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A Revised and Extended Classification of the Distal Arthrogryposes

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Since the group of disorders known as the distal arthrogryposes (DAs) were defined, additional disorders characterized by multiple congenital contractures of the distal limbs were described, and the distribution of phenotypic findings in the DAs has been expanded. The breadth of disorders labeled as DAs has diminished the usefulness of the DA classification. We propose a strict definition of DA and diagnostic criteria for DA disorders. Subsequently, we use these standards and propose a revised classification of discrete conditions that should be labeled DAs. Optimally, this serves as a framework for a DA classification based on underlying molecular and physiologic abnormalities.

KEY WORDS: arthrogryposis, congenital contractures, disease classification

INTRODUCTION

In 1982, Hall et al. summarized the data on 44 individuals with congenital distal limb contractures and defined a group of disorders designated the Distal Arthrogryposes (DAs). Hall et al. [1982a] recognized two major groups of patients and proposed that the DAs be divided into two distinct forms. Distal arthrogryposis type I (DAI) was characterized by overlapping fingers at birth, camptodactyly, ulnar deviation, and positional foot deformities and was not associated with any additional distinguishing physical abnormalities. Individuals with patterns of distal contractures similar to those found in DAI in addition to other discrete anomalies were grouped into five subcategories of distal arthrogryposis type II (DAII).

The purpose of defining the DAs, as is the logic behind disease classification in general, was to 1) understand the biologic basis of these disorders and their relationships to each other, if any, and 2) to facilitate clinical diagnosis and management of affected individuals. To this end the DA classification has been moderately successful. Categorization of a patient enables the clinician to provide more accurate counseling to the individual about the natural history and recurrence risks of a DA disorder. Yet, little has been learned about the relationships between disorders except that there is more intra- and inter-familial phenotypic variation and overlap than was originally appreciated, and that DAI exhibits locus heterogeneity. Furthermore, although Hall et al. [1982a] acknowledged that their DA classification was "preliminary," it has yet to be amended with more contemporary information. As part of an ongoing effort in our laboratory to identify genes causing human limb malformations, we became interested in defining further the basis of disorders characterized by congenital distal limb contractures. The purpose of this paper is to propose a revision and extension of the original DA classification utilizing a new and strict definition of DA as well as diagnostic criteria which should more effectively distinguish DA disorders from other conditions with distal limb contractures.

JUSTIFICATION

First, it appears that the label "distal arthrogryposis" is used to delineate individuals and disorders characterized by congenital contractures of the distal limbs regardless of cause. Since 1982 numerous associated abnormalities have been described in individuals with distal limb contractures [Chitayat et al., 1990, 1991; Moore and Weaver, 1989; Reiss and Sheffield, 1986; Stoll et al., 1992]. Additionally, inferences are commonly drawn from these descriptions and subsequently applied broadly across a wide variety of disorders. Furthermore, disorders with distal limb contractures not encompassed in the Hall et al. [1982a] DA classification (i.e., congenital contractural arachnodactyly, CCA; Freeman-Sheldon syndrome, and FSS) have been described in families with DAI [Bamshad, unpublished;

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Klemp and Hall, 1995]. Likewise, after application of the strict FSS criteria proposed by Carey et al. [1993], some individuals diagnosed with FSS were reclassified subsequently with DAI [Bamshad et al., 1994] or autosomal dominant multiple pterygium syndrome (ADMPS) [Carey et al., 1993]. Last, although disorders with manifestations similar to DAI and DAII, such as trismus-pseudocamptodactyly syndrome, were considered by Hall et al. [1982a], it is unclear why these disorders are not included in the original DA classification. Thus, it appears the boundaries of the DA disorders are becoming indistinct if not ambiguous.

Second, no strict criteria exist that define whether a disorder should be considered a form of DA, and no standardized definition of DA is available. This generates confusion as the clinician tries to distinguish between different disorders characterized by distal limb contractures. Does proximal joint involvement but no distal contractures denote a DA disorder? Do neurologic abnormalities exclude the diagnosis of a DA disorder?

Third, notable overlap in the distribution of abnormalities between different forms of DA has been described over the last 13 years [Reiss and Sheffield, 1986; Schrander-Stumpel et al., 1991]. Should an individual with short stature, distal limb contractures, and a normal palate whose sib has a cleft palate be diagnosed with DAI or Gordon syndrome? Fourth, the phenotypic defects of some of the DA disorders have been described further since Hall et al. [1982a] created the DA classification [Lai et al., 1991; Schrander-Stumpel et al., 1993]. Last, it has been suggested that DA disorders with similar phenotypic features exhibit locus heterogeneity [Bamshad et al., 1994].

DEFINING THE DISTAL ARTHROGRYPOSES

Our initial step toward revising the classification was to create strict criteria that defined which disorders could plausibly have a similar etiologic basis. DAI was used as the model disorder. Any disorder characterized by distal limb contractures and a heritable pattern was considered a candidate. DA was defined as an inherited primary limb malformation disorder characterized by congenital contractures of two or more different body areas and without primary neurologic and/or muscle disease that affects limb function. We argue that these disorders are malformations because it is suspected, and in some cases demonstrated (e.g., DAI, FSS, and trismus-pseudocamptodactyly), that distal limb contractures are caused by primary abnormalities of tendon growth and development [Hall et al., 1982a; O'Brien et al., 1984]. Additionally, mutations in extracellular matrix proteins such as fibrillin-2 (FBN2), which may partly guide tendon placement, can cause congenital contractures [Putnam et al., 1995]. Furthermore, at least one member of a kindred had to exhibit at least two of the following major diagnostic criteria in order to be included within the DA disorders (the diagnostic criteria were subsequently "relaxed" for additional affected relatives). Major diagnostic criteria of the upper limbs include ulnar deviation, camptodactyly (or pseudocamptodactyly), hypoplastic and/or absent flexion creases, and/or overriding fingers at birth. Major diagnostic criteria of the lower limbs include talipes equinovarus, calcaneovalgus deformities, a vertical talus and/or metatarsus varus. For DA disorders with additional defects, at least one individual within each kindred had to meet the requirements for DA as well as exhibit the distinguishing traits characteristic of a unique form of DA. For example, at least one individual in a kindred must have distal limb contractures, short stature, and a cleft palate to be categorized as having Gordon syndrome; otherwise it would be difficult to discriminate Gordon syndrome from other DA disorders (e.g., DA1). Consequently, the kindred described by Ioan et al. [1993] meets the criteria for DAI, but not Gordon syndrome. This DA definition excluded all disorders in which structural central nervous system anomalies, cognitive delay, abnormal neurologic tests, and/or abnormal muscle biopsies were primary features. For example, a child with holoprosencephaly or congenital myotonic dystrophy and distal limb contractures would not be diagnosed with a DA disorder. Likewise, a child with motor delays secondary to congenital contractures would not be excluded from the classification. This strict definition eliminated all the known X-linked forms of arthrogryposis [Hall et al., 1982b; Hennekam et al., 1991; Zori et al., 1993]. This strict definition of DA and the diagnostic criteria were subsequently used to reclassify disorders characterized by distal limb contractures.

CLASSIFICATION

The revised and extended DA classification is presented in Table I. The DA disorders with only distal limb contractures remain categorized as DA1 (formerly DAI). DA disorders with additional consistent and distinctive anomalies are labeled alphanumerically. The order of inclusion is such that disorders more similar to DA1 are identified with a lower number: DA2 (Freeman-Sheldon syndrome) is more similar to DA1 than DA9 (congenital contractural arachnodactyly) is to DA1. New designations can be assigned as novel conditions meeting the DA definition and strict criteria are described. Subtypes can be recorded as genes for each of these disorders are mapped and cloned.

The basic structure of the original classification remains largely intact. DA1 has been described in more than 30 families including 12 families that we have studied [Bamshad et al., 1994 and in press; Dhaliwal et al., 1985; Hall et al., 1975, 1982a; Ioan et al., 1993; McCormack et al., 1980; Sallis and Beighton, 1972; Stevenson et al., 1975]. DA1 is now divided into DA1A and DA1B depending upon whether the disease allele segregating in a family maps to the DA1A locus on chromosome 9 [Bamshad et al., 1994]. The chromosome 9 locus was mapped in a single large Utah kindred and genotypic data suggest that at least three additional families map to the same locus while eight families map to at least one additional locus [Bamshad, unpublished data]. It is possible that DA1 will eventually be further subdivided by mapping studies.

DA3 (Gordon syndrome and formerly DAIIA) is defined by at least ten published families, and its traits

TABLE I. The Distal Arthrogryposes

Syndrome	Former label	New label	OMIM number
Distal arthrogryposis type 1A	DAI	DA1	108120
Distal arthrogryposis type 2 (Freeman-Sheldon syndrome)	None	DA2	193700
Distal arthrogryposis type 3 (Gordon syndrome)	DAIIA	DA3	114300
Distal arthrogryposis type 4 (scoliosis)	DAIIB	DA4	
Distal arthrogryposis type 5 (opthalmoplegia, ptosis)	DAIID	DA5	108145
Distal arthrogryposis type 6 (sensorineural hearing loss)	None	DA6	
Distal arthrogryposis type 7 (trismus pseudo-camptodactyly)	None	DA7	158300
Distal arthrogryposis type 8 (autosomal dominant multiple pterygium syndrome)	None	DA8	178110
Distal arthrogryposis type 9 (congenital contractural arachnodactyly)	None	DA9	121050

seem to be consistent and well circumscribed [Gordon et al., 1969; Halal and Fraser, 1979; Hall et al., 1982a,b; Higgins, 1972; Moldenhauer, 1964; Robinow and Johnson, 1981; Say, 1980]. Scoliosis is a consistent manifestation in families with DA4 (formerly DAIIB), and this disorder has been described in at least two families [Baraitser, 1982; Hall et al., 1982a]. Likewise, the characteristics of DA5 (formerly DAIID) are well defined. It has been described in at least 12 individuals in seven families [Bamshad, unpublished data; Hall et al., 1982a; Krieger and Espiritu, 1972; Lai et al., 1991; Sack, 1978; Schrander-Stumpel, 1993]. Although ptosis is described as the most distinctive anomaly, we think that an external ophthalmoplegia is the most consistent trait among affected individuals. DAIIC and DAIIE have been eliminated from the revised classification. DAIIC was defined in two sporadic cases, and families with DAIIC have not been described. DAIIE was associated with a variety of additional defects including cognitive delay, and although these defects may define a specific disorder, no familial cases have been reported.

Five disorders which were not part of the original classification are incorporated in the revised version. The Freeman-Sheldon syndrome (DA2) has been documented in more than 20 families [Bamshad, unpublished data; Carey et al., 1993; Hall et al., 1982a] and numerous sporadic cases [Carey et al., 1993; Hall et al., 1982a; Freeman and Sheldon, 1938]. The most discriminating traits of FSS are its facial characteristics, in particular a small mouth with pursed lips and paramedian grooves between the lower lip and tip of the chin. Other distinctive findings include down-slanting palpebral fissures, hypoplastic alae nasi, prominent nasolabial folds, and a long philtrum. Anomalies overlapping with other DA disorders include scoliosis and mild cervical pterygia.

DA6 is distal arthrogryposis associated with a sensorineural hearing loss (originally described by Stewart and Bergstrom [1971]). We have also studied a large kindred with camptodactyly, microcephaly and sensorineural hearing loss (Bamshad, unpublished data). In each of these families the distal contractures appear to be limited to the upper limbs.

Trismus-pseudocamptodactyly (TPC; DA7) has been described in numerous families. The most characteristic defect in TPC is the unusual camptodactyly of the digits upon dorsiflexion of the hands. Trismus is a necessary prerequisite for making a diagnosis of TPC although it is relatively non-specific among the DA disorders. Short stature has also been reported among individuals with TPC.

Autosomal dominant multiple pterygium syndrome [Kawira and Bender, 1985; McKeown and Harris, 1988] is characterized by distal limb contractures, short stature, scoliosis, multiple pterygia, and distinctive facial anomalies. It is included in the DA classification as DA8.

Congenital contractural arachnodactyly (CCA) is incorporated into the classification as DA9. CCA was initially described by Beals and Hecht [1971] and was reported subsequently in more than 40 families [Viljoen, 1994]. It is characterized by a marfanoid habitus, an unusual external ear labeled "crumpled," arachnodactyly, camptodactyly, and foot deformities. Although it is considered a DA disorder by the definition and criteria that we have proposed, it differs substantially from other DAs. Proximal joint involvement (elbow and knee) is more common, foot deformities are less frequent and less severe, and muscular hypoplasia is more common in CCA than other DAs. Cardiac abnormalities such as mitral valve prolapse also occur in CCA but not other DA disorders. A gene for CCA has been mapped to the FBN2 locus at 5q23-31 [Tsipouras et al., 1992] and mutations of FBN2 have been identified in some families with CCA [Putnam et al., 1995].

DISCUSSION

Hall recognized more than 150 conditions with multiple congenital joint contractures as a component manifestation [Hall, 1985, 1989, 1992]. The list includes many aneuploidy syndromes, skeletal dysplasias, multiple congenital anomaly syndromes, and

neuromuscular disorders. Cause and pathogenesis of multiple congenital contractures in these disorders are heterogeneous. Hall et al. [1982a] defined a group of disorders characterized mainly, but not exclusively, by abnormalities of the distal limbs and called them the distal arthrogryposes. Not all of the conditions described by Hall et al. [1982a] were heritable. We have revised and extended the DA classification by creating diagnostic criteria, outlining a strict definition and proposing similarities between different DA disorders. The disorders that are included in the revised classification have similar characteristics. These include 1) a consistent pattern of distal joint involvement, 2) limited proximal joint involvement, 3) an autosomal dominant inheritance pattern, 4) reduced penetrance, and 5) variable expressivity.

There is significant variability of clinical expression within all of the DAs, especially DA1 and DA2 (FSS) [Bamshad et al., 1996; Carey et al., 1993]. Moreover, a distinction between DAs can be difficult if evaluating a sporadic case but is usually straightforward in a familial context. However, some families are difficult to classify. For example, in the Maori kindred described by Klemp and Hall [1995] different individuals have been diagnosed with DA1, DA2 (FSS), or DA9 (CCA). Hall et al. [1982a] re-classified a FSS kindred reported by Jorgenson [1974] as DA1, and Carey et al. [1993] evaluated 35 individuals diagnosed with DA2 (FSS) and re-classified some patients with DA1 or DA8 (ADMPS). Aside from the distal limb contractures, the distributions of defects between DAs demonstrate moderate overlap. Cleft palate occurs in DA3 (Gordon syndrome) and has been reported in DA8 (ADMPS), while a small mouth is described in DA1, DA2 (FSS), and DA8 (ADMPS). Short stature is described variably in DA1, DA2 (FSS), DA3 (Gordon syndrome), DA7 (TPC), and DA8 (ADMPS).

The DA classification excludes most of the disorders categorized as "camptodactyly" syndromes. These disorders been described in large part by Dr. Goodman and Dr. Cantú [Cantú et al., 1980; Goodman et al., 1972, 1976; Rozin et al., 1984]. In contrast, Rozin et al. [1984] include most of the DA disorders within the camptodactyly syndromes. Although many of the camptodactyly disorders are characterized by foot and hand deformities and thus diagnostic confusion with the DAs is possible, camptodactyly is often a non-specific physical finding. In fact, the camptodactyly classification of Rozin et al. [1984] includes chromosomal and neurogenic disorders as well as single gene disorders (e.g., Marfan syndrome). Although abnormal regulation of different elements in a developmental pathway(s) may produce the same anomaly (i.e., camptodactyly), the causes of the "camptodactyly syndromes" are diverse and we think the DA disorders should be more narrowly defined.

The purpose of our classification is to identify a grouping of discrete conditions that may be related etiologically to each other. We are using the label "DA" as a specific diagnostic term similar to the use of the term "neurofibromatosis" or "chondrodystrophy." This is different from the way that the term "syndromes with radial aplasia" or "syndromes with craniosynostosis" would be used. The former use is specific for a pathogenically related group of disorders while the latter usage simply lists syndromes characterized by a common defect.

Optimally this organization will be the framework for a DA classification based on underlying molecular and physiologic abnormalities. We suggest that clinicians use the label DA to refer to individuals having one of these discrete DA disorders. Other conditions should be referred to descriptively using phrases such as "distal limb contractures." We hope that this revision is recognized as refining and extending the classification proposed by Hall et al. [1982a], not replacing it entirely. This revision will facilitate the mapping of genes causing these malformations, and we anticipate that further changes will be incorporated as we unravel the genetic basis of DA disorders.

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