Supplementary Table S1. Detailed clinical findings in ARCAs muscle biopsies.

Disorder	Patients (N - age)	Findings	Reference
Degenerative ataxias			
MSS	2 - 4 to 13 years old	– Generalized muscle wasting – Severe hypotonia – Reduction in number and size of fibers – Fatty replacement	[1]
MSS	1 - 11 years old	– Generalized muscle weakness – Hypotonia – Variation in fibers size – Fatty replacement – Vacuolar degeneration with rimmed vacuoles – Myeloid bodies and vacuoles beneath the sarcolemma or near the nucleus	[2]
MSS	6 - 9 months to 14 years old	– Hypotonia – Atrophic muscle fibers – Variation in fibers size – Predominance of type 1 or type 2 fibres – Nuclear inclusions – Vacuolar degeneration	[3]
MSS	4 - unspecified age	– Increased abnormally enlarged number of lysosomes, containing whorled lamellar or amorphous inclusion bodies	[4]
MSS	4 - 10 to 30 years old	– Hypotonia – Variation in fibers size – Fatty replacement – Autophagic vacuoles, myeloid bodies in subsarcolemmal space	[5]
MSS	6 - 1.5 to 32 years old	– Degeneration in muscle fibers – Variation in fibers size – Fatty replacement – Internalised nuclei – Abnormal mitochondria	[6]
MSS	3 - 2 to 6 years old	– Variation in fibers size – Dense membrane surrounding the nuclei – Small vacuoles	[7]
MSS	12 - 1 to 36 years old	– Variation in fibers size – Autophagic vacuoles and rimmed vacuoles – Nuclear changes	[8]
MSS	1 - 33 months old	– Mild variation in fibers size – Type 2B fibers deficiency – Increase in fibers tissue – Rimmed vacuole	[9]
MSS	3 - 26 to 31 years old	– Generalized muscle atrophy and weakness – Marked variation in fibers size – Predominance of type 1 fibers	[10]

		– Deficiency of type 2B fibers – Fatty replacement – Rimmed vacuoles	
MSS	7 - 2 to 36 years old	– Variation in fibers size – Predominance of type 1 fibers – Internalised nuclei – Fatty replacement – Autophagic vacuoles with myeloid body formation – Massive necrosis with autophagic phenomenon	[11]
MSS	7 - 16 to 41 years old	– Loss of muscle fibers – Variation in fibers size – Fatty replacement – Internalised nuclei – Membrane-bound lipid vacuoles – Abnormal mitochondria	[12]
MSS	1 - 2 years old	– Variation in fibers size – Increased type 2C fibers – Intermyofibrillar collections of membranous whorls and glycogen particles	[13]
MSS	8 - 1 to 38 years old	– Nuclei degeneration – Membranous structures surrounding nuclei	[14]
MSS	2 - 5 and 6.5 years old	– Variation in fibers size – Rare necrotic and regenerating fibers – Membranous structures surrounding the nuclei – Rimmed vacuoles	[15]
MSS	2 - unspecified age	– Membranous autophagic/mitophagic material – Vacuolar structures contained granular or membranous electron-dense material – Aberrant, often widened endoplasmic reticulum structures	[16]
MSS	4 - unspecified age	– Normal variation of fibers size – Fibers necrosis – Defect of mitochondria	[17]
MSS	9 - 22 to 58 years old	– Hypotonia – Fatty and connective tissue replacement	[18]
MSS	4 - unspecified age	– Variation in fibers size – Atrophic fibers – Fatty replacement – Autophagic vacuoles associated with degenerating myonuclei – Internalised nuclei surrounded by membranous structures	[19]
MSS	3	– Atrophic fibers	[20]

	- 23 to 31 years old	– Degenerating nuclei surrounded by membranous structures – Autophagic vacuoles – Glycogen accumulation	
FRDA	4 - unspecified age	– Type 2 fibers atrophy – Fibers type grouping – Reduced MSA in myofibers – Fatty replacement – Aberrant mitochondria	[21]
FRDA	7 - unspecified age	– Type 2 fibers atrophy – Fibers type grouping – Reduced MSA in fibers – Fatty replacement – Aberrant mitochondria	[22]
ARCA-1	1 - unspecified age	– Variation in fibers size – Fatty and fibrous tissue replacement – Enlarged perinuclear space	[23]
SCAR2	1 - 4 years old	– Atrophic fibers – Internalised nuclei	[24]
Cerebellar ataxia with CoQ10 deficiency	13 - 6 to 35 years old	– No histological alterations	[25]
MIRAS	29 - Mean of 52 years old	– Mitochondrial dysfunctions	[26]
MIRAS	1 - 45 years old	– COX deficiency – Ragged-red fibres	[27]
MIRAS	1 - 52 years old	– COX deficiency	[28]
MIRAS	1 - 66 years old	– Increased subsarcolemmal mitochondrial proliferation – Ragged-red fibers	[29]
SPG7	1 - unspecified age	– No histological alterations	[30]
SPG7	1 - unspecified age	– Increased fat in vacuoles	[31]
SPG7	1 - 54 years old	– No histological alterations	[32]
SPG7	1 - 51 years old	– Increased type 2C fibers – COX defient fibers	[33]
SPG7	1 - 70 years old	– Variability in fiber size – Type 2 fibers atrophy – COX deficient fibers	[34]

SPG7	2 - Unspecified age	– Ragged red fibers – COX deficient fibers	[35]
SPG7	1 - Unspecified age	– Ragged red fibers – COX deficient fibers	[36]
Ataxias with impaired DNA repair mechanism			
A-T		**ND**	
AOA1	1 - 67 years old	– Variation in fibers size	[37]
AOA1	1 - 8 years old	– No alterations	[38]
AOA2	1 - 37 years old	– No histological alterations	[39]
Congenital ataxias			
Cayman ataxia		**ND**	
Joubert Syndrome	1 - 44 months	– No histological alterations – Decreased pyruvate oxidation rates – Decreased ATP production	[40]
Joubert Syndrome	2 - 12 and 15 years old	– Fatty and fibrous tissue replacement	[41]
Metabolic ataxias			
AVED	1 - 14.5 years old	– Myofibrillar derangement – Mild, non-specific cytoarchitectural abnormalities – Mild myopathic changes	[42]
AVED	1 - 69 years old	– Endomysial mononuclear inflammatory infiltrates – Rimmed vacuoles;	[43]
Wilson's disease	1 - 10 years old	– No histological alterations	[44]
Refsum's disease		**ND**	

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