

# Ophthalmological And Systemic Findings Associated With Arthrogryposis Multiplex Congenita

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## Citation

R Sharma, P Sharma, J Raina. *Ophthalmological And Systemic Findings Associated With Arthrogryposis Multiplex Congenita*. The Internet Journal of Tropical Medicine. 2006 Volume 3 Number 2.

## Abstract

Arthrogryposis multiplex congenital (AMC) is a birth defect characterised by multiple joint contractures and is associated with various other congenital abnormalities. Various systemic associations like cleft palate, low set ears, gastrointestinal (GI) abnormalities, congenital heart defects, hypoplastic lungs and cryptorchidism have been described with this syndrome.

A detailed clinical examination comprising of ophthalmic, orthopaedic, paediatric, orthoptic, ENT, haematologic and neurologic review of two babies with AMC was undertaken.

These cases presented some never before described ocular and systemic abnormalities in association with AMC. We suggest a close multidisciplinary follow up on all cases of arthrogryposis multiplex congenita to detect, report and better understand these associations.

## INTRODUCTION

Arthrogryposis multiplex congenital (AMC) is a birth defect characterised by multiple joint contractures and is associated with various other congenital abnormalities. The syndrome was first reported by Otto in 1841 as “congenital myodystrophy”<sup>(1)</sup>. The reported incidence is 3 in 10,000 live births<sup>(2)</sup>. The abnormality according to Banker<sup>(3)</sup> was neurogenic in origin (93%) as opposed to myopathic origin (7%), but Strehl and Vanasse, studied 24 cases, who underwent muscle biopsy and EMG, which showed that 40% were myopathic disorder<sup>(4)</sup>.

Irrespective of these, there are two common features i) onset in utero and ii) primary alteration in one structure comprising the final common pathway (anterior horn cells, roots, peripheral nerves, motor end plates, or muscles) leading to decreased fetal movement during pregnancy. The affected muscles clinically appear normal, atrophic, or absent but histologically they usually show fatty or fibrous tissue infiltration<sup>(3)</sup>. Treatment of AMC involves physical therapy, serial casting and finally surgery<sup>(5,6)</sup>.

Various systemic associations like cleft palate, low set ears, gastrointestinal (GI) abnormalities, congenital heart defects, hypoplastic lungs and cryptorchidism have been described

with this syndrome. We present two babies with AMC and unfamiliar anomalies of sickle cell anemia and G6PD deficiency.

We present the unusual and never before reported finding of unsteady fixation and delayed maturation of fixation in two babies with AMC.

## METHOD

A detailed clinical examination including complete ophthalmic assessment, orthopaedic, paediatric, orthoptic, otorhinolaryngologist (ENT), haematologist and neurologist review of two babies with AMC was undertaken.

## CASE REPORTS

Case 1 : She was a full term normal vaginal delivery in hospital setting with a birth weight of 7 lbs and 2 oz. Diagnosis of AMC was made at birth due to the presence of multiple joint contractures and talipes. Contractures were worse in lower limbs and was managed on splintage and physiotherapy. She was born with congenital pneumonia. Systemic abnormalities in this child included sickle cell anemia, laryngomalacia, severe gastroesophageal reflux and cleft palate. The neonate required ventilation in hospital in the initial period and phased therapy to combat various

anomalies. At the age of 6 months she was diagnosed to have otitis media with effusion impairing her hearing. Subsequent to grommet insertion her hearing improved in both ears. At 10 months of age she had an episode of bronchiolitis which was successfully treated.

The patient was first seen by us at the age of 3 months because the child did not appear to fix the objects and follow them well. Pupils were equal on both sides and were equally reacting. She had faint cortical cataract in both eyes which were not obscuring central vision. Fundus was normal on both sides. Refraction under atropine revealed a myopia of about 6 diopters in both eyes for which she was prescribed spectacles. She was next seen at age of 6 months. Unsteady fixation

persisted. There was moderate right divergent squint, left hypertropia, and intermittent fine nystagmus right eye. The horizontal extraocular movements appeared unrestricted but vertical movements were restricted especially elevation (Fig 1).

#### **Figure 1**

Figure 1: Case 1 showing right exotropia, right ptosis and decreased vertical eye movements.



At the age of 9 months there was some improvement in her fixation pattern. Fixation to light was unsteady. There was no change in her divergent squint, extraocular motility, nystagmus and cataracts. She was started on left eye occlusion half hour daily. At 14 months she could fix light but only poorly. There was no change in appearance of the squint, nystagmus and cataract. At 15 months she was fixing and following well. Her extraocular movements appeared full but slow. She was noted to have RE mild ptosis of 2mm. Her interpupillary distance was 65 mm and the inter medial canthal distance was 28 mm. Cataracts had not progressed. She was advised to continue myopic glasses and patching left eye.

Head ultrasound was done and subsequently MRI of brain was done, and both were reported to be normal. EMG showed no evidence of neuropathy, no musculoskeletal junction abnormality or anterior horn cell disease. Muscle

biopsy was reported to show non specific changes.

Case 2 : The second child was a full term birth following a caesarean section for extended breech. His limb deformities affected all four limbs and were diagnosed as arthrogryposis multiple congenita at birth. He was started on splintage. His G6 PD deficiency imposed some dietary restrictions in addition to having feeding difficulties due to gastroesophageal reflux. He underwent Nissen's fundoplication for the same. He had pooling of saliva which improved on glycopyrrolate 250mg three times a day. He was in the baby care unit for first five months of his life and was treated for recurrent right upper lobe pneumonia. At the age of 5 months he was diagnosed to have distal denervation of limbs.

He was brought to us at age of 5 months. He appeared to follow the target gradually and the fixation was unsteady. There was intermittent fine vertical nystagmus in both eyes. Ocular motility and alignment appeared normal. His pupils were equal in size and reaction to light. The patient was emmetropic with normal fundus and anterior segment appearance.

He was next seen at the age of 12 months. He was uncooperative for a formal visual acuity testing. His fixation had improved but was still unsteady and he appeared to follow targets slowly. His nystagmus was unchanged. No other abnormality was detected in his eyes. Systemic features in this child included AMC diagnosed at birth, gastroesophageal reflux, G6 PD deficiency, recurrent right upper lobe pneumonia and distal denervation in the limbs.

#### **DISCUSSION**

Although a variety of ophthalmological findings have been described in association with arthrogryposis multiplex congenita, the ocular manifestation of unsteady fixation and delayed maturation of fixation has not described.

The eye findings reported with AMC have been summarised in table 1.

**Figure 2**

Table 1: Eye findings previously reported with Arthrogryposis Multiplex Congenita.

No	Ophthalmological Abnormalities	Reported cases
1	Moebius syndrome(7-14)	14
2	Hypertelorism(14,15)	11
3	Microphthalmia(14,16)	9
4	Congenital glaucoma(14,17)	3
5	Congenital cataracts (14,17)	2
6	Lateral rectus palsy(10,18)	2
7	Duane's syndrome(19)	1
8	Lid abnormalities(20)	1
9	Optic atrophy(21)	1

Paez et al (18) has described a case of an infant with AMC having progressive bilateral paralysis of lateral recti along with decreased corneal blinking reflex. Our first patient was diagnosed to have right divergent squint with normal horizontal extraocular muscle movement. In addition to right divergent squint the other ocular features of restricted elevation, left hypertropia, myopia both eyes and intermittent fine nystagmus have not been reported in association with AMC earlier.

In the second case, there was delayed maturation of visual function and an unsteady fixation pattern till the age of 12 months in the absence of any detectable anterior segment, posterior segment or refractive abnormality.

Various systemic associations like cleft palate, low set ears, GI abnormalities, congenital heart defects, hypoplastic lungs and cryptorchidism have been described with this syndrome. Presence of certain systemic features in both these babies which have not been previously described with AMC include sickle cell anaemia, hearing loss and G6PD deficiency.

Joint abnormalities were managed by splintage and physiotherapy. Tracheostomy was performed for laryngomalacia and a hearing device was fitted for hearing loss. G6PD deficiency required to impose some dietary restrictions and avoidance of use of certain drugs.

AMC is a syndrome of congenital joint contractures with other associated abnormalities. These cases presented some never before described ocular and systemic abnormalities in association with AMC. We observed a delayed visual

maturation and fixation development in both our patients. The presence of sickle cell anemia in the first case and G6 PD deficiency in the second case have not been previously described. We suggest a close multidisciplinary follow up

on all cases of arthrogryposis multiplex congenita to detect, report and better understand these associations.

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