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ORIGINAL INVESTIGATION

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Recessive Schwartz-Jampel syndrome (SJS): confirmation of linkage to chromosome 1p, evidence of genetic homogeneity and reduction of the SJS locus to a 3-cM interval

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Abstract Schwartz-Jampel syndrome (SJS), or chondrodystrophic myotonia, is a rare autosomal recessive disorder characterized by generalized myotonia resulting in a particular, recognizable facies and osteoarticular abnormalities. Some of us have recently shown genetic linkage of SJS to a locus on 1p34–p36.1 in five families. Here, we show by homozygosity mapping and segregation analysis that eight new families are most likely linked to the SJS locus on chromosome 1, confirming the localization of SJS to chromosome 1p and suggesting genetic homogeneity. Recombination events reduced the SJS locus from a genetic interval of 8 to 3 cM, which should facilitate the identification of the SJS gene. Low clinical variability was observed between the studied families, except for osteoarticular abnormalities. Since the severity and the location of osteoarticular abnormalities varied from one individual to another, even in the same families, other factors than the SJS gene itself, genetic or epigenetic, might contribute to the phenotype.

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Introduction

Schwartz-Jampel syndrome (SJS), also known as chondrodystrophic myotonia, is an autosomal recessive disorder characterized by generalized myotonia, joint contractures, skeletal abnormalities and facial dysmorphism (Viljoen and Beighton 1992). The age at onset is usually in childhood. Earlier onsets have, however, been reported, even at birth or during pregnancy (Cao et al. 1978; Hunziker et al. 1989). Facial myotonia results in a typical facies with blepharophimosis and fixed facial expression. Skeletal abnormalities include hip dysplasia, bowing of the leg diaphyses and irregular epiphyses (Viljoen and Beighton 1992).

There is no known biochemical, cytogenetic or pathological defect specific to SJS. Electromyography (EMG) has revealed continuous electrical activity even during rest and generalized anesthesia. Myotonic discharges begin and end abruptly, and consist of several potential components. Their origin, neurogenic or myogenic, is not known: muscle fiber activity evidenced at rest by EMG studies was abolished following perfusion with D-tubocurarine in some reports (Taylor et al. 1972), whereas in others, the neuromuscular block did not decrease the extent of the muscle activity (Brown et al. 1975; Cao et al. 1978). Motor and sensory nerve conduction velocity measurements were in the normal range (Topaloglu et al. 1993). New light was shed on this debate when impaired muscle Na^+ channel inactivation was demonstrated in muscle biopsies of a patient displaying a phenotype close to SJS (Lehmann-Horn et al. 1990; Spaans et al. 1990). The identification of a Gly1306Glu mutation in the Na^+ channel gene established that this patient suffered from

Na^+ channel myotonia, called myotonia permanens, and not from SJS (Rüdel et al. 1993), and also provided the first molecular marker to distinguish myotonic syndromes resembling SJS, from SJS.

Using homozygosity mapping, some of us recently reported that SJS mapped within an 8-cM interval on chromosome 1p34–p36.1 (Nicole et al. 1995). We confirm and extend these findings, showing that 13 families of different ethnic origin are linked to this locus. Our results indicate genetic homogeneity, at least in the group of families studied, and reduce the SJS locus to a 3-cM interval. They also suggest that linkage to the SJS locus on chromosome 1p may be considered as a genetic marker that differentiates SJS from other syndromes sharing phenotypic features.

Subjects and methods

Patients

Thirteen families with SJS were available for this study, nine of which were consanguineous (Table 1, families A–I). In addition to the five families studied in Nicole et al. (1995), three consanguineous Tunisian families (A–C), one consanguineous family from South Africa (family I) and one nonconsanguineous Algerian family (family K), eight new families were studied, five of which were consanguineous and originated from Turkey (families D–H). The three remaining families originated from France (family J), The Netherlands (family L) and Mexico (family M) (Table 1). Inheritance in all families was recessive. Patient status was established by one of the authors. Families B, C, E, F, G, I, J, and M have already been described (Beighton 1973; Desbois et al. 1977; Ben Hamida et al. 1991; Soussi-Yanicostas et al. 1991; Figuera et al. 1993; Topaloglu et al. 1993). Altogether, 22 affected and 47 unaffected individuals were studied.

Genotyping

Blood samples were collected from all consenting family members and controls, according to the Helsinki convention. High molecular weight DNA was extracted as described (Gusella 1986). Genotyping was performed by the polymerase chain reaction (PCR) blotting technique (Gyapay et al. 1994). Polymorphic dinucleotide repeats from the Généthon human genetic map used in this study are described in Gyapay et al. (1994). To refine the SJS locus, six unpublished markers developed at Généthon were used: (1) AFMb040yb1 (forward primer: 5′-TGACTTCAGTGAGGCTGC-3′, reverse primer: 5′-CGGATACAAGGGCTTTTC-3′, PCR product (264 bp, heterozygosity = 0.77); (2) AFM303tg1 (forward primer: 5′-GGCTCCTGAACCTGGG-3′, reverse primer: 5′-AGCTTTGGCTGACCTTCC-3′, PCR product (267 bp, heterozygosity = 0.87); (3) AFMb016yb5 (forward primer: 5′-GGAGAATTGCTTGAACCTG-3′, reverse primer: 5′-GATTGCTTTCATGTATTGGC-3′, PCR product (176 bp, heterozygosity = 0.75); (4) AFMa114yd5 (forward primer: 5′-AAGAGTTGTCCAACCAAATTG-3′, reverse primer: 5′-GAATCTGGGATGGGATGT-3′, PCR product (100 bp, heterozygosity = 0.53); (5) AFMa048yh1 (forward primer: 5′-AGCTCTAATGCCCGAAACC-3′, reverse primer: 5′-TCTTTCATTCAGTCGCTCCC-3′, PCR product (141 bp, heterozygosity = 0.81); (6) AFMa300wb9 (forward primer: 5′-GGGAGTCCCTCAAACCAAAAGTTTA-3′, reverse primer: 5′-ACCACACCCGGCCTAGATT-3′, PCR product (135 bp, heterozygosity = 0.72). The relative positions of the genetic markers are shown in Table 1. The proposed order is deduced from the Généthon map and the recombination events observed in our families. The dinucleotide repeat alleles were numbered according to decreasing size on the same gel, to allow comparison between families (Table 1). Pair-wise lod scores were calculated using the MLINK program of the LINKAGE package (version 5.1) (Lathrop et al. 1985). Families L and M were not included in this calculation, because of their limited informativity (only one affected individual and no consanguinity loop). A disease gene frequency of 0.001 and a penetrance of 100% were assumed. Because the calculation of lod scores is sensitive to allelic frequency variation in consanguineous families, true allelic frequencies calculated in the control population were used. Genetic homogeneity was tested with the HOMOG program (version 3.10) (Ott 1991).

Allelic frequencies

Frequencies of AFMa114yd5 alleles were calculated in both a North-African and a European population: 43 unrelated individuals (86 chromosomes) from North Africa and 36 European individuals (72 chromosomes) were genotyped for AFMa114yd5. Allelic frequencies for the affected chromosome were deduced from both Mediterranean (Algerian, Tunisian and Turkish) and all families. Allelic frequencies in the affected population and in controls were compared using the χ^2 test.

Results

Clinical features of the SJS families

The 13 families presented the clinical features of SJS. The age at onset ranged from the first months of life to 4 years old with a mean at 21 months. Constant clinical features included myotonia, even if it was less intense in some cases (families B, C and J), the particular facies with a blepharophimosis, short stature and a myotonic EMG pattern. However, the osteoarticular abnormalities varied from one patient to another, even in the same families (families A, J). One patient (family M) presented no osteoarticular abnormalities. At onset, the severity of the clinical signs also differed among patients. For example, the first noticeable clinical sign was a difficult in walking in the Tunisian families (families A–C), whereas deformities or abnormal posture were first noted in families I and L, and a blepharophimosis in families E–H, J and M. Apart from the osteoarticular abnormalities, which can be evidenced by objective criteria (photographs and X-rays), the first clinical sign detected may depend on the examiner.

Genetic linkage to the SJS locus on chromosome 1p in eight new families

Nine markers, covering a genetic interval of 8-cM flanked by D1S199 and D1S234, were genotyped in the SJS families (Table 1). A region of homozygosity extending over at least 6-cM was demonstrated in affected children of consanguineous families B, C, and F–I (see family H in Fig. 1; Table 1). In inbred families D and E, homozygosity of affected children was found for four contiguous markers, corresponding to a genetic interval of 4 cM (Table 1). Segregation analyses in nonconsanguineous families were also compatible with genetic linkage to the

Table 1 Chromosome 1p haplotypes linked to Schwartz-Jampel Syndrome (SJS) in 13 families. No indicated allele means heterozygosity for at least one inbred affected child [*ND* not determined, *NI* noninformative (with the homozygous allele in brackets] An arrowhead indicates a recombination event on the affected chromosome; ⊢——⊣ 1 cM

Family[a]	Degree of consanguinity[a]	Ethnic origin	SJS locus										
			D1S199	AFMb040yb1	AFM343za9	AFM303tg1 AFMb016yb5		D1S478	AFMa114yd5	AFMa048yh1	D1S482	AFMa300wb9	D1S234
			tel										*cen*
A	2	Tunisia						▼	5	9	1	2	
B	1	Tunisia	11	8	3	3	4	1	5	$NI_{(9)}$	6	1	6
C	1	Tunisia	10	4	3	9	7	4	5	8	$NI_{(6)}$		
D	1.5	Turkey			▼	$NI_{(9)}$	$NI_{(6)}$	$NI_{(3)}$	6				ND
E	1	Turkey				7	4	4	5				ND
F	1	Turkey	8	4	5	7	$NI_{(6)}$	6	5	3	$NI_{(6)}$	4	ND
G	1.5	Turkey			3	3	6	4	$NI_{(5)}$	5	3	4	ND
H	1	Turkey	12	4	5	8	3	3	$NI_{(6)}$	9	6	3	ND
I	2	South Africa	4	3	4	8	6	4	5	12	$NI_{(6)}$	4	5
J	0	France	12	6	4	3	6	4	$NI_{(5)}$	3	4	5	ND
			5	9	2	9	6	4		5	6	1	
K	0	Algeria	7	7	3	10	1	7	5	8	5	4	4
			12	4	3	9	5	4	6	6	7	1	6
L	0	The Netherlands	ND	6	2	5	5	3	$NI_{(5)}$	ND	ND	5	ND
				4	3	3	6	3				4	
M	0	Mexico	10	1	2	8	$NI_{(6)}$	3	4	4	6	3	ND
			12	2	2	5		3	5	8	5	2	

[a] The degree of cousinship is given for each consanguineous family. A degree of 1 is a first-cousin mating and 2 is a second-cousin mating. 0 means no known consanguinity and 1.5 corresponds to marriage between an individual and the child of his first-degree cousin

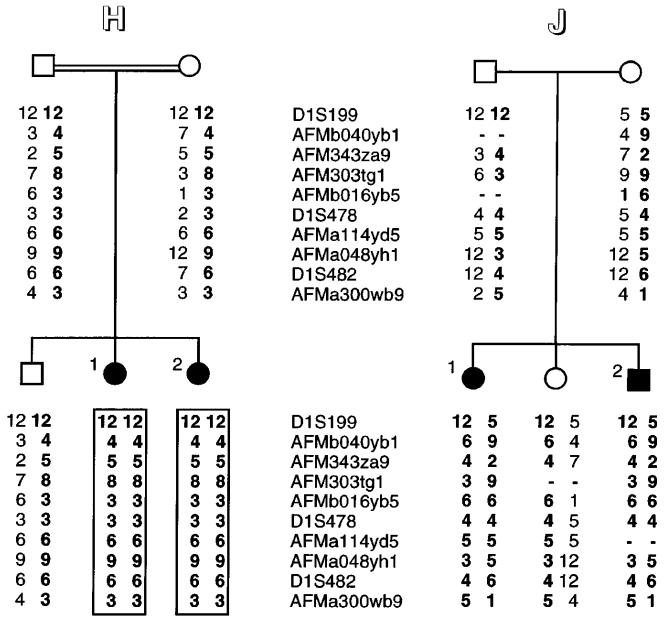

Fig. 1 Pedigrees of two families not previously published. Families H and J are consanguineous and nonconsanguineous families, respectively. The haplotype linked to Schwartz-Jampel syndrome (SJS) is in *bold type*. The homozygous region for inbred children is *boxed*. – not determined

SJS locus (see family J in Fig. 1). Pair-wise lod scores were positive for all markers tested. A peak lod score (z_{max}) of 5.44 was obtained for a recombination fraction of $\theta_{max} = 0.00$ for AFMa114yd5. This z_{max} was small because of the high frequency (0.56) of the linked allele 5 (100 bp) in the North African control population. HOMOG analysis confirmed the most likely genetic linkage of the families to the SJS locus on chromosome 1p (data not shown).

Heterozygosity of affected children in families D and E for AFMa048yh1 and a recombination event in family A reduced the SJS interval to 3-cM, flanked by D1S478 and AFMa048yh1 and comprising AFMa114yd5 (Table 1).

No preferential allelic association in SJS families

As indicated in Table 1, different haplotypes segregated with the disease, even in families of similar ethnic background. Since AFMa114yd5 was the only marker within the SJS interval, linkage disequilibrium between SJS and this marker was sought. No difference between the distributions of the AFMa114yd5 allele 5 in affected and control Mediterranean chromosomes was found, this allele being the most represented in the two populations (0.76 and 0.56, respectively).

Discussion

We analyzed 13 families of different background diagnosed as SJS, 8 of which had not previously been studied at the molecular level. All of them showed evidence of linkage to the SJS locus on chromosome 1p previously reported (Nicole et al. 1995). We were able to reduce the SJS locus from 8 to 3-cM. The latter contains several genes such as a receptor tyrosine kinase, *ERK* (Saito et al. 1995), and collagen genes, Col8A2 and Col16A1 (Muragaki et al. 1991; Pan et al. 1992). A kinase has been involved in another multisystemic myotonic disorder, i.e., myotonic dystrophy (Buxton et al. 1992; Fu et al. 1992; Mahadevan et al. 1992). Mutations in collagen genes such as Col11A2 and Col2A1 have been respectively identified in patients with an autosomal dominant form of Stickler syndrome and spondyloepiphyseal dysplasia (Vikkula et al. 1993, 1995).

SJS is a rare autosomal recessive disorder, expressed mostly in consanguineous families. One may therefore expect a small number of founding chromosomes. In contrast with other recessive disorders such as ataxia with vitamin E deficiency (Doerflinger et al. 1995) or chromosome 13-linked Duchenne-like muscular dystrophy (LGMD2C) (Ben-Othmane et al. 1995), we did not find a preferential association with an allele, although the tested marker (AFMa114yd5) was located within the SJS interval. The allele associated with 76% of SJS chromosomes was the most frequent in the population. Consequently, a preferential allelic association with SJS may be masked by the high frequency of this allele in the control population, but the low density of markers in the 3-cM SJS interval does not permit conclusive analysis. The characterization of new markers within the SJS interval will be necessary to address this issue.

Clinical data from the studied SJS families showed variation in the expression of the phenotype both between and within families, mostly in the severity and the location of the osteoarticular abnormalities, although genetic homogeneity was found. This has already been encountered in autosomal recessive disorders such as polycystic kidney disease or spinal muscular atrophy (Brzustowicz et al. 1990; Melki et al. 1990; Guay-Woodford et al. 1995). The clinical variability in SJS might be due to allelic mutations at the SJS locus. This hypothesis would not explain the intrafamilial variation. Genetic or epigenetic factors other than the SJS gene itself might contribute to the variable phenotype. One author has proposed that contiguous genes are involved: a complex rearrangement involving more than one gene would be necessary to obtain the complete SJS phenotype (Figuera et al. 1993). Complicating the issue, it is not known whether the osteoarticular abnormalities are primary, due to the deleterious action of the SJS locus, or secondary due to the myotonia. In favor of the latter hypothesis, mechanical tension has been suggested to alter local bone architecture by influencing bone remodeling (Erlebacher et al. 1995). For example, mice with inactivated *myf-5* and *myoD* genes lack deltoid tuberosity of the humerus and have a short anteriorly split sternum (Rudnicki et al. 1993). The intensity of myotonia and muscle activity during osteoarticular development could therefore explain the variable skeletal abnormalities.

In conclusion, our confirmation of genetic linkage of 13 SJS families to the SJS locus on chromosome 1p provides a new marker for the classification of myotonic disorders. The reduction of the SJS interval to a genetic distance of 3-cM should permit the rapid identification of the SJS gene.

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