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Congenital Contractural Arachnodactyly

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Congenital contractural arachnodactyly (CCA) is an autosomal dominant disorder of connective tissue, similar in many respects to Marfan syndrome. Beals and Hecht in 1972 reviewed their patients carrying the diagnosis of Marfans syndrome in literature and delineated a new syndrome(l). They introduced the term congenital contractural arachnodactyly for this condition(l). CCA is characterized by multiple contractures, dolichostenomelia, scoliosis, arachnodactyly and external ear anomalies(l,2). This new syndrome contrasted with Marfan by the absence of eye and heart anomalies(l,2) but with increasing number of cases reported, occasional association of cardiovascular and ocular anomalies have been reported in patients with CCA(3-5).

This paper reports 3 cases of CCA from different sibships and reviews the relevant literature emphasizing points of differentiation between CCA and Marfan's syndrome.

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The 3 subjects were isolated cases from 3 different sibships and were all products of non-consanguineous marriages. The various clinical features of the 3 cases studied by us are presented in *Table I* and *Figs. 1-4*.

Discussion

Congenital contractural arachnodactyly is an autosomal dominant connective tissue disorder which has emerged as a separate entity from Marfan syndrome. In 1972 Beals and Hecht described two new cases and identified 12 cases of this disorder from literature including the original patient described by Marfan in 1896(1). Subsequently, many affected families(2,3,6) and some isolated cases with no other member

TABLE I—*Summary of Clinical Features in Congenital Contractural Arachnodactyly*

Clinical features Age & Sex	Case I 1 yr F	Case II 13 yr F	Case III 45 days M	Literature	Total
Cranifacial					
Unusual shaped head	-	-	-	18/45	18/48
Micrognathia	-	-	-	15/51	15/54
High arched palate	-	-	-	18/56	18/59
Crumpled ears	+	+	-	44/66	46/69
Extremities:					
Elbows	+	+	+	59/69	62/72
Knee	+	+	+	54/67	57/70
Hips	-	-	-	21/66	21/69
Camptodactyly	+	+	+	61/68	64/71
Arachnodactyly	+	+	+	60/69	63/72
Adducted thumb	-	-	-	23/47	23/50
Club foot deformity	-	-	-	22/59	22/62
Bowed Long bones	+	-	-	36/67	37/70
Hypoplastic muscles	-	-	-	35/67	38/70
Hypoplastic muscles	-	-	-	35/67	38/70
Kyphosis/Scoliosis	-	-	-	35/57	35/60
Others					
Heart defects	-	-	-	11/65	11/68
Osteoporosis	-	-	-	3/53	3/56
Spontaneous improvement of contractures with time	-	-	-	45/48	45/51

of the family affected have been reported(4,7). The three additional cases presented here are all isolated cases. These may represent new mutations.

Our findings in these three cases are summarized in *Table I* and compared with those reported in the literature. Joint contractures are the important features of this disorder. These contractures are present at birth and tend to improve spontaneously with age(6). The most frequently involved joints are those of the fingers, elbow, knee and hip. Wrist, ankle and shoulder may also be affected(2,6). The motor development of these individuals may be delayed because of these contractures(2,6,8).

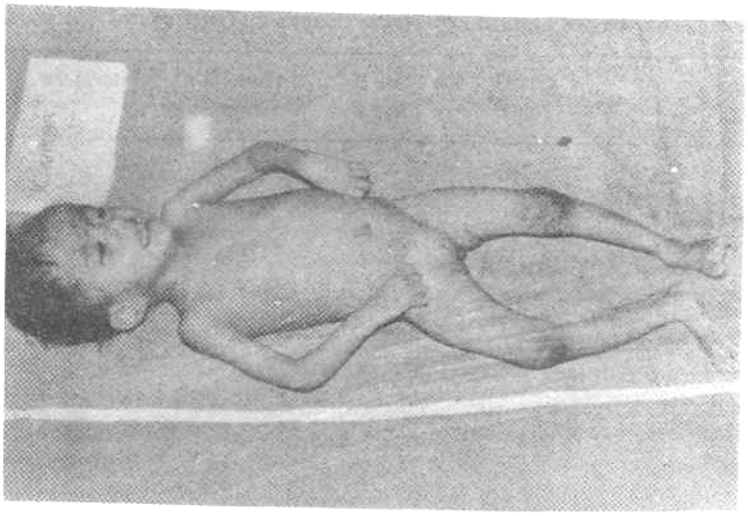

Fig. 1. Case 1 showing arachnodactyly, contractures of fingers, elbow and knee joints.

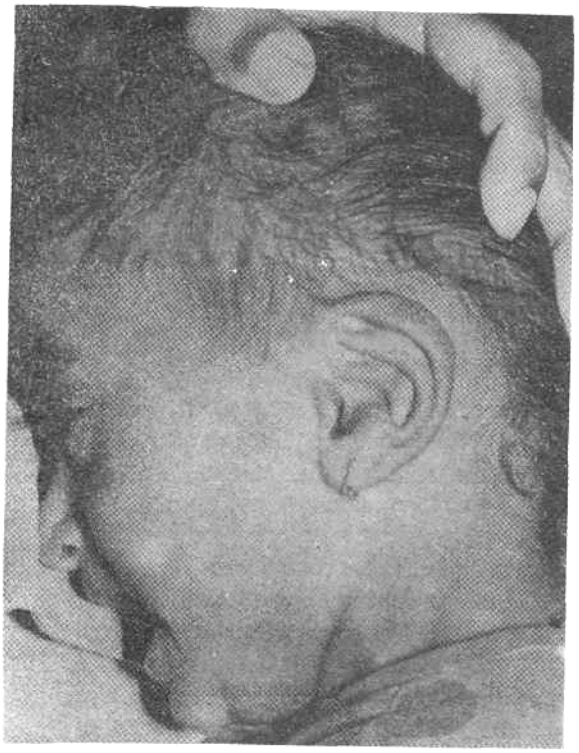

Fig. 2. Note the crumpled ear in Case 1.

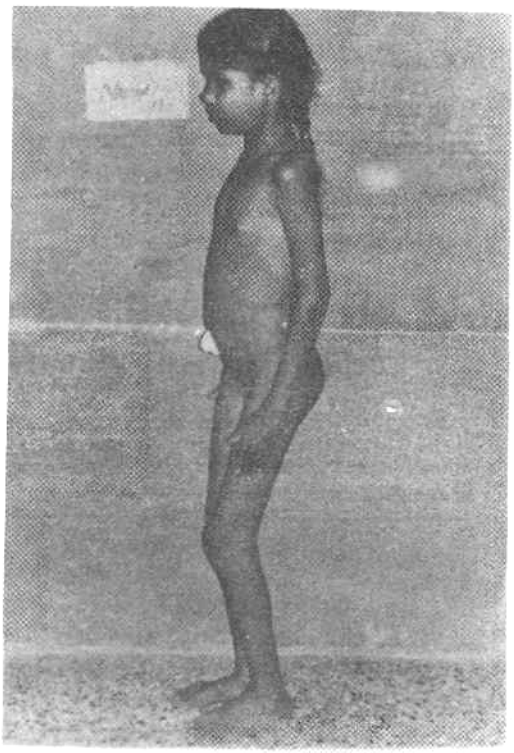

Fig. 3. Showing arachnodactyly and contractures of knee and elbow joints in case 3.

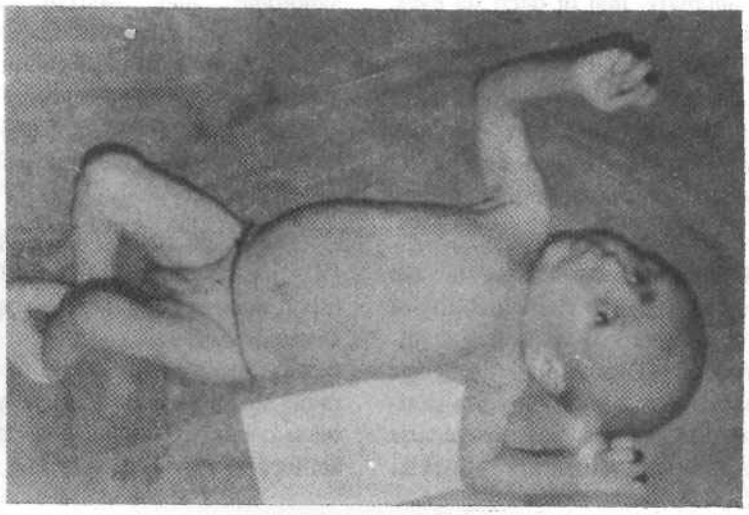

Fig. 4. Note the deep set eyes, arachnodactyly and contractures of elbow and knee joints in case 4.

The affected individuals tend to be tall with dolichostenomelia and arachnodactyly. Spinal abnormalities such as kyphosis and scoliosis are common. When present they usually are not congenital but develop later and progress with age(6). The neck appears slightly short. The head and face are apparently normal but there is a tendency for retrognathia. Crumpled ears are commonly present in these individuals. Other ear abnormalities include crumpled antihelix, prominent crura and partial obstruction of the concha(2,6).

It was previously believed that CCA in contrast to Marfan syndrome is not associated with ocular or cardiac abnormalities(l,2). Subsequently various reports proved that serious cardiac abnormalities can occur in CCA(2,4,5,9). Aortic aneurysm, aortic regurgitation and mitral valve prolapse are the common cardiac anomalies seen in Marfan syndrome. However, individuals with CCA are more likely to have structural heart defects like VSD, ASD, PDA, *etc.(6).* Mitral valve prolapse observed in some of these patients caused no significant problems(6).

In the recent past, two cases of CCA, one having a structurally defective heart who later developed great artery dilatation^) and the other having aortic root dilatation^) have been reported in two separate studies. These reports make the differentiation of CCA from Marfan syndrome much more difficult. In the present study, none of the patients had any cardiac anomaly.

Ocular anomalies like ectopia lentis, high myopia, retinal detachment, iritis and glaucoma are common in Marfan syndrome and are found in *80%* patients(6), while in CCA ocular anomalies are extremely rare. Recently a patient with CCA was reported as having bilateral ectopia lentis(5). Other ocular anomalies reported in the literature include keratoconus, iridodonesis and deeply set eyes. In our study, one patient

had ocular anomaly, that of deep set eyes. Reports show that patients with CCA usually have normal intelligence(l). Other anomalies associated with CCA that have been reported in literature include Klinefelter syndrome, tracheo esophageal fistula, duodenal atresia and unilateral limb deficiency(6,10).

Marked heterogeneity exists in the manifestation of CCA(6). The variability of expression is a feature of autosomal dominant inheritance. This variability gives problems in distinguishing CCA from similar ones like Achard syndrome, osteogenesis imperfecta, homocystinuria, Stickler syndrome and Marfan syndrome(6). Although it is relatively easy to distinguish CCA from most of these conditions, Marfan syndrome has many clinical features in common(2,6). *Table II* presents the differentiation points(2,6,ll) between these two syndromes.

The differentiation between Marfan and CCA has become still more difficult after a report documenting these two syndromes in a same family(12). Until a biochemical means becomes available to more accurately identify CCA, Marfan syndrome and other connective tissue disorders from one another making an accurate diagnosis is

TABLE II—*Differentiating Points Between CCA and Marfan Syndrome*

Congenital contractural arachnodactyly	Marfan syndrome
Contracture	
Always present	Rare
Congenital	Develop later
Spontaneous resolution with time	Worsen with time
Commonly involves elbow, knee, hip, and fingers	Hands & feet are commonly involved
Facial Features	
Oval head	Dolicocephalic head
Retrognathia is common	Prognathism
Ear crumpled	Ear normal
Eye Anomalies	
Very rare	Common
Cardiovascular anomalies rare	Common
Common lesions are VSD, ASD, PDA, MVP	Common lesions are aortic aneurysm, aortic regurgitation MPV
Joint laxity absent	Present
Long term prognosis—good	Poor

difficult. However, we hope that the recent advances in medical genetics localizing Marfan syndrome to chromosome No. 15, and evidence regarding the role of abnormal fibrillin protein in its pathogenesis will open new doors in this direction(13,14).

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