ARTHROGRYPOSIS MULTIPLEX CONGENITA

An Essay

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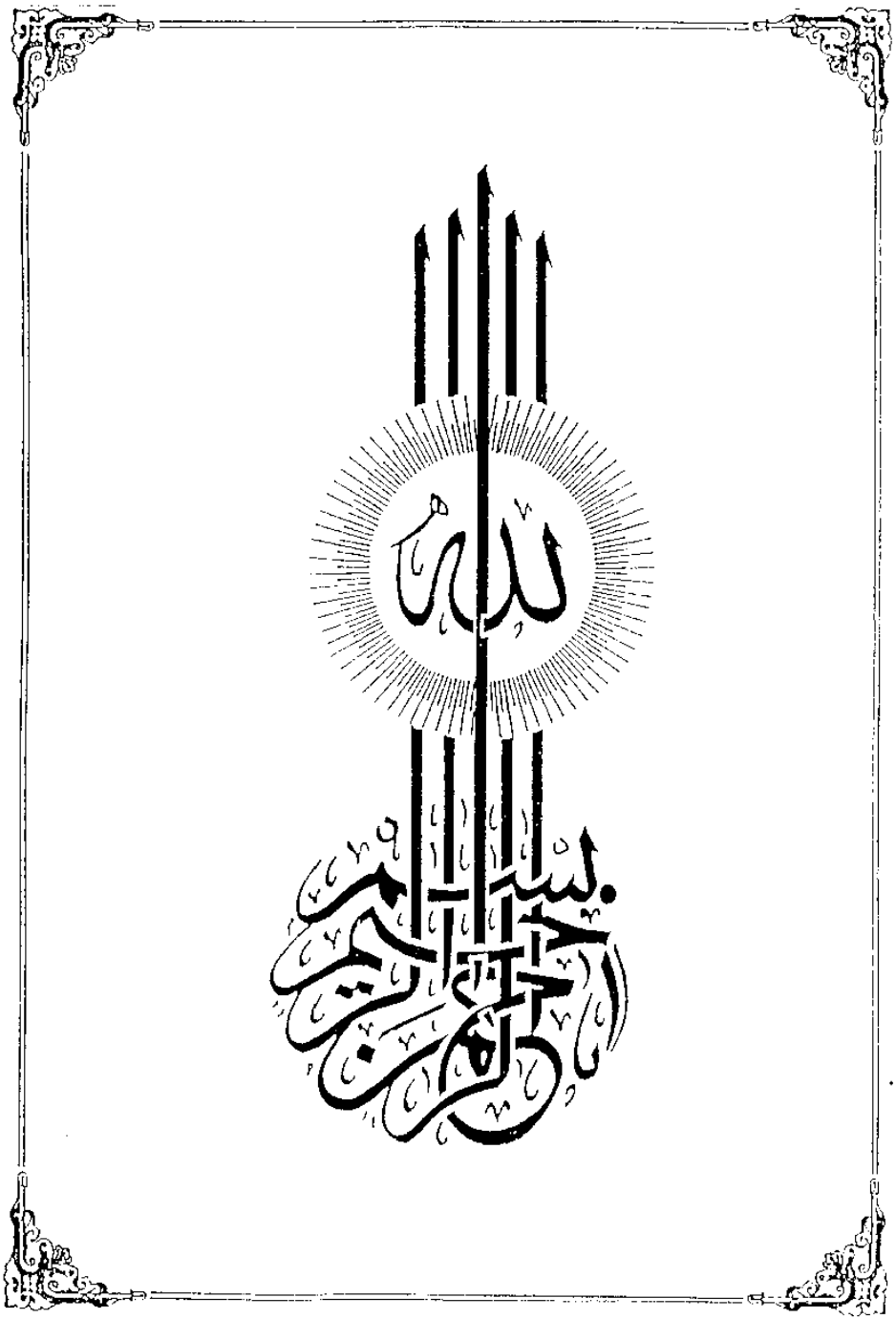
TO

MY MOTHER



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INTRODUCTION

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The syndrome of arthrogryposis multiplex congenita has been an enigma to orthopedic surgeon for decades.

The congenital joint contractures and frequent dislocations have been some of most challenging and difficult deformities to manage.

With advances in pathology, it has been demonstrated that any lesion affecting the final neuromuscular pathway during the formation of the limb in utero can result in congenital contractures.

Thus, lesions of the anterior horn cells roots, peripheral nerves, motor end plates and the muscles themselves are capable of producing congenital joint contracture provided that it occurs early enough in the fetal life to immobilize joints at various stages in their development.

Many of these disorders have other associated congenital abnormalities that can be as difficult to treat as the contractures themselves.

<u>The aim of this work</u>

Is to provide the current cencepts of arthrogryposis multiplex congenita regarding the aetiology, diagnosis, pathology, and management.

DEFINITION

* Arthrogryposis multiplex congenita [A.M.C] is a syndrome of non progressive but persistent joint contracture presenting at birth ***(Pellegrini, 1990)***.

HISTORY

* ***Otto***[*] ***(1841)***, was the first one to describe the condition, he described it as congenital myodystrophy, in ***1905 Rosenkranz*** was the first to use the term arthrogryposis, which is a Greek word meaning curved joints or hooked joints, however, in ***1923 Stern*** initiated the term Arthrogryposis multiplex congenita.

* For many years the condition was considered to be a discrete clinical entity or diagnosis. But in recent years,further studies have revealed more than 150 specific entities can be associated with what has been called A.M.C in the past. Thus, as suggested by Swinyard and Bleck, Multiple congenital contractures (M.C.C) is a more appropriate name for the condition ***(Ingram, 1987)***.

(*)Quoted from clinical orthopaedic and related research, 1985.

[Table 1]

Chronological list of descriptive terms that have been applied more than one time to designate M.C.C. in human Neonates *(Swinyard & Bleck, 1985)*.

Otto	1841	Congenital myodystrophy
Schanz	1897	Multiple Congenital Contractures
Rosenkranz	1905	Arthrogryposis
Novi-josserand and Rendu	1906	Multiple Congenital Articular rigidity
Howard	1907	Dystrophia muscularis congenita
Stern	1923	Arthrogryposis multiplex congenita
Sheldon	1932	Amyoplasia Congenita
Middleton	1934	Myodystrophia foetalis deformans
Dalmain	1947	Myophagism
Rossi	1947	pterygorthromyodysplasia
Lowenthal	1952	Hereditary myosclerosis
Rocher	1954	Dyplasia-myo-osteo articularis
Vellaavljev	1972	Guerin-stern syndrome

AETIOLOGY

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The deformities of arthrogryposis multiplex congenita [A.M.C] are known to occur as a component of a large group of heterogenous neurogenic and myopathic disorders as well as other disorders, such as skeletal dysplasia, the pathologic feature could be *a congenital or acquired defect in the motor unit* (anterior horn cells, roots, peripheral nerves, motor end plates, or muscle) producing severe weakness early enough in foetal life to immobilize joints at various stages in their development. Thus, persistent immobility leads to contractures as well as abnormal development of the joint surfaces.

The neurogenic form is the most common type accounting for over 90% of all cases and next to it is the myopathic type *(Swinyard et al., 1985)*.

Filogamo (1981) noted that mygonic determination of mesodermal tissue requires the influence of nerve fibers from motor neurons meaning that normal muscle development is dependent upon innervation.

Theories of Aetiology:

A) Connective tissue disorders:

Abnormal connective tissue specially collagen may interfere with the normal development of tendons, bones and cartilages. This can result in joint fibrosis and contractures.

Ionasescu et al.,(1970) demonstrated increased collagen synthesis in patients with A.M.C.

Ippolito and Ponseti (1980) observed increased fibrous tissue of the muscles, ligaments, fasciae and tendon sheaths in the feet of still born foetuses with club feet.

B) Mechanical limitation to intrauterine foetal movements:

This could be structural abnormalities in the uterus (myomata, malformations), amniotic bands, twins and oligo-hydramnios have also been associated with multiple contractures ***(Thompson, 1984)*** .

Mothers of newborn with A.M.C indicates that foetal movement was noted at the expected time but the character and intensity of movement significantly changed at variable times of the second trimester ***(Swinyard et al., 1985)***

A.M.C. with oligohydraminos:

Graham et al., (1980) have shown that mechanical fixation of limbs can result from pregnancy occurring in malformed uterus (bicornate) or by a fibroid tumor that encroaches upon available intrauterine space.

This observation suggests that loss of amniotic fluid for any reason brings the amniotic membrane into closer relation with the foetus restricting foetal limb movement and respiratory excursions.

Needle aspiration of amniotic fluid in pregnant rats on day 16.5 resulted in only 26% mortality, all surviving foetuses had growth retardation,multiple congenital contractures and short umbilical cord *(De Myer and Baird 1976)*.

C) Congenital infections:

Sever (1968), has indicated that there are some viruses known to be teratogenic in humans (Cytomegalo virus,rubella, herpes simplex and encephalitis).

The foetal neuro pathology of all these viruses is of a nature that A.M.C could result from such infections, especially that *Sever* has pointed out that immunoglobulins IgM are elevated in cases due to congenital infections.

Swinyard (1985) believes that too little attention has been given to the possibility that maternal infections transmitted to the foetus could be an aetiologic factor for A.M.C.

A.M.C. is well known to affect animals and it is known that it is lethal (because of inabilities of affected animals to feed themselves), Akabane virus is considered one cause of A.M.C. in animals (immunoglobulin in calves were found high, virus was recovered from brains and spinal cords of affected animals) now a formaline inactivated Akabane virus vaccine is available. However, there is no evidence that Akabane virus infection occurs in humans *(Kurgi et al.,1976)*

In laboratory animals prenatal contractures have been induced by viruses, neuromuscular blocking agents, toxins, insecticides, hyperthermia and limb immobilization *(Swinyard)*

D) A.M.C. with Polyhydraminos:

In syndromes related to Polyhydraminos,the contractures are related to either upper or lower motor neuron defects.

PENA-Shokeir (1974) described 2 types related to polyhydraminos.

PENA-Shokeir type I:

Pregnancy complicated by polydraminos and lung hypoplasia, decreased number of motor neurons, multiple contractures facial anomalies lowset malformed ears.

PENA-Shokeir type II:

Newborns had microcephaly, hypotonia, microopthalmus cataracts,large pinnae and multiple congenital contractures. Muscle biopsy showed increased connective tissue and fatty infiltration. Autosomal recessive inheritence was suggested.

E) **Miscellaneous:**

A.M.C. and neuromuscular blocking agents (curare):

In *1966 Drachman and Sokoloff* found that when a neuro muscular blocking agent was used to produce paralysis in chick embryos, joint cavities failed to develop even though joint formation had proceeded to the point where cavitation should occur. They also found that adventitious cartilages and sesamoid cartilages failed to develop.

A.M.C. and myasthenia gravis:

In *1961, Drachman and Barker* reported A.M.C associated with newborns having myasthenia gravis. This observation has been confirmed by several studies.

F) Genetics and A.M.C.:

There are only limited number of ways in which any condition can run in families (i.e. have genetic aspects).

The aetiologic and genetic basis of M.C.C. is very heterogenous (*Hall, 1985*)

These includes:

a) **Gene inheritence**

1. Autosomal dominant types:

I. Distal arthrogryposis -----> primarily hands and feet are affected, also knees, hips. hands with overlapping fingers clenched fist.

II. Coalitions, Synostosis and Symphalangism ------> many types of bony fusion or failure of bones to separate.

2. Autosomal recessive types:

I. **Multiple pterygium syndromes:-**

* Lethal type -------> hypoplastic lungs-fixed joints hypertelorism, polyhydraminos.

* Non lethal type ---> flexion contractures, clenched fists, foot anomalies, often scoliosis.