Arthrogryposis Multiplex Congenita: dental findings and treatment of an 8-year-old child

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Abstract

This report describes a case of Arthrogryposis Multiplex Congenita (AMC) with limited mouth opening and dental caries. Conservative dental treatment and physiotherapy exercises were prescribed. The aim of this case report is to describe the method and difficulties in the dental care of this patient and outline the importance of a preventive programme.

Key words: Arthrogryposis Multiplex Congenita, oral health, dental treatment

Introduction

Arthrogryposis Multiplex Congenita (AMC) is defined as multiple congenital malformations, which are due to neurological and muscular deficits at the time of birth and restrict the movement of the joints. The syndrome was first reported by Otto in 1841 as congenital myodystrophy (Otto, 1847). AMC is a disorder that is reported to occur in 1 in every 3,000-10,000 live births (Hall 1985, Steinberg et al., 1996; Thomas et al., 2001). The variation in the reported incidence is most probably related to different definitions of the disorder. Hall (1985) described arthrogryposis as a one of more than 150 conditions known to have congenital contractures on distal joints more frequently than proximal joints. In the study, the following three categories were used to distinguish the different types of arthrogryposis: those associated with limbs only, those with limb and other organ involvement (without central nervous system [CNS]), and syndromes with CNS involvement.

The cause of AMC varies, and may include neurogenic factors, myopathy, lack of foetal movement, and intrauterine vascular accident. (Steinberg et al., 1996). A number of studies have reported on changes in the spinal cord associated with AMC, such as a decrease in number and size of the anterior horn cells. In addition, a decrease in the size and number of muscle fibres, as well as fatty degeneration, have been reported by a number of other investigators. (Cohen and Isaacs, 1976). Hageman and Willemse (1983) suggested that animal studies support the theory that decreased foetal movement in the uterus can lead to congenital joint contractures. The timing and length of immobilisation may determine which joints are involved and the degree to which their function is limited. Williams (1978) suggested that a maternal viral infection may be part of the cause of AMC. Most reports show that AMC has a male predominance (Cohen and Isaacs, 1976). Inheritance is particularly relevant to so-called distal arthrogryposis which merits separate discussion. Arthrogryposis can be inherited in a number of ways: autosomal dominant, autosomal recessive, X-linked recessive and by mitochondrial inheritance (Gordon, 1998).

In some cases, only a few joints may be affected, and the range of motion may be nearly normal. In more severe cases, nearly every joint may be involved, including the jaws and spine (Alves et al., 2007). The disease is characterised by punctuate endochondral calcification of the apophysis or the epiphysis. The lesions appear in the elbow, wrist and malleolus joints which results in short limbs. Other characteristics of the disease are: ulnar ectopic continuous flexion of the joints, scoliosis, ichthyosis, strabismus and abnormal convergence of the parietal raphe. Usually, the patient is not intellectually impaired, but the disease is often associated with other congenital abnormalities.

Diagnosis is based on medical history and physical examination combined with laboratory tests, such as muscle biopsies, blood tests, chromosomal analysis, measurements of enzyme levels and radiographs. AMC can be diagnosed during pregnancy by using ultrasound at approximately 20 weeks of gestation (De Andrade et al., 2000). Sometimes, an electrical nerve or muscle conduction study is necessary. Computed Tomography (CT) scan or Magnetic Resonance Imaging (MRI) can identify any central nervous system abnormalities or myopathic forms and may provide important information (Alves et al., 2007).

In the area of the head, a review of the literature describes micrognathia, facial asymmetry, limited mobility in temporomandibular joints (TMJ), periodontal disease, cleft palate, high-arched palate, open-bite, facial muscle weakness, and delayed tooth exfoliation (Cohen and Isaacs, 1976, Mielnik-Blaszczyk and Borowska, 2002, Alves et al., 2007).
Dental treatment of AMC patients can be very complicated so it is better to follow an intensive oral preventive programme. (Mielnik-Blaszczak and Borowska, 2002). Orthodontic treatment is difficult because of limited mandibular opening and limited lateral movements (Alves et al., 2007).

Treatment of arthrogryposis should be undertaken by a multidisciplinary team, consisting of paediatrician, surgeon, physical therapist, psychologist, paediatric dentist and orthodontist. The main aim of treatment is to improve function. Through physiotherapy and other treatments, substantial improvement is normally possible by stretching out the contracted joints, developing the weak muscles, and increasing the range of motion. Parents are encouraged to become active participants by continuing their child’s therapy at home on a daily basis. Orthopaedic surgery may also relieve or correct joint problems. Treatment of TMJ problems through bilateral coronoidotomy surgery, right and left meniscectomy, capsular release, and lateral pterygoid myotomies are elective choices. It is necessary to develop muscle strength with early stimulation of movement, and periods of immobilisation should be minimised. Despite the recommendation of physical therapy to improve function, the literature does not relate the importance of orthodontic evaluation and monitoring from early childhood for development of problems (Alves et al., 2007; Ruff et al., 1988). The lifespan of an individual with arthrogryposis is usually long, but it may be altered by heart defects or central nervous system problems. Fortunately, AMC is not a progressive disorder (Alves et al., 2007).

This report presents a case of a patient with AMC and severe dental problems. It describes the difficulties associated with the dental treatment due to the restricted mouth opening as well as the importance of a preventive programme.

Case report

An 8-year-old boy was referred to the postgraduate clinic of the Paediatric Dentistry Department, Dental School, University of Athens from Athens Children’s Hospital Agia Sofia for oral assessment. The patient was diagnosed with Arthrogryposis Multiple Congenita at the time of birth at Athens Children’s Hospital Agia Sofia. The parents had declined genetic counselling and a search on www.possum-web.com.au after listing all the traits found on the patient, revealed two possible syndromes, distal arthrogryposis type 4 or Sonuda Kudo. The traits listed were: intellectual impairment, absence of language, strabismus in the right eye, camptodactyly, equinovarus, scoliosis, navicular thorax, short arms and legs, club foot, asthma and previous operations for cryptorchism and cheilioschisis (Figures 1 and 2).

Intraoral examination revealed the patient was in the mixed dentition phase with multiple carious lesions (dmft:10). Orthodontic examination revealed a Class II, Div II malocclusion with severely crowded mandibular anterior teeth, limited mouth opening (22 mm), anterior horizontal protrusion (10 mm), limited lateral mandibular movements and poor oral hygiene (Figures 3-5). The patient had received no previous dental care. Dietary analysis showed that the patient’s diet included frequent consumption of sweets and sugary drinks between meals.

In order to best treat the patient he was assessed by a multidisciplinary team consisting of a paediatric dentist, an orthodontist, and an oral surgeon. His paediatrician, general surgeon and physiotherapist were consulted as well. During the first visit, the patient was very attached to his mother, stressed and frightened which, along with his intellectual impairment, limited understanding and the lack of speech led to uncooperative behaviour. However, he did respond to behaviour modification techniques during dental treatment such as distraction, tell – show – do and reinforcement of positive behaviour.

It was very difficult for the patient to stand still for lateral cephalometric and panoramic radiographs. Nevertheless, impressions were taken and casts were made for orthodontic evaluation. (Figure 6). Four periapical radiographs were taken. The radiographic examination showed the extent of the caries and the existing furcation lesions (Figure 7).

The dental treatment plan consisted of:

• The use of a jaw motion rehabilitation system (Therabite®) three times a day under the supervision of his mother before and during the dental treatment, in order to improve the opening of the mouth (Figure 8).
• Pulp therapy followed by placement of stainless steel crowns or resin restorations on affected primary molars.
• Extraction of necrotic primary teeth.
• Composite resin restorations or sealants placed on the first permanent molars.
• Prophylaxis and fluoride varnish application (Figures 9-11).

Orthodontic evaluation was undertaken by an orthodontist. It was suggested that the patient should wait at least one year before commencing orthodontic treatment. After eight weeks using the Therabite® appliance, mouth opening improved by 12 mm, from 22 to 34 mm.

The home preventive programme included instructions to the parents for oral hygiene (brushing twice daily with a 1450 ppm fluoride toothpaste) night flossing and dietary counselling. The first recall was at three months for clinical evaluation, prophylaxis and fluoride varnish application. At the six month recall all the above as well as radiographic evaluation were undertaken (Figure 12). No new carious lesions were observed. Mouth opening was 31 mm. A marked improvement in attitude towards the dentist, to oral hygiene and dietary habits was observed. Brushing instructions to the parents were reinforced once more. It was suggested to continue the use of the mouth opening appliance. At the twelve month clinical and radiographic evaluation, findings were normal. Radiographs showed normal root resorption of primary teeth.

Discussion
Figure 1 Postural position of the patient

Figure 2 Characteristic position of the hands with capto-dactyly

Figure 3 Anterior view before treatment. Poor oral hygiene can be observed

Figure 4 Maxilla view- multiple dental caries

Figure 5 Mandibular arch with multiple dental caries and crowded anterior teeth

Figure 6 Study models. Anterior horizontal protrusion and small mandibular jaw. Class II molar relationship: Overjet 10mm, Overbite 4mm
Figure 7 Periapical radiographs before dental treatment

Figure 8 Use of the Therabite® appliance

Figure 9 Anterior view after dental treatment. Improved oral hygiene can be observed

Figure 10 Maxillary arch after dental treatment

Figure 11 Mandibular arch after dental treatment

Figure 12 Periapical radiographs after dental treatment
Discussion

AMC is a rare disorder that causes multiple joint contractures and generalised hypoplasia of skeletal muscles. This disease usually coexists with other congenital anomalies and syndromes.

Manifestations of AMC can involve the maxillofacial region. In this case, the patient had abnormalities that involved the cranio-maxillofacial area. Similar manifestations were present in the study of Cohen and Isaacs (1976) as well as in the study of Steinberg (1996).

Several papers observe that patients with AMC have restricted mandibular opening as well as weakness of the masticatory musculature and high-arched palate (Steinberg et al., 1996). This patient had limited mouth opening, facial muscle weakness and a high palatal arch as well.

Treatment of the maxillofacial manifestations of AMC depends on the severity of the case and is problematic. Management of the TMJ and the limited mandibular opening is equally challenging. The use of mouth opening appliances can improve the opening of the mouth and simplify the dental treatment (De Andrade et al., 2000; Thomas et al., 2001). De Andrade et al. (2000) described a case of AMC and concomitant bruxism with limited mouth opening and pain in the temporomandibular joint. At first, they prescribed the use of a muscle relaxing splint in order to relieve the signs and symptoms of bruxism, as well as to reduce pain. Following splint therapy, physiotherapy exercises were employed to complement the splint treatment. After two months of physiotherapy, an improvement on mouth opening was noticed.

In our case, the use of a muscle relaxing splint was not useful, since the patient felt no pain. However, in order to improve the opening of his mouth, we used a jaw rehabilitation appliance (TheraBite®), which helped in the strengthening of the muscles. The same appliance was used by Thomas et al. (2001) before and after surgery. In our case, surgery was not necessary and the mouth opening improved by 12mm in eight weeks. The use of the appliance was recommended regularly, just like physical exercise. Every time it ceased for long periods of time, for example, during surgery to correct limp malformations, mouth opening relapsed although not to the original value.

Patients with AMC usually have generalised gingivitis and dental carries (Mielnik-Blaszczak and Borowska, 2002; Alves et al., 2007). Our patient received restorative and preventive dental treatment. Mielnik-Blaszczak and Borowska (2002) reported a patient with AMC who had delayed teething. Our patient had a normal dentition.

All writers stress the importance of a preventive programme customised to the needs of the patient. Our patient was placed on a three-month follow up programme. The cooperation of the family members was paramount to the success of the treatment and outcome maintenance.

Conclusion

A multidisciplinary approach is recommended to provide optimal functional and improved aesthetics in cases with AMC, due to the complexity of the facial and associated problems. In the case reported here, use of the TheraBite® appliance improved the opening of the mouth, so dental treatment was more easily performed. The continuance of the exercises with the TheraBite® device was suggested to maintain the opening of the mouth.

Restriction of joint movement also limits oral hygiene. It is very important that a strict preventive programme is followed and reviewed every three months, to include dental examination, application of fluoride varnish and reinforcement of oral hygiene instructions.

References


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