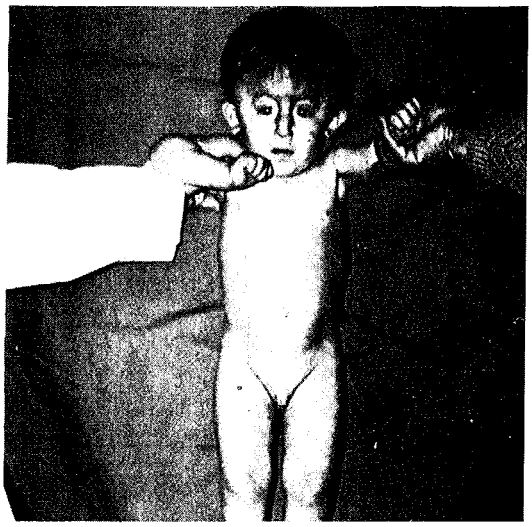

Fig - 3 : Short stature, short neck, wide spaced nipples.

DISCUSSION

The pathogenesis of Rubalcaba syndrome is not clear yet. Rubalcaba suggested a possible X-linked mode of inheritance (Rubalcaba et al. 1971). Our patient is the single case in his family. The mode of inheritance suggested as an autosomal dominant disease with incomplet penetrance in three generations of a family (Hunter et al. 1977). In literature there is no report of chromosome anomaly except the report of a patient showing a balanced Robertsonian translocation involving chromosomes 13 and 14. That was also present in his unaffected father (Bialer et al. 1989). The choromosome analysis of our patient was normal. The radiographic examinations of hands of the parents were normal as well.

The clinical features of previously reported cases are summarized in table 1. The patient described herein shows most of the clinical features of the Rubalcaba syndrome.

Ocular involvement reported in one case include central tapetoretinal dystrophy (Bialer et al. 1989).

The previously reported genitourinary abnormalities include hypoplastic genitalia, hypospadias, cryptorchidism, abnormal positioning of the kidney, small kidney, megaureter, hydronephrosis, accessory ovary, ovarian cyst, atretic fallopian tube (Bialer et al. 1989; Bianchi et al. 1984; Rubalcaba et al. 1977). The urogenital finding in our patient was cryptorchidism.

The skeletal features of the syndrome include norrow trunk, pectus deformities, scoliosis, kypho-

Systems	Features	Percentage of Patients affected
Skin	Hypoplastic skin, sparsity of hair*, arched eyebrows*, large areolae Wide spaced nipples*	 > 10 10
Skeleton	Short stature* Microcephaly, high forehead*, narrow maxilla Joint limitation* Broad hips	> 50
	Pectus deformity Narrow trunk	> 10
	Vertebral anomalies*, Scoliosis*, kyphosis, osteochondritis of spine	30-50
	Short extremities, 5th finger clinodactyly, short toes, short metacarpals, and phalanges, coned epiphyses, bulbous ends of metacarpals and metatarsals, fusion of carpals, proximally placed thumbs, small hands and feet	> 10
	Shot metatarsals	> 50
Dental	Irregular, crowded teeth*	> 10
Ocular	Central tapetoretinal dystrophy, pale optic disc*	10
Soft tissue	Cerebral atrophy* Downslanting palpebral fissures*, long upper lip*	> 10
	Mental deficiency*, abnormal nose*, microstomia*, downturned mouth corner*, thin vermillion border	> 50
	Hypoplastic genitalia, pubertal delay, crypthorchidism*, inguinal hernia*, abnormal kidney position, sacral dimple*	> 10

* Indicates the features of our case.
Table 1 : Clinical features of reported cases of Rubalcaba syndrome (Bialer et al. 1989, Bianchi et al. 1984; Hunter et al. 1977; Rubalcaba et al. 1971).

sis, osteochondritis of spine, vertebral anomalies, broad hips, joint limitation, short extremities, small hands and feet, 5th finger clinodactyly, short metacarpals and metatarsals, short phalanges, bulbous ends of metacarpals, fusion of carpals, proximally placed thumbs, craniosynostosis (Bialer et al. 1989; Bianchi et al. 1984; Hunter et al. 1977; Rubalcaba et al. 1971). Our patient shows slighter skeletal features such as cervical spina bifida occulta, scoliosis, joint limitiation, radially deviating thumbs.

Although Hunter et al. discuss that the six cases they reported do not resemble some features of Rubalcaba syndrome and each of these cases or families have unique syndromes, their cases showed several similarities with Rubalcaba syndrome.

In conclusion, it is obvious that we should have several more case reports to specify this syndrome more clearly.

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REFERENCES

1. Bilaer MG, Wilson WG, Kelly TE : Apparent Rubalcaba Syndrome with Genitourinary Abnormalities. Am J Med Genet 33: 314-317, 1989
2. Bianchi E, Livieri C, Arico M, Cattaneo E, Podesta AF, Beluffi G : Rubalcava syndrome: a case report. Eur J Pediatr 142: 301-303, 1984
3. Hunter AGW, Mc Alpine PJ, Rudd NL, Fraser FC : A "new" syndrome of mental retardation with characteristic facies and brachyphalangy. J Med Genet 14: 430-437, 1977
4. Rubalcaba RHA, Reichert A, Smith DW : A new familial syndrome with osseous dysplasia and mental deficiency. J Pediatr 79: 450-455, 1971