Arthrogryposis multiplex congenita

Part 1: Clinical and electromyographic aspects

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SUMMARY Sixteen cases with arthrogryposis multiplex congenita were examined clinically and electromyographically; three of them were re-examined later. Joint deformities were present in all extremities in 13 of the cases; in eight there was some degree of mental retardation. In two cases, there was clinical and electromyographic evidence of a myopathic disorder. In the majority, the appearances of the shoulder-neck region suggested a developmental defect. At the same time, selective weakness of muscles innervated by C5-C6 segments suggested a neuropathic disturbance. EMG revealed, in eight of 13 cases, clear evidence of denervation of muscles, but without any regenerative activity. The non-progressive nature of this disorder and capacity for improvement in muscle bulk and power suggest that denervation alone cannot explain the process. Re-examination of three patients after two to three years revealed persistence of the major deformities and muscle weakness noted earlier, with no appreciable deterioration.

Otto (1841) appears to have been the first to recognize this condition. Decades later, Magnus (1903) described it as multiple congenital contractures with muscle defects. In 1913, Rocher gave the designation 'multiple congenital articular rigidity' and suggested that intrauterine pressure, such as that due to hydramnios, might restrict foetal movement and produce joint changes secondarily. In 1923, Stern proposed the term 'arthrogryposis multiplex congenita' (AMC), from two Greek words, meaning a 'crooking' of joints. Up to this time, joint deformities were generally considered the primary defect. There are also some reports in the literature suggesting a primary muscle disorder. These are reviewed by Dastur, Razzak, and Bharucha (1972).

It was not till 1933 that attention shifted to the central nervous system as the primary site of pathology in AMC. In this year, Price (1933) and Scarzella (1933) independently described diminution in size, degeneration, and a fallout of anterior horn cells in the spinal cord in cases of this disorder that went to necropsy. Since then, many papers have stressed the spinal cord changes in AMC (Gilmour, 1946; Brandt, 1947; Kanof, Aronson, and Volk, 1956; Fowler, 1959; and Swinyard, 1960). Anomalies of the cerebral cortex, such as abnormal gyri and dilated

ventricles, have been described (Adams, Denny-Brown, and Pearson, 1953; Fowler, 1959), in addition to the spinal cord changes.

In the past decade, with the use of electromyography, the nervous system and especially the spinal cord remained the focus of attention, as a large proportion of cases revealed denervation potentials (Lefebvre and Chaumont, 1957; Swinyard, 1960; Smith, Bender, and Stover, 1963; Amick, Johnson, and Smith, 1967).

Thus, by the mid-1960s, two major types of AMC were being recognized—the neurogenic and the myopathic. At times a primary process of maldevelopment of neuromuscular elements was also envisaged.

The study reported here was undertaken in an attempt to resolve some of these differences of opinion regarding the pathogenesis of this condition, through a combined clinical, electromyographic, and histopathological evaluation. The presence of limiting deformities at two or more joints (excepting talipes) from birth onwards, was considered essential for the diagnosis.

METHODS

During the three years 1966 to 1968, we examined and studied 15 cases (and added one of the older cases) of AMC, as observed at the Children's

Orthopaedic Hospital in Bombay. A detailed antenatal, natal, and postnatal history, with a family history and a careful evaluation of the developmental milestones, was obtained from the parents of each child. This was followed by a detailed neurological examination and, generally, a psychological evaluation. Radiographs of the bones and joints of limb and trunk were available in 13 cases.

Electromyography was carried out on 13 of the cases, using either a RAF type IIIC single channel or a Disa two-channel electromyograph coupled with a Tektronix type 564 storage oscilloscope, and concentric needle electrodes. A general sampling of one or two muscles each of the upper and the lower limb, usually including the deltoid and the quadriceps, was carried out in all cases. Nerve conduction velocity could be measured in only one case. In each muscle as many sites were needled as were necessary to study 10 motor units. With the RAF type IIIC single channel instrument only rough visual measurements were possible, but use of the coupled DISA and Tektronix instrument enabled accurate determination of amplitude and duration.

Three of these children could be recalled two to three years later and were examined carefully and repeat biopsy specimens obtained (Dastur *et al.*, 1972).

OBSERVATIONS

INITIAL EXAMINATION

HISTORY: POSSIBLE PREDISPOSING FACTORS *Genetic* (Cases 1, 8, 9, 15.) The parents of case 1 were related to each other; there was a sib with arthrogryposis multiplex in the family of case 8. The father and sister of case 15 had a congenital heart lesion, while an uncle of the same patient had bilateral talipes equinovarus (TEV). On the other hand, case 9 had an unaffected twin sister.

Pregnancy (Cases 1, 14.) In the majority of cases, this was quite normal. However, a rash all over the body in case 1's mother at the seventh month of pregnancy may have been due to measles. Another mother (case 14) had vaginal bleeding from the third to the seventh months of pregnancy. Two

mothers complained of abdominal pain 15 days and two months before delivery respectively.

Delivery (Cases 2, 8, 12, 13, 14.) It was surprising to note how frequently these children with fixed limb deformities were born at home without obstetrical assistance. Two were vertex presentations, with legs flexed at the hips and extended at the knees. One breech presenting baby also had legs extended at the knees. However, case 13 required a caesarean section after prolonged labour. During delivery of case 12 the head came out 20 minutes before the rest of the body, with the legs entwined around the neck and the arms tethered to the side by loops of umbilical cord.

There was no clear history of hydramnios or oligohydramnios in any of these cases. Some mothers however did complain of feebleness of foetal movements during this pregnancy compared with their other pregnancies. Cases 6 and 16 had a delay in crying after birth, while case 1 sucked poorly and had a feeble cry for the first month.

Developmental milestones A delay in motor development was present in 15 of 16 cases. The sole exception was case 7, who was seen only once at the age of 5 months, when it was difficult to establish motor retardation. In eight cases, there was also mental retardation, suggesting an associated cerebral defect. Isolated motor delay with normal mental status was present in the remainder, and here the gross mechanical handicap resulting from arm and leg deformities was probably the major contributory factor. This is particularly true of cases 4 and 14, where normal mental development accompanied normal early motor milestones with delay in standing and walking.

CLINICAL FEATURES All 16 cases of arthrogryposis multiplex congenita in the present series showed deformities of two or more joints from birth. Their ages at the first examination varied from 2 days to 15 years and there were nine males and seven females. The salient clinical findings are summarized in Table 1.

TABLE 1
SUMMARY OF MAIN CLINICAL AND ELECTROMYOGRAPHIC FINDINGS IN 16 CASES

Case no.	Sex	Age (yr)	Main deformities and/or contractures	Other clinical features		Electromyography				EMG diagnosis	Other information	
						Fibrillations	Motor unit potential					Interference pattern
							Duration (msec)	Amplitude (μV)	Polyphasic (%)			
1 A.L.	F	2 2/12	Arms: 'policeman-tip' Legs: mild flexion	Mild myopathic facies; proximal weakness	No	1-2	300	50	Full	Myopathic		
2 A.K.C.	F	8/12	Arms: normal Legs: marked extension with TE	Weak arms, proximal distal. Hip-movements fair despite dislocations	No	6	1000	None	Full	Normal		

TABLE 1—(continued)

Case no.	Sex	Age (yr)	Main deformities and/or contractures	Other clinical features	Electromyography				EMG diagnosis	Other information	
					Fibrillations	Motor unit potential					Interference pattern
						Duration (msec)	Amplitude (μV)	Polyphasic (%)			
3 M.N.	M	3 6/12	Arms: minimal flexion deformity at elbows Legs: in flexion	'Bird-like facies', first 3 fingers almost equal MR +	No	5	800	None	Full	Normal	Prominent soft tissue shadows in x-rays
4 M.R.G.	M	2	Arms: 'policeman-tip' Legs: almost normal	Right foot prehensile Takes a few steps when led. MR +	High frequency discharges	6	1000	100	Single oscillations	Neuro-pathic	
5 H.B.	M	11	Arms: 'policeman-tip' Legs: some flexion at hips, knees; ankles fixed in mid-position	Spina bifida occulta several operations done on all these 3 joints; walking with crutches	—	—	—	—	—	—	
6 L.P.S.	F	1 3/12	Arms: 'policeman-tip' Legs: kept in 'diamond shaped position	Stands with support	No	6	1000	10	Single oscillations	Neuro-pathic	
7 J.R.A.	M	5/12	Legs: in flexion, with 'hockey-stick' feet	Bilateral hip dislocation and spina bifida	—	—	—	—	—	—	
8 A.K.	F	3	Arms: only slight elbow flexion Legs: knees—posterior dislocation with hyperextension	Drooping eyelids Bilateral hip dislocation with lateral rotation of leg. MR +	No	1-3	800	25	Full	Myo-pathic	One older sib similarly affected
9 M.G.	F	5 9/12	Arms: normal (mild spasticity of left arm) Legs: marked lordosis with hips flexed, knees extended	MR—mild VSD—heart	Yes	14	400	100	Single oscillations	Neuro-pathic	One twin sister normal
10 D.G.M.	M	1 2/12	Arms: 'policeman-tip' Legs: in extension	No power, except some at hips	No	6	600	10	Single oscillations	Neuro-pathic	
11 T.M.	M	3	Arms: mild, modified 'policeman-tip' Legs: generalized flexion	'Bird-like' facies, kyphoscoliosis Walks a little bent forward. MR +	No	5	200	None	Full	Normal	
12 S.D.	M	3 10/12	Arms: 'policeman-tip' Legs: hips extended, knees flexed, TEV	Odd facies with hypognathism. MR—mild	—	—	—	—	—	—	
13 K.P.	F	11	Arms: 'policeman-tip' at birth; surgical corrections later Legs: only ankle plantar flexed	Marked wasting of C5-C6 muscles. At regular school in VI standard. Walks with support	No	6	800	10	Single oscillations	Neuro-pathic (upper and lower limbs)	
14 S.M.S.	F	2	Arms: 'policeman-tip' Legs: generalized flexion. Bilateral TEV	Arms and calves extremely thin. Shuffles on bottom. MR +	No	10	600	50	Single oscillations	Neuro-pathic	
15 C.K.	M	16	Shoulders: elevated and forwards, merging with neck Legs: fixed in extension	Arms very muscular but poor abduction Legs: several operations; walks with crutches	Yes	10	1500	50	Single oscillations	Neuro-pathic	Good improvement with rehabilitative and surgical procedures. Studying well
16 R.V.D.	M	1 6/12	Arms: 'policeman-tip' Legs: mild generalized flexion; bilateral TEV	Most joints fixed; all movements very poor. MR +	—	—	—	—	—	—	

TEV = talipes equinovarus.

MR = Mental retardation.

VSD = Ventricular septal defect.

FIG. 1. Case 7. Typical case with arms in 'policeman-tip' position; legs flexed at the hips with 'hockey-stick' deformity of the ankles.

FIG. 2. Case 11. Bird like facies, generally flexed attitude. Arm in 'policeman-tip' position.

DEFORMITIES Face In some of the cases (especially nos. 3, 11, and 12) the striking features were large protruding eyes, a bulging forehead, small pointed bird-like nose, small tight mouth with thin lips, a poorly developed chin (hypognathism), and receding jaw (Figs 1-4).

Figure 5 shows the distribution of the deformities. In the majority (13 cases) all limbs were involved, in the remainder only two were affected (upper limbs in one case, lower limbs in two cases). The limb postures consisted of universal flexion (five cases), a universal extension (one case) and a combination of flexion and extension (10 cases).

Neck and shoulder The neck and shoulders of these children frequently showed a peculiar configuration. The scapulae were often elevated, laterally displaced, with the shoulders pointing forwards and winging of the inner border of the scapulae (Fig. 2) (cases 5, 11, 12, 13). This often gave rise to a short neck (case 15).

In some cases the neck and the narrow rounded shoulders sloped smoothly into the arms without the presence of shoulder tips (cases 7, 15). In addition, in case 15, the ill-defined borders of the sternomastoid muscle merged into the posterior neck muscles.

Upper limbs In nine of the 16 cases the position was one of adduction and inversion at the shoulder, pronation at the elbow and flexion at the wrist and fingers (Fig. 1). This so-called 'policeman-tip' position was accompanied by extension at the elbow in six cases and flexion in three. In two cases the arms were normal. Mild flexion at the elbow was the sole defect in another case and a mild adduction deformity of the shoulder in one instance. In three of the remaining cases there were minor degrees of flexion, at the wrist and fingers, forming a C-shaped curve of the hand and fingers.

Lower limbs The common position at the hip was one of flexion (Fig. 3) (six cases), or external rotation (Fig. 4) (two cases). In one case each, external rotation was combined with abduction or with flexion of the hip. In one case the hips were extended.

The knees were either flexed (Fig. 2) or extended

FIG. 3. Case 9. Marked lordosis with hips flexed, knees extended; note one ankle plantar flexed and one dorsiflexed.

FIG. 4. Case 8. Only slight flexion at elbow. Bilateral dislocation with external rotation of the hips, with hyperextension and posterior dislocation of knees.

(Fig. 3) in 14 of the cases. At times the knees were hyperextended, with a posterior bulge (Fig. 4). The ankles and feet were in a position of talipes equinovarus (10 cases), pes calcaneus (one case) or a combination of these with one deformity on each side (Fig. 3) (one case). One patient had a very unusual deformity, the foot being adducted and inverted looking rather like a hockey stick.

NEUROLOGICAL SIGNS *Face and neck* Case 1 had a myopathic facies, with weakness of the neck muscles. Case 8 showed drooping eyelids.

Motor power (In minimally affected joints.) Proximal weakness and wasting were met with only in one case (case 1). This child also had a myopathic facies.

The power at the shoulder and hip was much less than at the elbow and knee.

In most cases a fair degree of movement was possible at the shoulders and hips. However, weakness and wasting (or poor development as will be discussed) were much more a feature of the shoulder girdle muscles than of those of the hip where the power was good even with restricted movement. This weakness and wasting varied in distribution. Thus case 6 had very thin deltoid muscles with well-formed scapular muscles. In cases 12 and 14 the entire shoulder girdle musculature was weak. In case 13 all the muscles supplied by C5 and C6 nerve roots including the biceps and brachioradialis muscles were affected. The shoulder girdle weakness was also present in cases 1, 2, and 5. In cases 4 and 7,

FIG. 5. Diagram showing the pattern and incidence of deformities at six major joints of the limbs and in the neck in 16 cases. (The small digits indicate the serial number of cases.)

there was tethering of the arm by a pectoral strap causing restricted movement at the shoulder with corresponding weakness.

Case 12 showed wasting of the right thigh muscles. In case 9 the left arm was small and spastic, with athetoid movements, and there was a history of left-sided focal convulsions and presumably an infantile hemiplegia in addition to the arthrogryposis multiplex.

Deep reflexes These were rarely obtainable even in the minimally affected limb. The knee jerks were brisk in cases 7 and 13, the ankle jerks in case 8, and both knee and ankle jerks in case 14. The arm jerks were obtainable in cases 8, 9, and 15. The plantar responses were either flexor or absent.

Effective function Six patients were walking with crutches and four others with manual support, generally after many orthopaedic corrective procedures. Similarly, five patients had an almost normal grasp, in addition to two whose upper limbs

were unaffected. Some patients, notably case 15 (Fig. 6), improved considerably with physiotherapy and corrective surgical procedures carried out over the years.

RADIOGRAPHIC FINDINGS AND ASSOCIATED ANOMALIES These were available in 12 of the 16 cases. In three (cases 6, 13, 14) radiographs of the bones and joints of both upper and lower limbs were within normal limits. The findings in the other nine cases were as follows: spina bifida occulta in three cases (nos. 5, 7, 12), lumbar lordosis in one (case 9), unilateral or bilateral dislocation or subluxation of hip joint (Fig. 7) (cases 2, 5, 7, 8, 9, 12); coxa valga (case 12), anterior dislocation of the knee (case 8), and a valgus deformity (case 12); mid-tarsal dislocation of the foot (case 2) and valgus deformity in another (case 5); unilateral subluxation of the shoulder (Fig. 8) (case 4), anterior dislocation of the elbow (case 4) and subluxation at the radio-ulnar joint with bowing of the bones of upper and lower extremity (Fig. 9) (case 11). Prominent subcutaneous soft

FIG. 6. *Case 15. Scapular and arm muscles over-developed with exercise; winging of scapulae; legs straightened after repeated operations.*

FIG. 7. *Case 8. Dislocation of the hip.*

FIG. 8. *Case 4. Subluxation at the shoulder joint.*

FIG. 9. *Case 11. Subluxation at the radio-ulnar joint.*

tissue shadows were observed in two cases (nos. 3 and 4). Thus the joint affected most often was the hip and the changes observed there were due both to an ill-formed head of the femur and to a shallow acetabulum.

In addition, clinical examination revealed pigeon chest in one (case 1), kyphoscoliosis in two (cases 5 and 11). There was a ventricular septal defect in case 9 and a congenital inguinal hernia in case 16.

ELECTROMYOGRAPHIC FINDINGS Electromyography (EMG) performed in 13 of the 16 cases, showed a clear neuropathic pattern in eight cases, a myopathic picture in two (cases 1 and 8). It was normal in three (cases 2, 3, 11). The main findings are summarized in Table 1.

It will be seen that the essential neuropathic

units. Even clinically normal muscles showed myopathic changes in case 8.

SUBSEQUENT EXAMINATION

Three of the patients, cases 3, 4, and 14, could be recalled for a follow-up study. Patient 3, now aged 6 years 9 months, still has the same 'bird-like' facies and essentially the same mild degree of flexion deformity at the elbows and hips. There was some weakness of flexor and extensor muscles of the forearm and to a lesser degree of the flexors and extensors of the hip and knee. The fingers were kept extended at metacarpophalangeal and flexed in contracture at the distal interphalangeal joints, with only a pincer-like action possible with thumb and index finger.

FIG. 10. Case 9. Single unit pattern on electromyography. For details see Table 1.

features observed in the eight cases (4, 6, 7, 9, 10, 13, 14, 15) were fibrillation potentials in three, and loss of interference pattern with only single unit activity in all—for example, Fig. 10. Five of these cases showed 50–100% polyphasic units. While the duration of these potentials was prolonged in all cases (up to 14 msec), their amplitude was *not* increased in any case. Thus, there was no evidence of giant motor units—that is, units over 5 mV in amplitude. Neuropathic changes were observed even in clinically normal muscles. The nerve conduction velocity was measured only in case 15. It was normal in the upper limb where denervation potentials were obtained from the muscle but no response could be obtained on nerve stimulation in the lower limb, the muscles of which were also totally unresponsive to voluntary innervation.

The two cases (1 and 8) with a myopathic picture were characterized by typical short-duration (average duration less than 4 msec) and low-amplitude motor unit potentials (average less than 300 μ V) with full interference pattern, and up to 50% polyphasic

Patient 4, now aged 4 years, is still short for his age but walks well and can run a little, he still uses his feet as prehensile organs. The upper limbs persist in the 'policeman-tip' position, with stiffness at the elbows, in extension on the right side, and in fixed flexion on the left. The upper limb muscles are still flabby and undefinable as before, the biceps being more affected than the deltoid muscles.

Patient 14, now aged 4½ years, weighs only 8 kg and still maintains the 'policeman-tip' position of both upper limbs with elbows extended. Muscles were as flabby as before and pectoral straps tethering the arms to the sides were noticed. Though the hips were still kept at 20° of flexion, the left knee was found straightened (by fasciotomy). There was partial correction of talipes equinovarus by plastering bilaterally. She could stand with support.

DISCUSSION

To make a better assessment of the disabilities resulting from the deformities of the limbs, which constituted the major presenting feature

of our cases of AMC, an attempt was made to correlate them with the functional status of the limbs. Deformities were graded in the arms and legs as I, II, and III, according to the number of joints affected. The functional loss in the upper limbs was graded as A, B, or C, according to the ability to grip with the hands. Thus with the severest degree of loss (grade C), no prehension was possible with the hands. With grade B, a feeble grasp was possible, while with grade A, a fairly good grasp was present. In the legs, if the child was over 1½ years and could not walk, the functional loss was considered to be grade C. With grade B loss, walking was possible with appliances, and with grade A, only a stick or slight support was required. In children under 1½ years who had not yet started to walk, the power was graded according to the ability to kick freely while lying in bed. Table 2 shows these relationships for the arms and legs.

TABLE 2
RELATION OF DEFORMITY TO FUNCTION

<i>Deformity</i>	<i>Case no.</i>	<i>Functional loss</i>	<i>No. of cases</i>
A: in arms (16 cases)			
Normal	2, 3	Nil	2
Grade I	5, 8, 9, 15,	Normal	1
		Grade A	2
		Grade C	1
Grade II	7, 11, 13	Grade A	2
		Grade C	1
Grade III	1, 4, 6, 10, 12, 14, 16	Grade C	7
B: in legs (16 cases)			
Normal	4	Nil	1
Grade I	11, 13	Grade A	1
		Grade B	1
		Grade A	2
Grade II	3, 6, 7, 8, 10, 14	Grade B	2
		Grade C	2
		Grade B	3
Grade III	1, 2, 5, 9, 12, 15, 16	Grade B	3
		Grade C	4

The important joints in the upper limbs appear to be those of the wrist and fingers. As the Table shows, with only one pair of joints involved (grade I deformity), if the wrist and fingers were affected there could be a grade C functional loss. Contrariwise, a grade II deformity involving the elbow and shoulder only might be compatible with grade A function. In the legs there was greater correlation between the number of joints involved and functional incapacity. However, a person with all three joints

involved may walk with support, provided that the feet sit well on the ground, the knees are fixed in extension, and some movements are available at the hip. Hence, occasionally it is found (cases 6, 8) that the functional incapacity is less than the degree of deformity would have warranted.

There were two features of note in the follow-up study of three of the children, carried out two to three years after the initial examination. The major deformities associated with actual fibrotic contractures—for example, the ‘policeman-tip’ position, had remained in status quo. The overall growth of the children was also poor. However, there was no deterioration in any specific movement, deformity, or function of a muscle group.

Our ideas on the pathogenesis of arthrogryposis multiplex congenita will be elaborated in Part 2 but it appears necessary to draw some inferences from the clinical and electrical data. It is noteworthy that even clinically, most of our 16 cases did not reveal any features suggestive of a myopathic disorder. While nine of the cases evidenced weakness of the shoulder girdle, only two of these presented bilateral facial weakness and proximal muscle weakness of the limbs consistent with a myopathy. Only these two patients showed EMG evidence of myopathy (Table 1). The fact that the proximal weakness of the upper limbs did not equally involve all the abductor muscles of the shoulder spoke against a typical muscular dystrophy. Thus, for instance, either the deltoid alone was weak, or the deltoid and spinati muscles were affected. Furthermore, the weakness also tended to involve the biceps and brachialis muscles, creating the impression of a C5–C6 segmental lesion of the spinal cord. The most frequently observed picture of the upper limbs in these cases—namely, the ‘policeman-tip’ position, seemed to have been brought about by this selective muscular involvement, and was reminiscent of upper brachial plexus birth injury as in Erb’s palsy.

The majority of cases also did not manifest any clear signs of a neurogenic disorder. However, it is realized that against a background of multiple joint deformities and contractures which are most pronounced distally, the demonstration of distal weakness and the elicitation of deep and superficial reflexes are bound to be difficult or impossible. Fasciculations were not clinically demonstrable. Nevertheless, as revealed by electromyography, eight of the 13 patients had evidence of a denervation process. While single

motor unit potentials were constantly present and fibrillations were frequent, giant polyphasic motor unit potentials (indicating regeneration in the motor units) were never detected. Considering the fact that a number of sites in several muscles were examined, the absence of such re-innervation seemed significant, being unusual for most motor neurone diseases of long duration.

There are some clinical and surgical features also pointing in the direction of a primary defect in muscle formation. The characteristic position of the shoulder region—namely, high placed laterally rotated scapulae with a short neck merging with the arms without clear shoulder tips—points to an incomplete embryonic descent of the scapulae and lack of differentiation of the shoulder girdle muscles. Obvious congenital malformation of the hip joint may occasionally be accompanied by retention of a fair amount of power in the muscles of the hip. This suggests a primary developmental defect or a dysplastic process. Clinical improvement is noticed over the years with gain in muscle bulk and power, as the infant with AMC grows up, especially with the help of physiotherapy, as for instance in our case 15.

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