ARTHROMYODYSPLASIA CONGENITA
SIMULATING THE ARTHRITIC MANIFESTATION OF
"RHEUMATOID DISEASE"

BY

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"Rheumatoid disease" in its acute or chronic form with systemic or local manifestations may simulate many diseases in general medicine. Local manifestations may resemble many specific arthropathies and bony and muscular lesions with secondary joint involvement (Ellman, 1947; Ellman and Ball, 1948), and two cases of a chondro-osseous dystrophy, which had been regarded as an arthritic manifestation of "rheumatoid disease", have already been reported (Ellman, 1949).

The purpose of this paper is to give an account of a unique case of so-called "amyoplasia congenita" or "arthrogryposis multiplex congenita", which we, for reasons to which we shall refer in the discussion, prefer to designate "arthromyodysplasia congenita". This condition occurred in a road sweeper aged 40, who was initially referred as a case of "rheumatoid arthritis". The flexion deformities of fingers, wrists, elbows, and knees, and the fixation of these joints, give the impression of a fibrous ankylosis, hence the terms "multiple articular rigidity" and "arthrogryposis multiplex". As we shall show, the symmetrical joint immobility and flexion deformity are never associated with any inflammatory change, but depend upon some congenital developmental defect, probably associated with a dysplasia of certain groups of muscles with changes occurring in and around the joints.

Case Report

Male, aged 40, single, roadsweeper, was admitted to the Rheumatism Unit at St. Stephen's Hospital as a possible case of "rheumatoid arthritis". At the time of admission he was complaining of pain and stiffness in the knees, ankles, and toes, together with occasional aching of the shoulders and upper limbs in general for the past 2 months. In addition to his polyarthralgia he had been complaining of some breathlessness on exertion.

His past history showed that ever since he could remember he had had several severe joint deformities, in fact since the age of 2 ½ years when he had attended the paediatric department of a London hospital for an apparently congenital condition of his limbs. He had been under their supervision until he had reached the age of 16, but in spite of these very marked deformities he had worked as a road sweeper until 7 weeks before admission to hospital. There was no relevant family history.

Clinical Examination.—Pupils equal and reacted to light and accommodation; disks healthy; no evidence of disease clinically in heart, lungs, abdomen, or central nervous system. Blood pressure 130/80. No clinical evidence of anaemia. No lymphadenopathy, hepatomegaly, or splenomegaly.

X-Ray Examination of the heart was within normal limits.

Electrographic Examination normal.

Locomotor System.—Severe limitation of movement in elbow joints which were almost fixed at a right angle. Flexion deformities of hands and wrists (Fig. 1). No abnormality of the spine. Hip joints considerably restricted in range of movement. Knees similarly restricted, the right one being painful. Ankles also limited to a few degrees of movement in the middle range (Fig. 2, overleaf). Joint lesions all of a secondary degenerative nature.

Fig. 1.—Flexion deformities of hands and wrists.
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Fig. 2.—Body posture (anterior and lateral views), showing flexion deformities in upper and lower limbs.

Fig. 3.—Secondary degenerative joint changes involving ankles.

Fig. 4.—Right shoulder, showing presence of several loose bodies.

Range of movement in the joints:

**Elbows:**
- Right: Extension limited to 85°.
- Flexion full, 150°.
- Left: Extension limited to 80°.
- Flexion 80-135°.

**Shoulders:**
- Both: Abduction to 60°.
- Internal rotation full.
- External rotation limited.
- Flexion to 90°.

**Wrists:**
- Both: Dorsiflexion 10°.
- Plantar flexion 70°.

**Hands:**
- All fingers flexed to 70°.

**Knees:**
- Right: Flexion 80°.
- Extension 150°.
- Left: Flexion 70°.
- Extension 150°.

**Ankles:**
- Both: Dorsi-plantar flexion limited.
- Range 30°.

**Hips:**
- Both: Flexion 50°.
- Abduction to 17 in. between feet when standing on one.
- Abduction to 20 in. between feet when both abducted.
Fig. 5.—Hands, showing short metacarpals and flexion deformity with secondary degenerative changes.

X-Ray Examination confirmed the clinical findings and showed degenerative changes in all joints, especially the knees. These changes were regarded as secondary to an older lesion which had occurred during the period of growth. A noteworthy radiological feature was the existence of several loose bodies in the shoulders and elbows. There was a generalized trabeculation of the bone with some generalized osteoporosis of the epiphyseal ends. The bones showed evidence of a very uneven growth. The metacarpals were short. The dorsilumbar spine showed no abnormality. In the elbows the upper ends of the radii were ankylosed to the ulnae, and above each and apparently attached to it was a body resembling a radial head (Figs 3, 4, 5, and 6).

Pathological Investigations:
Blood-count: normal.
Blood sedimentation rate (Westergren): normal.
Blood urea, plasma uric acid, plasma proteins: all within limits of the normal.
Alkaline phosphatase: 17.2 units per 100 ml.
Liver function tests: normal.
Wassermann reaction and Kahn test: negative.

Discussion
This case of amyoplasia congenita (Sheldon, 1932) or arthrogryposis multiplex congenita (Stern, 1923) is apparently the only case yet described in a patient as old as 40, still actively employed in manual labour. (Albeaux-Fernet and Weissenbach, 1952, described a case in a man, aged 23, observed from birth.) In regard to terminology we prefer the recently suggested name, arthromyodysplasia congenita (Sürder, 1952). It is impossible to say that the joints are affected secondarily to the muscles, or that the muscles are affected secondarily to the joints. It is more likely to be a primary developmental abnormality of both the muscles and the joints. Many other names for the disease have been
employed (see Parkes Weber, 1947a, b; Kallio, 1948; Keizer, 1949; and Hagberg and others, 1952). The disease occurs in widely different forms and degrees, and in combination with various other developmental abnormalities or syndromes.

Sheldon (1932), in his paper on amyoplasia congenita, recorded the case of a child, aged 2 yrs, with congenitally rigid arms and legs, associated with aplasia of certain muscle groups. He stated that under the names "multiple congenital articular rigidity" and "arthrogryposis multiplex congenita" a rare but well defined condition had been described:

The characteristic features consisted of immobility of one or more joints of the limbs, generally symmetrical in distribution, and dating from intra-uterine life. The immobility may be absolute, or movement may be severely limited. The fixation of the joints has the clinical appearance of fibrous ankylosis, but evidence of inflammatory change to account for this is absent, and it would appear more probable that the condition depends primarily upon some developmental defect. In this connection the incomplete development, or even entire failure of development of certain groups of muscles in the limbs, which has been recorded in cases specifically examined from this point of view, has been a striking feature.

Sheldon thought that the most likely explanation was that the initial defect was a developmental aplasia or dysplasia of certain groups of limb muscles, secondary developmental changes occurring in and around the joints leading to the clinical picture of fibrous ankylosis. This does not signify that congenital abnormalities may not sometimes be caused by faulty position, fold, bends, etc., in utero.

Amongst recent writers using the term arthro-
gryposis multiplex congenita are Kallio (1948), Keizer (1949), Metcalfe (1951), Albeaux-Fernet and Weissenbach (1952), and Hagberg and others (1952); Jeune and Bruel (1950) prefer the name amyoplasia congenita.

Middleton (1934) prefers to call the condition myodystrophia foetalis deformans. He draws an analogy between it and the muscular dystrophies of post-natal life. He says it was first described by Otto (1841).

In 1929 Sir Heneage Ogilvie and his resident medical officer, F. J. Lees, showed Parkes Weber some cases at St. Vincent’s Orthopaedic Hospital (Parkes Weber, 1947a, b). In Ogilvie’s series the occurrence of associated developmental abnormalities was noteworthy. Thus, in one of the female patients, there was a urachus abnormality, the urinary bladder reaching nearly up to the umbilicus.

Another patient had a brother or sister affected with meningocoele or encephalocoele, and another seems to have had a fellow sib with cleft palate.

Dr. D. M. Greig, of Edinburgh, kindly directed our attention in 1929 to investigations by Hutt and Greenwood (1929) on embryonic mortality in the fowl, and chick monsters in relation to embryonic mortality. Greig wrote to one of us (F.P.W.) as follows:

In addition to cranial and facial deformities there are included congenital malformed limbs, thickened and flattened tarsometatarsus, unilateral absence of muscles, and absence of one or two toes. The joints are bent on account of the muscular anomalies and many cases of twisted feet and toes are mentioned as occurring, but are not described.

Greig himself had seen the chicks and had no doubt of the congenital distortions of the limbs.

Rocher (1913), in his comprehensive account of the disease referred to by Sheldon, pointed out that first-born children are not especially affected and that in his cases there was no familial or hereditary tendency. He directed attention to abnormalities of the hands and feet, notably flexion of the fingers, claw-hand, and club-foot; shortening of the flexor tendons of the fingers has been reported as in Volkmann’s ischaemic contracture, flexion of the fingers being more easily performed when the wrists are also flexed. The patella is often abnormal, being displaced, small or even absent.

The muscles never show a reaction of degeneration, so that the muscular atrophy is probably not of nervous origin, but the reactions to faradism and galvanism are diminished or absent, indicating hypoplasia or complete aplasia of muscle. There are no sensory or trophic changes. The tendon reflexes are naturally difficult to obtain, but when present, they are not increased.

The children in his cases were of normal intelligence. He also noted the thickened appearance of the subcutaneous tissues obliterating the normal bony markings in parts of limbs, but in a case reported by Magnus (1903) this subcutaneous thickening was absent, and the muscular aplasia was so great that the child appeared to be simply skin and bone. Rocher stated that there might be some shortening of the affected limb or segment of a limb. Amongst associated conditions there might be ankylosis of the mandible and some vertebral stiffness or scoliosis. A later paper (Rocher and Ouarry, 1930) recorded the case of a girl, aged 3 months, with fixation of the legs in extension and double talipes, associated with several malformations of the lumbar vertebrae and aplasia of the sacrum. According to these authors there may be abnormality of the synovio-capsular arrangement of joints.

Moncrieff and Wiles (1934), observing that Middleton (1934) had noted the resemblance of amyoplasia congenita to a sporadic disease occurring in sheep, wrote:

Should the two prove to be identical, it will be of great interest, because Fraser Roberts (1926, 1929) has been able to show by selective breeding that in sheep the condition depends upon the homozygous state of an autosomal recessive factor.

Edwards (1938), demonstrating an infant with “webbing of the lower limbs, associated with congenital bilateral contractions of flexor muscles of elbow and wrists” (which we suppose to be allied to arthromyodysplasia congenita), referred to the question of such “webbing” representing an atavism analogous to the webbed wing of a bat, and observed that similar folds are seen in the neck of a chimpanzee. He said that Bruns and Kredel (1890) maintained that the conditions of webbing owed its origin to misplaced and abnormal muscular developments, bridging the flexor surfaces of joints and displacing the overlying skin in web-like formation. Traces of muscular tissue are often encountered between these skin folds.*

In regard to associated developmental abnormalities, an interesting case was described by Herson (1947). The patient was a woman, aged 61 years, who in addition to amyoplasia congenita had a condition of hyperostosis frontalis interna. A more complicated case was that of a boy, aged 14 years, demonstrated by Williams (1948), in which there was maldevelopment of the osseous, muscular, and subcutaneous tissue, with central nervous system dysfunction.

Various questions arise from the consideration of arthromyodysplasia congenita:

* In this connection compare also scattered literature on "Brevicollis", "Klippel Feil Syndrome", and "Webbed Neck". 

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Perhaps some abnormalities of the hands, such as congenital camptodactyly, with or without webbing (compare Parkes Weber, 1938, 1947a) might be regarded as minor varieties of arthromydysplasia congenita. We would instance especially the case of a man, aged 47 years, with congenital or early developmental camptodactyly of both little fingers and considerable atrophy (or more probably hypoplasia) of the intrinsic muscles of the hands, who had likewise had a kind of facial telangiectasia of the Rendu-Osler type for as long as he could remember (Parkes Weber, 1938?).

Should the term arthromydysplasia congenita be used to include cases of localized muscular aplasia of the whole part of the pectoralis major muscle, of certain muscles of the abdominal wall, of bigger congenital defects of the thoracic or abdominal walls, and of fibrous dysplasia of a sternomastoid muscle?

May not some post-natal cases of local muscular dystrophy be regarded as representing a deferred arthromydysplasia congenita?

Is there a condition of developmental dysplasia of subcutaneous tissue analogous to, and sometimes associated with, arthromydysplasia congenita?

Middleton (1934) discusses the relation of arthromydisplasia congenita to congenital tibial kyphosis (congenital angulation of the tibia) and to congenital high shoulder (congenital elevation of the scapula, "Sprengel's shoulder"). Congenital hypoplasia or aplasia of the patellae have also been recorded in association with various types of congenital ectodermal defects (Parkes Weber, 1929).

The remarkable case described by Batten (1904) as "myositis fibrosa" might conceivably be regarded as a rare or even unique variant of arthromydysplasia congenita. The curious case described by Huber and others (1951) was apparently a chance combination of arthromydysplasia congenita with a hypervitaminosis D2 in the foetus, the latter condition being due to the mother having taken large doses of vitamin D2 throughout her pregnancy.

As stated above, we prefer the name arthromo- dysplasia congenita (Sürder, 1952). Moreover, we regard the term "dysplasia" as preferable to "aplasia" in most of the recorded cases. Sürder points out that, though there is abundant evidence of the occurrence of familial congenital contractures of joints in animals, these contractures are rare in human beings. In solitary cases, without any evidence of familial or hereditary tendency, neither exogenous causes, nor new mutations can be absolutely excluded.

There seems to be no evidence that the disease is ever due to intra-uterine toxaemia or infection due to an infectious disease in the mother, such as rubella at an early stage in the pregnancy.

Confusion with changes due to the rheumatoid or osteo-arthritic type of arthritis may occur, but as the foregoing shows, this should create no real problem.

In regard to treatment, little can be said. Though most authors mention that no treatment has been found of any use, Metcalfe (1951) mentions as useful Sir Thomas Fairbank's advice in the case of infants to stretch the tight tissues gently two or three times a day.

Summary

A case of arthromydysplasia in a man, aged 40, is described. The causation, symptomatology, nomenclature, and literature of the disease are discussed, together with its not infrequent association with other developmental abnormalities. The possible confusion of this condition with rheumatoid disease is noted.

Various minor varieties of congenital (or early developmental) contracture deformities of the extremities deserve to be regarded as possible slight cases of arthromydysplasia congenita. Foremost amongst these are examples of congenital camptodactylia with or without "webbing" and "hypoplasia" of the corresponding muscles. There is no reason why such slight developmental abnormalities should hinder the attainment of ordinary longevity.

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RÉSUMÉ
On décrit un cas d'arthromyodysplasie chez un homme de 40 ans. On en discute l'etiologie, la symptomatologie, la nomenclature et la litterature, ainsi que le fait que cette maladie est souvent associee a d'autres anomalies evolutives. On note qu'elle prête à confusion avec la maladie rhumatismale.

De différentes variétés mineures de contracture avec déformation congénitale (ou acquise précocement) des extrémités devraient être considérées comme des cas probables d'arthromyodysplasie congénitale légère. Un des meilleurs exemples de tels cas est la comptodactylie congénitale, avec ou sans palmeadura, et "hypoplasie" des muscles correspondants. Il n'y a pas lieu de croire que ces anomalies évolutives légères affectent la probabilité de survie.

SUMARIO
Se describe un caso de artromiosdisplasia en un hombre de 40 años. Se discute su etiología, sintomatología, nomenclatura y literatura, así como el hecho de que esta enfermedad se ve frecuentemente en asociación con otras anomalías de desarrollo. Se nota la posibilidad de confusión de este disturbio con la enfermedad reumática.

Diferentes variedades menores de contractura con deformidad congénita (o evolutiva precoz) de las extremidades deberían considerarse como casos probables de artromiosdisplasia congénita ligera. El primer lugar entre ellos ocupan los ejemplos de comptodactilia congénita, con o sin palmeadura, y "hipoplasia" de los músculos correspondientes. No hay razón de pensar que estas anomalías evolutivas ligeras afecten la probabilidad de supervivencia.
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