

List of publications on ARSACS

As of January 19, 2019

(1-163)

1. Zesiewicz TA, Wilmot G, Kuo SH, Perlman S, Greenstein PE, Ying SH, et al. Comprehensive systematic review summary: Treatment of cerebellar motor dysfunction and ataxia: Report of the Guideline Development, Dissemination, and Implementation Subcommittee of the American Academy of Neurology. *Neurology*. 2018;90(10):464-71.
2. Vogel AP, Rommel N, Oettinger A, Stoll LH, Kraus EM, Gagnon C, et al. Coordination and timing deficits in speech and swallowing in autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS). *J Neurol*. 2018;265(9):2060-70.
3. Vogel AP, Rommel N, Oettinger A, Stoll LH, Kraus EM, Gagnon C, et al. Coordination and timing deficits in speech and swallowing in autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS). *J Neurol*. 2018.
4. Vill K, Muller-Felber W, Glaser D, Kuhn M, Teusch V, Schreiber H, et al. SACS variants are a relevant cause of autosomal recessive hereditary motor and sensory neuropathy. *Hum Genet*. 2018;137(11-12):911-9.
5. Tardif J, Pratte A, Laberge AM. Experience of carrier couples identified through a population-based carrier screening pilot program for four founder autosomal recessive diseases in Saguenay-Lac-Saint-Jean. *Prenat Diagn*. 2018;38(1):67-74.
6. Synofzik M, Nemeth AH. Recessive ataxias. *Handb Clin Neurol*. 2018;155:73-89.
7. Souza PVS, Bortholin T, Naylor FGM, Pinto W, Oliveira ASB. Early-onset axonal Charcot-Marie-Tooth disease due to SACS mutation. *Neuromuscul Disord*. 2018;28(2):169-72.
8. Picher-Martel V, Dupre N. Current and Promising Therapies in Autosomal Recessive Ataxias. *CNS Neurol Disord Drug Targets*. 2018;17(3):161-71.
9. Parkinson MH, Bartmann AP, Clayton LMS, Nethisinghe S, Pfundt R, Chapple JP, et al. Optical coherence tomography in autosomal recessive spastic ataxia of Charlevoix-Saguenay. *Brain*. 2018;141(4):989-99.
10. Menade M, Kozlov G, Trempe JF, Pande H, Shenker S, Wickremasinghe S, et al. Structures of ubiquitin-like (Ubl) and Hsp90-like domains of saccin provide insight into pathological mutations. *J Biol Chem*. 2018;293(33):12832-42.
11. Machuca C, Vilches A, Clemente E, Pascual-Pascual SI, Bolinches-Amoros A, Artero Castro A, et al. Generation of human induced pluripotent stem cell (iPSC) line from an unaffected female carrier of mutation in SACSIN gene. *Stem Cell Res*. 2018;33:166-70.
12. Li S, Chen Y, Yuan X, Wei Q, Ou R, Gu X, et al. [Identification of compound heterozygous mutations of SACS gene in two patients from a pedigree with spastic ataxia of Charlevoix-Saguenay]. *Zhonghua Yi Xue Yi Chuan Xue Za Zhi*. 2018;35(4):507-10.

13. Lessard I, Brais B, Cote I, Lavoie C, Synofzik M, Mathieu J, et al. Assessing mobility and balance in Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay population: Validity and reliability of four outcome measures. *J Neurol Sci.* 2018;390:4-9.
14. Incecik F, Herguner OM, Bisgin A. Autosomal-Recessive Spastic Ataxia of Charlevoix-Saguenay: A Turkish Child. *J Pediatr Neurosci.* 2018;13(3):355-7.
15. Gentil BJ, Lai GT, Menade M, Lariviere R, Minotti S, Gehring K, et al. Sacsin, mutated in the ataxia ARSACS, regulates intermediate filament assembly and dynamics. *FASEB J.* 2018:fj201801556R.
16. Gagnon C, Lessard I, Lavoie C, Cote I, St-Gelais R, Mathieu J, et al. An exploratory natural history of ataxia of Charlevoix-Saguenay: A 2-year follow-up. *Neurology.* 2018;91(14):e1307-e11.
17. Gagnon C, Lessard I, Brais B, Cote I, Lavoie C, Synofzik M, et al. Validity and Reliability of Outcome Measures Assessing Dexterity, Coordination, and Upper Limb Strength in Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay. *Arch Phys Med Rehabil.* 2018.
18. Gagnon C, Brais B, Lessard I, Lavoie C, Cote I, Mathieu J. From motor performance to participation: a quantitative descriptive study in adults with autosomal recessive spastic ataxia of Charlevoix-Saguenay. *Orphanet J Rare Dis.* 2018;13(1):165.
19. Dougherty SC, Harper A, Al Saif H, Vorona G, Haines SR. A Chromosomal Deletion and New Frameshift Mutation Cause ARSACS in an African-American. *Front Neurol.* 2018;9:956.
20. Biswas A, Varman M, Yoganathan S, Subhash PK, Mani S. Teaching NeuroImages: Autosomal recessive spastic ataxia of Charlevoix-Saguenay: Typical MRI findings. *Neurology.* 2018;90(14):e1271-e2.
21. Audet O, Bui HT, Allisse M, Comtois AS, Leone M. Assessment of the impact of an exercise program on the physical and functional capacity in patients with autosomal recessive spastic ataxia of Charlevoix-Saguenay: An exploratory study. *Intractable Rare Dis Res.* 2018;7(3):164-71.
22. Arellano CM, Vilches A, Clemente E, Pascual-Pascual SI, Bolinches-Amoros A, Castro AA, et al. Generation of a human iPSC line from a patient with autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS) caused by mutation in SACSIN gene. *Stem Cell Res.* 2018;31:249-52.
23. Ady V, Toscano-Marquez B, Nath M, Chang PK, Hui J, Cook A, et al. Altered synaptic and firing properties of cerebellar Purkinje cells in a mouse model of ARSACS. *J Physiol.* 2018;596(17):4253-67.
24. Zeng H, Tang JG, Yang YF, Tan ZP, Tan JQ. A Novel Homozygous SACS Mutation Identified by Whole-Exome Sequencing in a Consanguineous Family with Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay. *Cytogenet Genome Res.* 2017;152(1):16-21.
25. Vogel AP, Rommel N, Sauer C, Horger M, Krumm P, Himmelbach M, et al. Clinical assessment of dysphagia in neurodegeneration (CADN): development, validity and reliability of a bedside tool for dysphagia assessment. *J Neurol.* 2017;264(6):1107-17.
26. Saffie P, Kauffman MA, Fernandez JM, Acosta I, Espay AJ, de la Cerda A. Teaching Video NeuroImages: Spastic ataxia syndrome: The Friedreich-like phenotype of ARSACS. *Neurology.* 2017;89(14):e178-e9.
27. Lessard I, Lavoie C, Cote I, Mathieu J, Brais B, Gagnon C. Validity and reliability of the LEMOCOT in the adult ARSACS population: A measure of lower limb coordination. *J Neurol Sci.* 2017;377:193-6.

28. Krygier M, Konkel A, Schinwelski M, Rydzanicz M, Walczak A, Sildatke-Bauer M, et al. Autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS) - A Polish family with novel SACS mutations. *Neurol Neurochir Pol.* 2017;51(6):481-5.
29. Iqbal Z, Rydning SL, Wedding IM, Koht J, Pihlstrom L, Rengmark AH, et al. Targeted high throughput sequencing in hereditary ataxia and spastic paraplegia. *PLoS One.* 2017;12(3):e0174667.
30. Duncan EJ, Lariviere R, Bradshaw TY, Longo F, Sgarioto N, Hayes MJ, et al. Altered organization of the intermediate filament cytoskeleton and relocalization of proteostasis modulators in cells lacking the ataxia protein saccin. *Hum Mol Genet.* 2017;26(16):3130-43.
31. Ding M, Weng C, Fan S, Cao Q, Lu Z. Purkinje Cell Degeneration and Motor Coordination Deficits in a New Mouse Model of Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay. *Front Mol Neurosci.* 2017;10:121.
32. Burguez D, Oliveira CM, Rockenbach M, Fussiger H, Vedolin LM, Winckler PB, et al. Autosomal recessive spastic ataxia of Charlevoix-Saguenay: a family report from South Brazil. *Arq Neuropsiquiatr.* 2017;75(6):339-44.
33. Bui HT, Gagnon C, Audet O, Mathieu J, Leone M. Measurement properties of a new wireless electrogoniometer for quantifying spasticity during the pendulum test in ARSACS patients. *J Neurol Sci.* 2017;375:181-5.
34. Bui HT, Audet O, Mathieu J, Gagnon C, Leone M. Computer-based assessment of upper-limb incoordination in autosomal recessive spastic ataxia of Charlevoix-Saguenay patients: A pilot study. *J Neurol Sci.* 2017;380:68-73.
35. Borruat FX, Holder GE, Bremner F. Inner Retinal Dysfunction in the Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay. *Front Neurol.* 2017;8:523.
36. van Lint M, Hoornaert K, Ten Tusscher MPM. Retinal nerve fiber layer thickening in ARSACS carriers. *J Neurol Sci.* 2016;370:119-22.
37. Tranchant C, Anheim M. Movement disorders in mitochondrial diseases. *Rev Neurol (Paris).* 2016;172(8-9):524-9.
38. Shah CT, Ward TS, Matsumoto JA, Shildkrot Y. Foveal hypoplasia in autosomal recessive spastic ataxia of Charlevoix-Saguenay. *J AAPOS.* 2016;20(1):81-3.
39. Palmio J, Karppa M, Baumann P, Penttila S, Moilanen J, Udd B. Novel compound heterozygous mutation in SACS gene leads to a milder autosomal recessive spastic ataxia of Charlevoix-Saguenay, ARSACS, in a Finnish family. *Clin Case Rep.* 2016;4(12):1151-6.
40. Nascimento FA, Canafoglia L, Aljaafari D, Muona M, Lehesjoki AE, Berkovic SF, et al. Progressive myoclonus epilepsy associated with SACS gene mutations. *Neurol Genet.* 2016;2(4):e83.
41. Liu L, Li XB, Zi XH, Shen L, Hu Zh M, Huang Sh X, et al. A novel hemizygous SACS mutation identified by whole exome sequencing and SNP array analysis in a Chinese ARSACS patient. *J Neurol Sci.* 2016;362:111-4.
42. Bradshaw TY, Romano LE, Duncan EJ, Nethisinghe S, Abeti R, Michael GJ, et al. A reduction in Drp1-mediated fission compromises mitochondrial health in autosomal recessive spastic ataxia of Charlevoix Saguenay. *Hum Mol Genet.* 2016;25(15):3232-44.
43. Armour CM, Smith A, Hartley T, Chardon JW, Sawyer S, Schwartzentruber J, et al. Syndrome disintegration: Exome sequencing reveals that Fitzsimmons syndrome is a co-occurrence of multiple events. *Am J Med Genet A.* 2016;170(7):1820-5.
44. Ali Z, Klar J, Jameel M, Khan K, Fatima A, Raininko R, et al. Novel SACS mutations associated with intellectual disability, epilepsy and widespread supratentorial abnormalities. *J Neurol Sci.* 2016;371:105-11.

45. Sanchez MG, Perez JE, Perez MR, Redondo AG. Novel SACS mutation in autosomal recessive spastic ataxia of Charlevoix-Saguenay. *J Neurol Sci.* 2015;358(1-2):475-6.
46. Pilliod J, Moutton S, Lavie J, Maurat E, Hubert C, Bellance N, et al. New practical definitions for the diagnosis of autosomal recessive spastic ataxia of Charlevoix-Saguenay. *Ann Neurol.* 2015;78(6):871-86.
47. Nickerson SL, Marquis-Nicholson R, Claxton K, Ashton F, Leong IU, Prosser DO, et al. SNP Analysis and Whole Exome Sequencing: Their Application in the Analysis of a Consanguineous Pedigree Segregating Ataxia. *Microarrays (Basel).* 2015;4(4):490-502.
48. Li X, Menade M, Kozlov G, Hu Z, Dai Z, McPherson PS, et al. High-Throughput Screening for Ligands of the HEPN Domain of Sacsin. *PLoS One.* 2015;10(9):e0137298.
49. Li X, Gehring K. Structural studies of parkin and sacsin: Mitochondrial dynamics in neurodegenerative diseases. *Mov Disord.* 2015;30(12):1610-9.
50. Lariviere R, Gaudet R, Gentil BJ, Girard M, Conte TC, Minotti S, et al. Sacs knockout mice present pathophysiological defects underlying autosomal recessive spastic ataxia of Charlevoix-Saguenay. *Hum Mol Genet.* 2015;24(3):727-39.
51. Kwon KY, Huh K, Eun BL, Yoo HW, Kamsteeg EJ, Scheffer H, et al. A Probable Korean Case of Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay. *Can J Neurol Sci.* 2015;42(4):271-3.
52. Kurt S, Kartal E, Aksoy D, Cevik B, Eken AG, Sahbaz I, et al. Coexistence of autosomal recessive spastic ataxia of Charlevoix Saguenay and spondyloepiphyseal dysplasia in a Turkish patient. *J Neurol Sci.* 2015;357(1-2):290-1.
53. Criscuolo C, Procaccini C, Meschini MC, Cianflone A, Carbone R, Doccini S, et al. Powerhouse failure and oxidative damage in autosomal recessive spastic ataxia of Charlevoix-Saguenay. *J Neurol.* 2015;262(12):2755-63.
54. Blumkin L, Bradshaw T, Michelson M, Kopler T, Dahari D, Lerman-Sagie T, et al. Molecular and functional studies of retinal degeneration as a clinical presentation of SACS-related disorder. *Eur J Paediatr Neurol.* 2015;19(4):472-6.
55. Yu-Wai-Man P, Pyle A, Griffin H, Santibanez-Korev M, Horvath R, Chinnery PF. Abnormal retinal thickening is a common feature among patients with ARSACS-related phenotypes. *Br J Ophthalmol.* 2014;98(5):711-3.
56. van de Warrenburg BP, van Gaalen J, Boesch S, Burgunder JM, Durr A, Giunti P, et al. EFNS/ENS Consensus on the diagnosis and management of chronic ataxias in adulthood. *Eur J Neurol.* 2014;21(4):552-62.
57. Takiyama Y. [Japan Spastic Paraplegia Research Consortium (JASPAC)]. *Brain Nerve.* 2014;66(10):1210-7.
58. Storey E. Genetic cerebellar ataxias. *Semin Neurol.* 2014;34(3):280-92.
59. Roberts JL, Hovanes K, Dasouki M, Manzardo AM, Butler MG. Chromosomal microarray analysis of consecutive individuals with autism spectrum disorders or learning disability presenting for genetic services. *Gene.* 2014;535(1):70-8.
60. Mignarri A, Tessa A, Carluccio MA, Rufa A, Storti E, Bonelli G, et al. Cerebellum and neuropsychiatric disorders: insights from ARSACS. *Neurol Sci.* 2014;35(1):95-7.
61. McKenzie ED, Sharma PN, Parboosingh JS, Suchowersky O. Novel SACS Mutation Deviates from the French Canadian ARSACS Phenotype. *Can J Neurol Sci.* 2014;41(1):88-9.
62. Masciullo M, Silvestri G, Modoni A, Tessa A, Bianchi ML, Santorelli FM. Do not jump to easy conclusions! Lessons from pitfall in the molecular diagnosis of ARSACS. *Clin Genet.* 2014;86(4):396-7.

63. Leavitt JA, Singer W, Brown WL, Pulido JS, Brodsky MC. Retinal and pontine striations: neurodiagnostic signs of autosomal recessive spastic ataxia of Charlevoix-Saguenay. *J Neuroophthalmol.* 2014;34(4):369-71.
64. Koutras C, Braun JE. J protein mutations and resulting proteostasis collapse. *Front Cell Neurosci.* 2014;8:191.
65. Gazulla J, Mayayo-Sinues E, Benavente I, Modrego PJ, Berciano J. Ataxia of Charlevoix-Saguenay: MR and Clinical Results in Lower-Limb Musculature. *Can J Neurol Sci.* 2014;41(1):37-41.
66. Garcia-Martin E, Bambo MP, Gazulla J, Larrosa JM, Polo V, Fuertes MI, et al. [Finding of retinal nerve fiber layer hypertrophy in ataxia of Charlevoix-Saguenay patients]. *Arch Soc Esp Oftalmol.* 2014;89(5):207-11.
67. Gagnon C, Lavoie C, Lessard I, Mathieu J, Brais B, Bouchard JP, et al. The Virtual Peg Insertion Test as an assessment of upper limb coordination in ARSACS patients: a pilot study. *J Neurol Sci.* 2014;347(1-2):341-4.
68. Tzoulis C, Johansson S, Haukanes BI, Boman H, Knappskog PM, Bindoff LA. Novel SACS mutations identified by whole exome sequencing in a norwegian family with autosomal recessive spastic ataxia of Charlevoix-Saguenay. *PLoS One.* 2013;8(6):e66145.
69. Thiffault I, Dicaire MJ, Tetreault M, Huang KN, Demers-Lamarche J, Bernard G, et al. Diversity of ARSACS mutations in French-Canadians. *Can J Neurol Sci.* 2013;40(1):61-6.
70. Synofzik M, Soehn AS, Gburek-Augustat J, Schicks J, Karle KN, Schule R, et al. Autosomal recessive spastic ataxia of Charlevoix Saguenay (ARSACS): expanding the genetic, clinical and imaging spectrum. *Orphanet J Rare Dis.* 2013;8:41.
71. Stevens JC, Murphy SM, Davagnanam I, Phadke R, Anderson G, Nethisinghe S, et al. The ARSACS phenotype can include supranuclear gaze palsy and skin lipofuscin deposits. *J Neurol Neurosurg Psychiatry.* 2013;84(1):114-6.
72. Shimazaki H, Takiyama Y, Honda J, Sakoe K, Namekawa M, Tsugawa J, et al. Middle cerebellar peduncles and Pontine T2 hypointensities in ARSACS. *J Neuroimaging.* 2013;23(1):82-5.
73. Romano A, Tessa A, Barca A, Fattori F, de Leva MF, Terracciano A, et al. Comparative analysis and functional mapping of SACS mutations reveal novel insights into saccin repeated architecture. *Hum Mutat.* 2013;34(3):525-37.
74. Pyle A, Griffin H, Duff J, Bennett S, Zwolinski S, Smertenko T, et al. Late-onset saccinopathy diagnosed by exome sequencing and comparative genomic hybridization. *J Neurogenet.* 2013;27(4):176-82.
75. Prodi E, Grisoli M, Panzeri M, Minati L, Fattori F, Erbetta A, et al. Supratentorial and pontine MRI abnormalities characterize recessive spastic ataxia of Charlevoix-Saguenay. A comprehensive study of an Italian series. *Eur J Neurol.* 2013;20(1):138-46.
76. Oguz KK, Haliloglu G, Temucin C, Gocmen R, Has AC, Doerschner K, et al. Assessment of whole-brain white matter by DTI in autosomal recessive spastic ataxia of Charlevoix-Saguenay. *AJNR Am J Neuroradiol.* 2013;34(10):1952-7.
77. McElroy JP, Krupp LB, Johnson BA, McCauley JL, Qi Z, Caillier SJ, et al. Copy number variation in pediatric multiple sclerosis. *Mult Scler.* 2013;19(8):1014-21.
78. Liew WK, Ben-Omran T, Darras BT, Prabhu SP, De Vivo DC, Vatta M, et al. Clinical application of whole-exome sequencing: a novel autosomal recessive spastic ataxia of Charlevoix-Saguenay sequence variation in a child with ataxia. *JAMA Neurol.* 2013;70(6):788-91.

79. Gregianin E, Vazza G, Scaramel E, Boaretto F, Vettori A, Leonardi E, et al. A novel SACS mutation results in non-ataxic spastic paraplegia and peripheral neuropathy. *Eur J Neurol*. 2013;20(11):1486-91.
80. Garcia-Martin E, Pablo LE, Gazulla J, Vela A, Larrosa JM, Polo V, et al. Retinal segmentation as noninvasive technique to demonstrate hyperplasia in ataxia of Charlevoix-Saguenay. *Invest Ophthalmol Vis Sci*. 2013;54(10):7137-42.
81. Garcia-Martin E, Pablo LE, Gazulla J, Polo V, Ferreras A, Larrosa JM. Retinal nerve fibre layer thickness in ARSACS: myelination or hypertrophy? *Br J Ophthalmol*. 2013;97(2):238-41.
82. Duquette A, Brais B, Bouchard JP, Mathieu J. Clinical presentation and early evolution of spastic ataxia of Charlevoix-Saguenay. *Mov Disord*. 2013;28(14):2011-4.
83. Dibilio V, Cavalcanti F, Nicoletti A, Mostile G, Bruno E, Annesi G, et al. Sacsin-related spastic ataxia caused by a novel missense mutation p.Arg272His in a patient from Sicily, southern Italy. *Cerebellum*. 2013;12(4):589-92.
84. Chen Z, Wang JL, Tang BS, Sun ZF, Shi YT, Shen L, et al. Using next-generation sequencing as a genetic diagnostic tool in rare autosomal recessive neurologic Mendelian disorders. *Neurobiol Aging*. 2013;34(10):2442 e11-7.
85. Berciano J, Garcia A, Infante J. Peripheral nerve involvement in hereditary cerebellar and multisystem degenerative disorders. *Handb Clin Neurol*. 2013;115:907-32.
86. Anantharaman V, Makarova KS, Burroughs AM, Koonin EV, Aravind L. Comprehensive analysis of the HEPN superfamily: identification of novel roles in intra-genomic conflicts, defense, pathogenesis and RNA processing. *Biol Direct*. 2013;8:15.
87. Verhoeven WM, Egger JI, Ahmed AI, Kremer BP, Vermeer S, van de Warrenburg BP. Cerebellar cognitive affective syndrome and autosomal recessive spastic ataxia of charlevoix-saguenay: a report of two male sibs. *Psychopathology*. 2012;45(3):193-9.
88. Pyle A, Griffin H, Yu-Wai-Man P, Duff J, Eglon G, Pickering-Brown S, et al. Prominent sensorimotor neuropathy due to SACS mutations revealed by whole-exome sequencing. *Arch Neurol*. 2012;69(10):1351-4.
89. Narendra DP, Youle RJ. Neurodegeneration: Trouble in the cell's powerhouse. *Nature*. 2012;483(7390):418-9.
90. Miyatake S, Miyake N, Doi H, Saito H, Ogata K, Kawai M, et al. A novel SACS mutation in an atypical case with autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS). *Intern Med*. 2012;51(16):2221-6.
91. Masciullo M, Modoni A, Tessa A, Santorelli FM, Rizzo V, D'Amico G, et al. Novel SACS mutations in two unrelated Italian patients with spastic ataxia: clinico-diagnostic characterization and results of serial brain MRI studies. *Eur J Neurol*. 2012;19(8):e77-8.
92. Haga R, Miki Y, Funamizu Y, Kon T, Suzuki C, Ueno T, et al. Novel compound heterozygous mutations of the SACS gene in autosomal recessive spastic ataxia of Charlevoix-Saguenay. *Clin Neurol Neurosurg*. 2012;114(6):746-7.
93. Girard M, Lariviere R, Parfitt DA, Deane EC, Gaudet R, Nossova N, et al. Mitochondrial dysfunction and Purkinje cell loss in autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS). *Proc Natl Acad Sci U S A*. 2012;109(5):1661-6.
94. Gazulla J, Benavente I, Vela AC, Marin MA, Pablo LE, Tessa A, et al. New findings in the ataxia of Charlevoix-Saguenay. *J Neurol*. 2012;259(5):869-78.
95. De Michele G, Filla A. Other autosomal recessive and childhood ataxias. *Handb Clin Neurol*. 2012;103:343-57.

96. Vingolo EM, Di Fabio R, Salvatore S, Grieco G, Bertini E, Leuzzi V, et al. Myelinated retinal fibers in autosomal recessive spastic ataxia of Charlevoix-Saguenay. *Eur J Neurol*. 2011;18(9):1187-90.
97. Takiyama Y. [Hereditary spastic paraplegia in Japan]. *Rinsho Shinkeigaku*. 2011;51(11):1125-8.
98. Pedroso JL, Braga-Neto P, Abrahao A, Rivero RL, Abdalla C, Abdala N, et al. Autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS): typical clinical and neuroimaging features in a Brazilian family. *Arq Neuropsiquiatr*. 2011;69(2B):288-91.
99. Pablo LE, Garcia-Martin E, Gazulla J, Larrosa JM, Ferreras A, Santorelli FM, et al. Retinal nerve fiber hypertrophy in ataxia of Charlevoix-Saguenay patients. *Mol Vis*. 2011;17:1871-6.
100. Narayanan V, Rice SG, Olfers SS, Sivakumar K. Autosomal recessive spastic ataxia of Charlevoix-Saguenay: compound heterozygotes for nonsense mutations of the SACS gene. *J Child Neurol*. 2011;26(12):1585-9.
101. Kozlov G, Denisov AY, Girard M, Dicaire MJ, Hamlin J, McPherson PS, et al. Structural basis of defects in the saccin HEPN domain responsible for autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS). *J Biol Chem*. 2011;286(23):20407-12.
102. H'Mida-Ben Brahim D, M'Zahem A, Assoum M, Bouhlal Y, Fattori F, Anheim M, et al. Molecular diagnosis of known recessive ataxias by homozygosity mapping with SNP arrays. *J Neurol*. 2011;258(1):56-67.
103. Gazulla J, Vela AC, Marin MA, Pablo L, Santorelli FM, Benavente I, et al. Is the ataxia of Charlevoix-Saguenay a developmental disease? *Med Hypotheses*. 2011;77(3):347-52.
104. Desserre J, Devos D, Sautiere BG, Debruyne P, Santorelli FM, Vuillaume I, et al. Thickening of peripapillar retinal fibers for the diagnosis of autosomal recessive spastic ataxia of Charlevoix-Saguenay. *Cerebellum*. 2011;10(4):758-62.
105. Bouhlal Y, Jennewein DM, Anderson B, Reynoldson J, Maamouri W, Hentati F, et al. Computational analysis of a novel SACS gene mutation with BioExtract server. *J Mol Neurosci*. 2011;44(1):53-8.
106. Bouhlal Y, Amouri R, El Euch-Fayeche G, Hentati F. Autosomal recessive spastic ataxia of Charlevoix-Saguenay: an overview. *Parkinsonism Relat Disord*. 2011;17(6):418-22.
107. Anheim M. [Autosomal recessive cerebellar ataxias]. *Rev Neurol (Paris)*. 2011;167(5):372-84.
108. Anesi L, de Gemmis P, Pandolfo M, Hladnik U. Two novel homozygous SACS mutations in unrelated patients including the first reported case of paternal UPD as an etiologic cause of ARSACS. *J Mol Neurosci*. 2011;43(3):346-9.
109. Anderson JF, Siller E, Barral JM. The neurodegenerative-disease-related protein saccin is a molecular chaperone. *J Mol Biol*. 2011;411(4):870-80.
110. Takiyama Y, Ishiura H, Shimazaki H, Namekawa M, Takahashi Y, Goto J, et al. [Japan spastic paraplegia research consortium (JASPAC)]. *Rinsho Shinkeigaku*. 2010;50(11):931-4.
111. Guernsey DL, Dube MP, Jiang H, Asselin G, Blowers S, Evans S, et al. Novel mutations in the saccin gene in ataxia patients from Maritime Canada. *J Neurol Sci*. 2010;288(1-2):79-87.
112. Gerwig M, Kruger S, Kreuz FR, Kreis S, Gizewski ER, Timmann D. Characteristic MRI and funduscopic findings help diagnose ARSACS outside Quebec. *Neurology*. 2010;75(23):2133.
113. Baets J, Deconinck T, Smets K, Goossens D, Van den Bergh P, Dahan K, et al. Mutations in SACS cause atypical and late-onset forms of ARSACS. *Neurology*. 2010;75(13):1181-8.

114. Anheim M, Fleury M, Monga B, Laugel V, Chaigne D, Rodier G, et al. Epidemiological, clinical, paraclinical and molecular study of a cohort of 102 patients affected with autosomal recessive progressive cerebellar ataxia from Alsace, Eastern France: implications for clinical management. *Neurogenetics*. 2010;11(1):1-12.
115. Anderson JF, Siller E, Barral JM. The saccin repeating region (SRR): a novel Hsp90-related supra-domain associated with neurodegeneration. *J Mol Biol*. 2010;400(4):665-74.
116. Terracciano A, Casali C, Grieco GS, Orteschi D, Di Giandomenico S, Seminara L, et al. An inherited large-scale rearrangement in SACS associated with spastic ataxia and hearing loss. *Neurogenetics*. 2009;10(2):151-5.
117. Parfitt DA, Michael GJ, Vermeulen EG, Prodromou NV, Webb TR, Gallo JM, et al. The ataxia protein saccin is a functional co-chaperone that protects against polyglutamine-expanded ataxin-1. *Hum Mol Genet*. 2009;18(9):1556-65.
118. McMillan HJ, Carter MT, Jacob PJ, Laffan EE, O'Connor MD, Boycott KM. Homozygous contiguous gene deletion of 13q12 causing LGMD2C and ARSACS in the same patient. *Muscle Nerve*. 2009;39(3):396-9.
119. Bouhlal Y, El Euch-Fayeche G, Hentati F, Amouri R. A novel SACS gene mutation in a Tunisian family. *J Mol Neurosci*. 2009;39(3):333-6.
120. Vermeer S, Meijer RP, Pijl BJ, Timmermans J, Cruysberg JR, Bos MM, et al. ARSACS in the Dutch population: a frequent cause of early-onset cerebellar ataxia. *Neurogenetics*. 2008;9(3):207-14.
121. Ouyang Y, Segers K, Bouquiaux O, Wang FC, Janin N, Andris C, et al. Novel SACS mutation in a Belgian family with saccin-related ataxia. *J Neurol Sci*. 2008;264(1-2):73-6.
122. Kamada S, Okawa S, Imota T, Sugawara M, Toyoshima I. Autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS): novel compound heterozygous mutations in the SACS gene. *J Neurol*. 2008;255(6):803-6.
123. Garcia A, Criscuolo C, de Michele G, Berciano J. Neurophysiological study in a Spanish family with recessive spastic ataxia of Charlevoix-Saguenay. *Muscle Nerve*. 2008;37(1):107-10.
124. Dupre N, Chrestian N, Thiffault I, Brais B, Rouleau GA, Bouchard JP. [Hereditary ataxias, spastic parapareses and neuropathies in Eastern Canada]. *Rev Neurol (Paris)*. 2008;164(1):12-21.
125. Breckpot J, Takiyama Y, Thienpont B, Van Vooren S, Vermeesch JR, Ortibus E, et al. A novel genomic disorder: a deletion of the SACS gene leading to spastic ataxia of Charlevoix-Saguenay. *Eur J Hum Genet*. 2008;16(9):1050-4.
126. Bouhlal Y, Zouari M, Kefi M, Ben Hamida C, Hentati F, Amouri R. Autosomal recessive ataxia caused by three distinct gene defects in a single consanguineous family. *J Neurogenet*. 2008;22(2):139-48.
127. Anheim M, Chaigne D, Fleury M, Santorelli FM, De Seze J, Durr A, et al. [Autosomal recessive spastic ataxia of Charlevoix-Saguenay: study of a family and review of the literature]. *Rev Neurol (Paris)*. 2008;164(4):363-8.
128. Takiyama Y. Saccinopathies: saccin-related ataxia. *Cerebellum*. 2007;6(4):353-9.
129. Shimazaki H, Sakoe K, Nijima K, Nakano I, Takiyama Y. An unusual case of a spasticity-lacking phenotype with a novel SACS mutation. *J Neurol Sci*. 2007;255(1-2):87-9.
130. Martin MH, Bouchard JP, Sylvain M, St-Onge O, Truchon S. Autosomal recessive spastic ataxia of Charlevoix-Saguenay: a report of MR imaging in 5 patients. *AJNR Am J Neuroradiol*. 2007;28(8):1606-8.
131. Hara K, Shimbo J, Nozaki H, Kikugawa K, Onodera O, Nishizawa M. Saccin-related ataxia with neither retinal hypermyelination nor spasticity. *Mov Disord*. 2007;22(9):1362-3.

132. Yamamoto Y, Nakamori M, Konaka K, Nagano S, Shimazaki H, Takiyama Y, et al. Sacsin-related ataxia caused by the novel nonsense mutation Arg4325X. *J Neurol*. 2006;253(10):1372-3.
133. Takiyama Y. Autosomal recessive spastic ataxia of Charlevoix-Saguenay. *Neuropathology*. 2006;26(4):368-75.
134. Ouyang Y, Takiyama Y, Sakoe K, Shimazaki H, Ogawa T, Nagano S, et al. Sacsin-related ataxia (ARSACS): expanding the genotype upstream from the gigantic exon. *Neurology*. 2006;66(7):1103-4.
135. Okawa S, Sugawara M, Watanabe S, Imota T, Toyoshima I. A novel saccin mutation in a Japanese woman showing clinical uniformity of autosomal recessive spastic ataxia of Charlevoix-Saguenay. *J Neurol Neurosurg Psychiatry*. 2006;77(2):280-2.
136. Dupre N, Bouchard JP, Brais B, Rouleau GA. Hereditary ataxia, spastic paraparesis and neuropathy in the French-Canadian population. *Can J Neurol Sci*. 2006;33(2):149-57.
137. Yamamoto Y, Hiraoka K, Araki M, Nagano S, Shimazaki H, Takiyama Y, et al. Novel compound heterozygous mutations in saccin-related ataxia. *J Neurol Sci*. 2005;239(1):101-4.
138. Shimazaki H, Takiyama Y, Sakoe K, Ando Y, Nakano I. A phenotype without spasticity in saccin-related ataxia. *Neurology*. 2005;64(12):2129-31.
139. Hara K, Onodera O, Endo M, Kondo H, Shiota H, Miki K, et al. Saccin-related autosomal recessive ataxia without prominent retinal myelinated fibers in Japan. *Mov Disord*. 2005;20(3):380-2.
140. Criscuolo C, Sacca F, De Michele G, Mancini P, Combarros O, Infante J, et al. Novel mutation of SACS gene in a Spanish family with autosomal recessive spastic ataxia. *Mov Disord*. 2005;20(10):1358-61.
141. Richter AM, Ozgul RK, Poisson VC, Topaloglu H. Private SACS mutations in autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS) families from Turkey. *Neurogenetics*. 2004;5(3):165-70.
142. Ogawa T, Takiyama Y, Sakoe K, Mori K, Namekawa M, Shimazaki H, et al. Identification of a SACS gene missense mutation in ARSACS. *Neurology*. 2004;62(1):107-9.
143. Grieco GS, Malandrini A, Comanducci G, Leuzzi V, Valoppi M, Tessa A, et al. Novel SACS mutations in autosomal recessive spastic ataxia of Charlevoix-Saguenay type. *Neurology*. 2004;62(1):103-6.
144. Gomez CM. ARSACS goes global. *Neurology*. 2004;62(1):10-1.
145. Gagnon C, Desrosiers J, Mathieu J. Autosomal recessive spastic ataxia of Charlevoix-Saguenay: upper extremity aptitudes, functional independence and social participation. *Int J Rehabil Res*. 2004;27(3):253-6.
146. Criscuolo C, Banfi S, Orio M, Gasparini P, Monticelli A, Scarano V, et al. A novel mutation in SACS gene in a family from southern Italy. *Neurology*. 2004;62(1):100-2.
147. El Euch-Fayache G, Lalani I, Amouri R, Turki I, Ouahchi K, Hung WY, et al. Phenotypic features and genetic findings in saccin-related autosomal recessive ataxia in Tunisia. *Arch Neurol*. 2003;60(7):982-8.
148. Tallaksen CM, Durr A, Brice A. Recent advances in hereditary spastic paraplegia. *Curr Opin Neurol*. 2001;14(4):457-63.
149. Mercier J, Prevost C, Engert JC, Bouchard JP, Mathieu J, Richter A. Rapid detection of the saccin mutations causing autosomal recessive spastic ataxia of Charlevoix-Saguenay. *Genet Test*. 2001;5(3):255-9.

150. Mrissa N, Belal S, Hamida CB, Amouri R, Turki I, Mrissa R, et al. Linkage to chromosome 13q11-12 of an autosomal recessive cerebellar ataxia in a Tunisian family. *Neurology*. 2000;54(7):1408-14.
151. Engert JC, Berube P, Mercier J, Dore C, Lepage P, Ge B, et al. ARSACS, a spastic ataxia common in northeastern Quebec, is caused by mutations in a new gene encoding an 11.5-kb ORF. *Nat Genet*. 2000;24(2):120-5.
152. Richter A, Rioux JD, Bouchard JP, Mercier J, Mathieu J, Ge B, et al. Location score and haplotype analyses of the locus for autosomal recessive spastic ataxia of Charlevoix-Saguenay, in chromosome region 13q11. *Am J Hum Genet*. 1999;64(3):768-75.
153. Engert JC, Dore C, Mercier J, Ge B, Betard C, Rioux JD, et al. Autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS): high-resolution physical and transcript map of the candidate region in chromosome region 13q11. *Genomics*. 1999;62(2):156-64.
154. Bouchard M, Langlois G. Orthopedic management in autosomal recessive spastic ataxia of Charlevoix-Saguenay. *Can J Surg*. 1999;42(6):440-4.
155. Bouchard JP, Richter A, Mathieu J, Brunet D, Hudson TJ, Morgan K, et al. Autosomal recessive spastic ataxia of Charlevoix-Saguenay. *Neuromuscul Disord*. 1998;8(7):474-9.
156. Vermeer S, van de Warrenburg BP, Kamsteeg EJ. Arsacs. In: Adam MP, Ardinger HH, Pagon RA, Wallace SE, Bean LJH, Stephens K, et al., editors. *GeneReviews*(R). Seattle (WA)1993.
157. Richter A, Morgan K, Bouchard JP, Poirier J, Mercier J, Gosselin F, et al. Clinical and molecular genetic studies on autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS). *Adv Neurol*. 1993;61:97-103.
158. De Braekeleer M, Giasson F, Mathieu J, Roy M, Bouchard JP, Morgan K. Genetic epidemiology of autosomal recessive spastic ataxia of Charlevoix-Saguenay in northeastern Quebec. *Genet Epidemiol*. 1993;10(1):17-25.
159. Richards C, Bouchard JP, Bouchard R, Barbeau H. A preliminary study of dynamic muscle function in hereditary ataxia. *Can J Neurol Sci*. 1980;7(4):367-77.
160. Langelier R, Bouchard JP, Bouchard R. Computed tomography of posterior fossa in hereditary ataxias. *Can J Neurol Sci*. 1979;6(2):195-8.
161. Bouchard RW, Bouchard JP, Bouchard R, Barbeau A. Electroencephalographic findings in Friedreich's ataxia and autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS). *Can J Neurol Sci*. 1979;6(2):191-4.
162. Bouchard JP, Barbeau A, Bouchard R, Bouchard RW. Electromyography and nerve conduction studies in Friedreich's ataxia and autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS). *Can J Neurol Sci*. 1979;6(2):185-9.
163. Bouchard JP, Barbeau A, Bouchard R, Bouchard RW. Autosomal recessive spastic ataxia of Charlevoix-Saguenay. *Can J Neurol Sci*. 1978;5(1):61-9.