Dominant Form of Arthrogryposis Multiplex Congenita with Limited Mouth Opening: A Clinical and Imaging Study

Antonio Sergio Guimarães, DDS, MS, PhD
Professor
Department of Oral Physiology
Head Institute
São Paulo Federal University (UNIFESP)
São Paulo, SP, Brazil

Suely Kazue Nagahashi Marie, MD, PhD
Chair
Laboratory of Neuromuscular Disorders
Department of Neurology
School of Medicine
University of São Paulo
São Paulo, SP, Brazil

Correspondence to:
Dr Sergio Guimarães
Rua Visconde da Luz # 60/31
Cep 04537 070 São Paulo, SP, Brasil
Fax: +55 11 3842 9262
E-mail: asgatm@uol.com.br

Aims: *Arthrogryposis multiplex congenita (AMC) is characterized by congenital contractures and joint deformities, but there are only a few reports of temporomandibular joint (TMJ) involvement. The objective of this investigation was to study the cause of limited mouth movement in this disease.* ***Materials and Methods:*** *Four individuals from a family affected by AMC over 5 generations were examined clinically and by magnetic resonance imaging (MRI) and 3-dimensional computerized tomography (3D-CT).* ***Results:*** *The CT scans of the 4 individuals showed hyperplasia of the coronoid process protruding into the infratemporal fossa in 2 of them and cranially to the zygomatic arch in the other 2; the hyperplasia was associated with mechanical limitation of the mouth opening. The MRI showed a disc displacement with reduction in 1 patient and a disc displacement without reduction in another; disc displacement could not be evaluated because of the limited mouth opening in the other 2. The condyle-disc complex of these last 2 individuals could only rotate. The MRI on T2-weighted images showed disc hyposignal in all cases but no alterations in the masticatory muscle tissue. The pedigree of the family suggests an autosomal dominant form of inheritance.* ***Conclusions:*** *The restriction of mouth opening in the 4 individuals affected by AMC was likely due to osseous dysplasia.* J OROFAC PAIN 2005;19:82–88

Key words: arthrogryposis, coronoid process, jaw locking, magnetic resonance imaging, mouth opening, 3-dimensional computed tomography

Arthrogryposis multiplex congenita (AMC) is an uncommon congenital disorder, characterized by multiple fixed-joint deformities, that occurs in 1 out of every 5,000 to 10,000 births.[1] It is defined as a congenital, nonprogressive limitation of movement in 2 or more joints in different body areas.[1] There are both neuropathic and myopathic types of AMC.[1] Maxillofacial manifestations of AMC include micrognathia, limited jaw opening, high vaulted palate, cleft palate, and weakness of the masticatory muscles.[2–4] It has been suggested that joint immobility occurs as a consequence of a lesion in the neuromuscular system (anterior horn cells, roots, peripheral nerve, or motor end plate). Limitation of movement during intrauterine development or connective tissue disorder, with disturbances in the proper development of cartilage, tendon, or bone, have been suggested as other causes for this condition.[1,5] The genetic form of AMC has also recently been reported.[6,7]

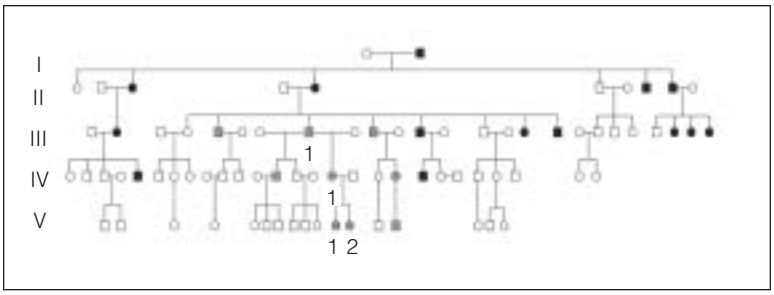

Fig 1 Pedigree of the family. Black box = affected male; black circle = affected female; gray box = affected male with limited mouth opening; gray circle = affected female with limited mouth opening; white box = unaffected male; white circle = unaffected female. I–V correspond to the generations. 1 and 2 correspond to the members studied in each generation.

Very few cases of AMC with limited opening and range of jaw movements have been described where this restriction has been attributed to involvement of the temporomandibular joint (TMJ) and the masticatory muscles.[4,8–11] In this report, 4 individuals from 3 generations from a family afflicted with AMC for 5 generations participated in an imaging study of the TMJs, infratemporal fossa, and coronoid process in order to clarify the cause of their jaw movement restrictions.

Materials and Methods

Subjects

The subjects were members of a family affected by AMC over 5 generations. The family pedigree is presented in Fig 1. There was 1 affected individual in the first generation, 4 affected individuals from 6 siblings in the second generation, 10 affected from 16 offspring in the third generation, 5 out of 21 in the fourth generation, and 3 out of 17 in the fifth generation. Thus, 23 individuals (12 males and 11 females) from 5 generations of this family were affected by AMC, thereby revealing a dominant form of inheritance. Females and males were equally affected over the second and the third generations, and 3 males and 2 females were affected in the fourth generation, and 2 females and 1 male were affected in the fifth generation; 9 out of 23 affected subjects had limited mouth opening. No information was available concerning the degree of mouth opening from the other 14, becuase the could not be found to be evaluated. Limitation in mouth opening, which had been perceived since birth, was the main complaint of these 9 individuals.

Two affected girls (V-1, V-2) from the fifth generation, along with their mother (IV-1) and grandfather (III-1) were examined clinically, and the TMJ/coronoid process area was imaged by computerized tomography (CT) and magnetic resonance imaging (MRI) scans. Subjects' rights were protected by an appropriate institutional review board, and informed consent was granted by the subjects.

Clinical Examination

All 4 subjects were examined for muscular contractures in the upper and lower limbs as well as spinal deformities, limited range of jaw movements (maximum opening, protrusive and lateral movements), number of teeth, periodontal conditions, and articular sounds.

Imaging

Two types of imaging were used:

- *CT (Elscint hellicoidal):* Corrected sagittal multiplanar reconstruction images on sagittal and coronal planes were analyzed. Three-dimensional (3D) images were used to study the spatial relationship of the structures involved, including the TMJs, infratemporal fossas, zygomatic arches, body and ascending ramus of mandibles, and coronoid processes.
- *MRI (GE 1.5T):* Corrected sagittal and coronal images were created in closed- and opened-mouth positions with the spin echo technique on T1-weighted images (repetition time [RT] = 500 ms, echo time [ET] = 10 ms, field of view [FOV] = 140 mm) and T2-weighted images (RT = 200 ms, ET = 100 ms, FOV = 140 mm), including full lateromedial width with a slice thickness of 4 mm, with no gap between slices. The articular disc position was evaluated according to a technique previously described in the literature.[12]

Masticatory muscles including the masseter, temporalis, and lateral and medial pterygoids were analyzed by means of T2-weighted images for fatty replacement of muscular tissue. The presence of fatty degeneration was diagnosed if a hypersignal was visible within the investigated muscle.

Table 1 Findings at Clinical Examination

Clinical examination	III-1	IV-1	V-1	V-2
Sex	M	F	F	F
Age at examination	84	39	13	11
Maximum jaw opening (mm)	30	24	2	10
Protrusion (mm)	NP	5	2	2
Laterotrusion R/L (mm)	NP	5/5	2/2	1/2
Angle class	I	II	II	II
No. of teeth	26	28	28	23 mix
Periodontal condition	Regular	Good	Good	Good
Chewing side	Bilateral	Right	Bilateral	Right
Pain on chewing	No	No	No	No
Articular sounds	No	DDwR	No	No
Contractures				
Hips	–	–	+	+
Knee	–	–	+	–
Ankle	–	+	+	+
Feet, toes	–	–	+	+
Elbow	–	–	–	–
Wrist	+	+	+	+
Hand, fingers	+	+	+	+
Spine	–	Hyperlordosis	Hyperlordosis	Hyperlordosis

III-1 = grandfather; IV-1 = mother; V-1 and V-2 = daughters; M = Male; F = Female; DDwR = disc displacement without reduction; + = present; – = absent; NP = not possible.

Results

Clinical Examination Findings

The 84-year-old grandfather (III-1) presented with 26 teeth, 30 mm of maximum jaw opening, and Angle's Class I malocclusion. The 39-year-old mother (IV-1) presented with 28 teeth, 24 mm of maximum jaw opening, and Class II malocclusion. Her elder daughter, who was 13 years old, had 28 teeth and 2 mm of maximum jaw opening, and her younger daughter, who was 11 years old, had a mixed dentition of 23 teeth and 10 mm mandibular opening; both daughters presented with Class II malocclusion.

Additional detailed findings of the stomatognathic examination and distribution of the contractures are presented in Table 1.

Imaging Findings

The 3-D CT scans revealed a striking increase in the length of the coronoid process in all 4 subjects. The coronoid process protruded into the infratemporal fossa in the grandfather and mother and cranially to the zygomatic arch in the 2 daughters (Fig 2). The images obtained in the opened-mouth position clearly demonstrated that the coronoid process impinged on the anterior part of the zygomatic process or the lateral aspect of the infratemporal fossa and mechanically blocked mouth-opening movements. The least degree of coronoid process increase was seen in the mother and grandfather, which was consistent with their wider jaw openings of 24 mm and 30 mm, respectively. The CT scans of all 4 subjects showed a normal TMJ morphology with normal articular surfaces (Fig 2).

The MRI of the grandfather showed normal condyle-disc-fossa relationships on the right side (Fig 3) and anterior disc displacement without reduction on the left side (Fig 3). In the case of the mother, anterior disc displacement with reduction on both sides was observed (Fig 3). The MRI scans of her 2 daughters showed only a discrete condylar rotation without translation. In these cases, it was not possible to evaluate the form and position of the disc since the MRI hyposignal on T1-weighted images of the condyle, disc, and fossa structures did not allow a determination of the disc position. Furthermore, the very limited mouth opening did not allow evaluation of disc displacement. The MRI hyposignal on the T1- and T2-weighted images was observed in the discs of all 4 subjects (Fig 3), and no volumetric or signal alterations were detected in the masticatory muscles on qualitative analysis (Fig 4).

Discussion

The limitation of jaw function that occurs in AMC may directly and significantly affect mastication, oral hygiene, and dental care. The maxillofacial manifestation of AMC has been considered a

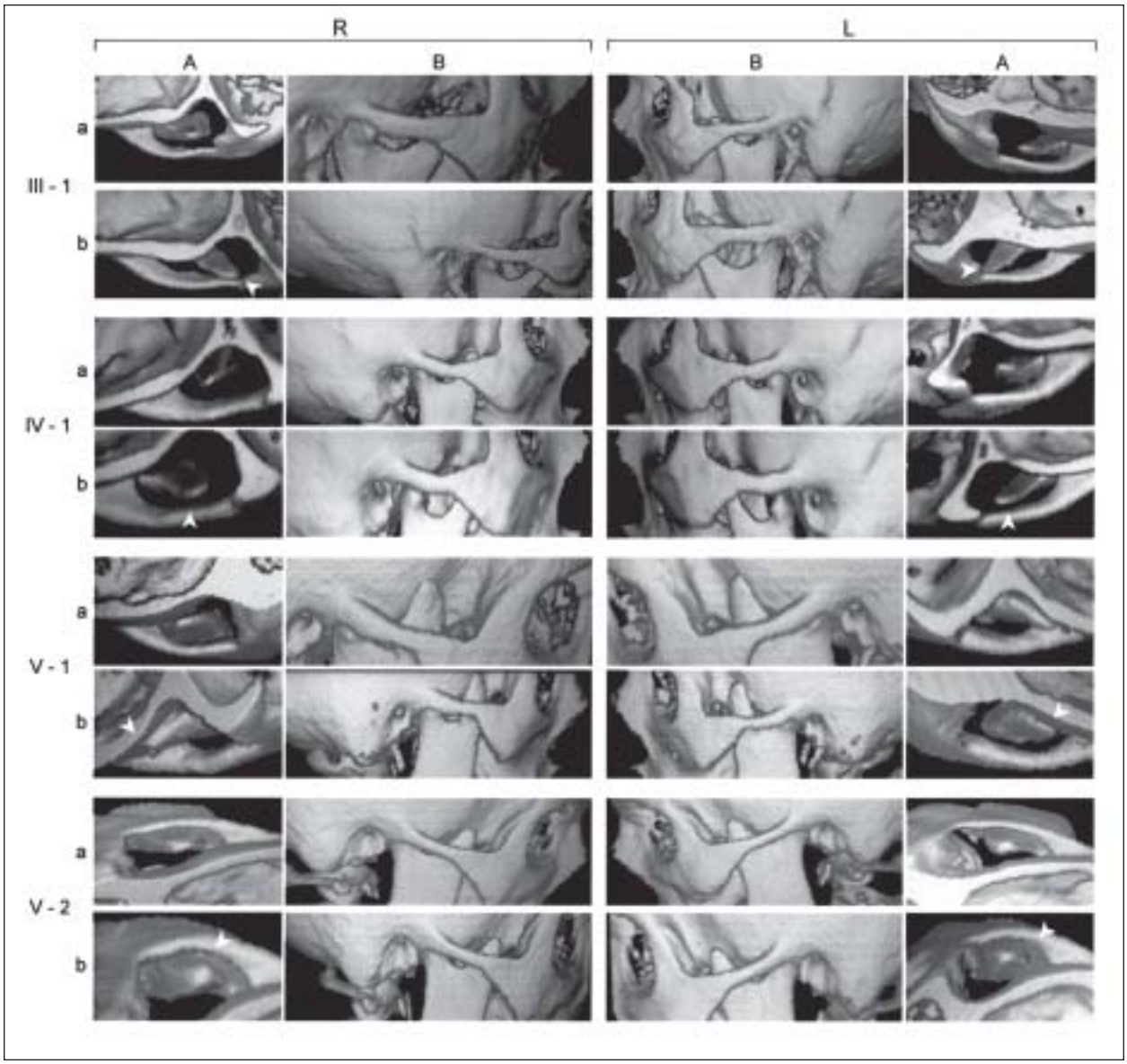

Fig 2 CT scans reconstructed from 1-mm-thick axial images. A = axial 3-D, upper view at the level of the zygomatic arch, at the best angle of reconstruction to demonstrate the mechanical limitation of the coronoid process during mouth-opening movements in the 4 subjects, III-1, IV-1, V-1, and V-2. B = sagittal 3-D, lateral view. Arrowheads indicate the most probable areas of mechanical limitation. R = right, L = left; a = maximal intercuspation position; b = wide mouth-opening position.

congenital abnormality or a secondary positional abnormality.[2–4,13] The joint hypomobility has been previously explained as being the result of muscle deficiency and tension arising from hypoplasia or atrophy.[5] In contrast to these hypotheses, the present findings have demonstrated no morphological abnormalities in the TMJs and masticatory muscles but instead an elongated coronoid process that impinged against the zygomatic arch or the lateral aspect of the infratemporal fossa and blocked the jaw-opening movements. The demonstration of an increased coronoid process in the 4 cases studied reinforces the hypothesis of an osseous contribution to the restriction of mouth opening. The pathological factors leading to this abnormality remain an open question. The patients presented no morphological alteration at the insertion site of the temporalis muscles into the coronoid process. However, further functional study of this muscle may clarify whether a factor related to tension developed by the temporalis muscle over the coronoid process might be respon-

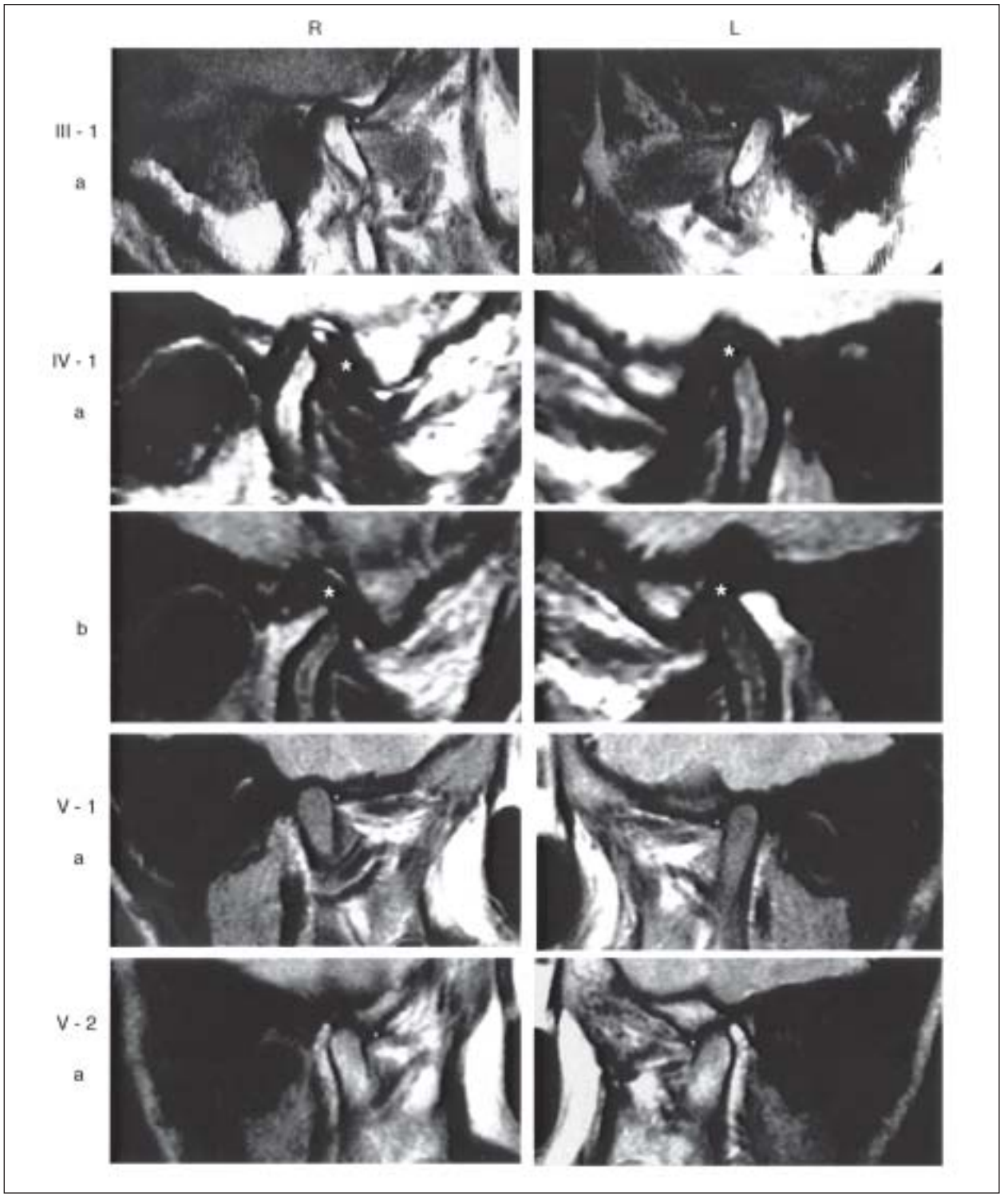

Fig 3 Corrected sagittal MRI of TMJ on spin-echo T1-weighted images (RT = 500 ms, ET = 10 ms, FOV = 140 mm, slice thickness = 4 mm). Normal condyle-disc-fossa relationships are demonstrated in III-1 aR. V-1 and V-2 aR and aL show discrete translatory movements of the condyles but no rotational movements. The disc position could not be evaluated because of the hyposignal demonstrated in condyle-disc-fossa structures. Anterior disc displacement is shown in III-1 aL and IV-1a; a reduction is demonstrated in in IV-1b. R = right, L = left; a = maximal intercuspation position; b = wide mouth-opening position. *Hyposignal in the disc.

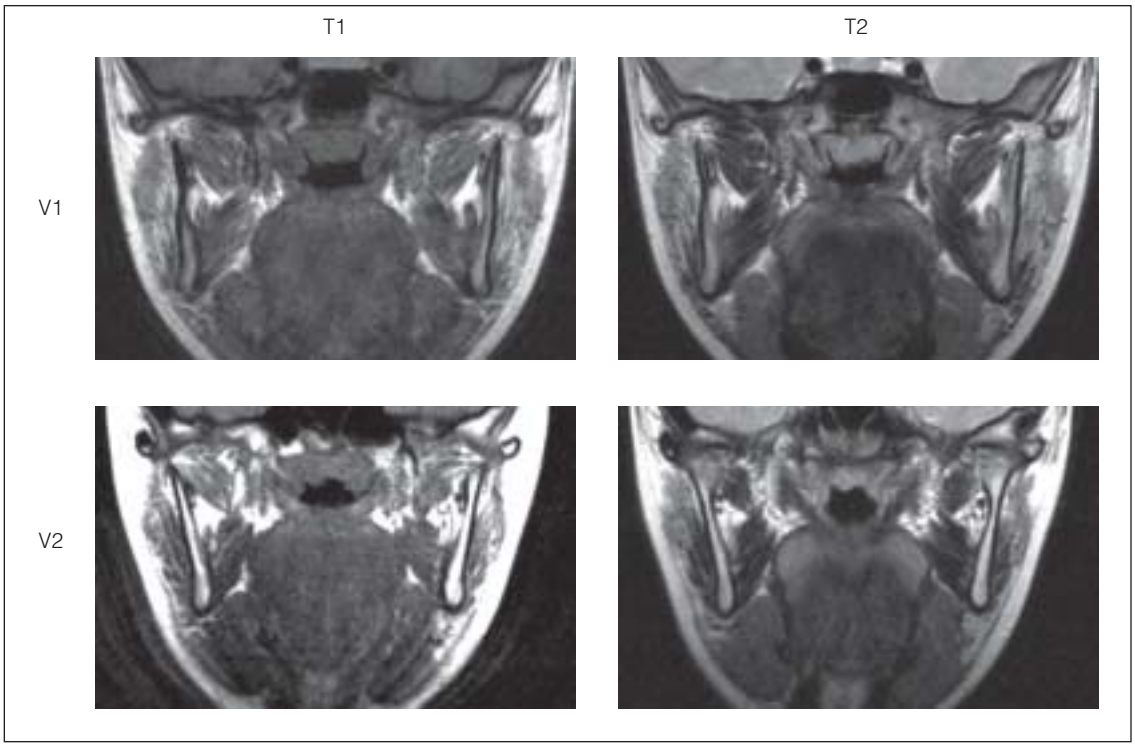

Fig 4 Coronal spin-echo T1-weighted (RT = 500 ms, ET = 10 ms, FOV = 140 mm, 4-mm slice thickness) and T2-weighted (RT = 200 ms, ET = 100 ms, FOV = 140 mm, 4-mm slice thickness) MRIs of the 2 daughters (V-1 and V-2) show medial and lateral pterygoid muscles with usual muscle MRI signals and no morphological alterations.

sible for its modification in shape and size. Two previous studies of AMC with TMJ involvement also reported alterations in the osseous architecture. Heffez et al[4] described a 5-year-old boy with normal chronologic and morphologic dental development and with a mouth opening limited to 25 mm. The subject presented with slight micrognathia, normal antegonial notching, and an absence of bilateral condylar translation that was confirmed by facial radiographs and TMJ tomograms. The authors observed slender and pointed coronoid processes, rudimentary condyles, and smooth condylar surfaces. In the other study, Hodgson et al[11] reported the case of a 30-year-old man who had had arthrogryposis of the extremities since birth and who had developed TMJ deformity after a trauma to the mandible. Interestingly, they observed a unilaterally increased size and gross deformity of the condylar head. The TMJ dysfunction in this case was attributed to a proliferative bone alteration of the condyle.

The existence of an anterior disc displacement with and without reduction in 2 of the cases in the present study (III-1, IV-1) probably is not associated with the limitation of jaw movement, as disc displacement could be considered a functionally adapted state observed in normal subjects.[14] Moreover, a unilateral anterior disc displacement without reduction causing a dysfunctional state usually leads to asymmetric mouth opening, which was not observed in those cases.

Limited opening of the jaw has been described in 2 other conditions: type IIE distal arthrogryposis and trismus pseudocamptodactyly syndrome (TPS; also known as Hecht syndrome). In the first pathological condition, an unusual hand position has been consistently observed together with limited jaw opening.[14] In the second, complete finger extension on wrist flexion and fingers flexion contractures on wrist dorsiflexion due to short finger-flexor tendons has been reported.[15,16] These 2 conditions can be ruled out in the present 4 cases since no such alterations were observed. Further definition of these syndromes may be possible in the future if molecular markers are found.

Although the masticatory muscles showed the usual signal on MRI analysis, a neuromuscular problem, such as contracture of the capsule or muscles (eg, due to proliferation of connective

tissue within the muscle), could be a further cause of jaw locking. Histologic and histochemical studies of these muscles might clarify this issue.

In conclusion, the present study has revealed a dominant form of inheritance of AMC wherein limited jaw movement is a phenotypic characteristic in at least 40% of the affected subjects in the present family. As no relevant TMJ or masticatory muscle morphological alterations were detected on imaging analysis in the 4 family members studied, it is likely that the jaw-opening restriction was of mechanical origin, ie, due to the increased length of the coronoid process.

Acknowledgments

The authors are very grateful to CTC and INCOR Imaging Services for kindly allowing them to do the imaging examinations for the patients. Supported by an FAPESP grant (process no. 01/12844-1).

References

1. Hall JG. An approach to congenital contractures (arthrogryposis). Pediatr Ann 1981;10:15–26.
2. Garfunkel A, Steiner JE. Orofacial manifestations in arthrogryposis: A case report. J Oral Med 1971;26:77–81.
3. Cohen SR, Isaacs H Jr. Otolaryngological manifestations of arthrogryposis multiplex congenita. Ann Otol Rhinol Laryngol 1976;85(4 Pt 1):484–490.
4. Heffez L, Doku HC, O'Donnell JP. Arthrogryposis multiplex complex involving the temporomandibular joint. J Oral Maxillofac Surg 1985;43:539–542.
5. Hageman G, Willemse J. Arthrogryposis multiplex congenita. Review with comment. Neuropediatrics 1983;14: 6–11.
6. Bamshad M, Watkins WS, Zenger RK, et al. A gene for distal arthrogryposis type I maps to the pericentromeric region of chromosome 9. Am J Hum Genet 1994;55: 1153–1158.
7. Krakowiak P, O'Quinn JR, Bohnsack JF, et al. A variant of Freeman-Sheldon syndrome maps to 11 p15.5-pter. Am J Hum Genet 1997;60:426–432.
8. de Andrade CM, Hotta TH, Mazetto MO, de Felicio CM, Bataglion A. Arthrogryposis multiplex congenita in a patient with limited mouth opening: A case report. Cranio 2000;18:66–70.
9. Steinberg B, Nelson VS, Feinberg SE, Calhoun C. Incidence of maxillofacial involvement in arthrogryposis multiplex congenita. J Oral Maxillofac Surg 1996;54: 956–959.
10. Thomas JA, Chiu-Yeh M, Moriconi ES. Maxillofacial implications and surgical treatment of arthrogryposis multiplex congenita. Compend Contin Educ Dent 2001;22: 588–592.
11. Hodgson P, Weinberg S, Consky C. Arthrogryposis multiplex congenita of the temporomandibular joint. Oral Surg Oral Med Oral Pathol 1988;65:289–291.
12. Tasaki MM, Westesson PL. Temporomandibular joint: diagnostic accuracy with sagittal and coronal MR imaging. Radiology 1993;186:723–729.
13. Smith EM, Bender LF, Stover CN. Lower motor neuron deficit in arthrogryposis. An EMG study. Arch Neurol 1963;8:97–100.
14. Staheli LT, Hall JG, Jaffe KM, Paholke DO. Arthrogryposis: A text atlas. Cambridge, UK: Cambridge University Press, 1998.
15. Robertson RD, Spence MA, Sparkes RS, Neiswanger K, Field LL. Linkage analysis with the trismus-pseudocamptodactyly syndrome. Am J Med Genet 1982;12:115–120.
16. Tsukahara M, Shinozaki F, Kajii T. Trismus-pseudocamptodactyly syndrome in a Japanese family. Clin Genet 1985;28:247–250.