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Friedreich ataxia: the clinical picture

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350	Fetal reversed constrictive effect of indomethacin and postnatal delayed closure of the ductus arteriosus following administration of transplacental magnesium sulfate in rats. 2009 , 96, 125-31		7
349	Diverse effects in Friedreich's ataxia place PGC-1alpha center-stage. 2009 , 76, 345-7		1
348	Iron-binding activity in yeast frataxin entails a trade off with stability in the alpha1/beta1 acidic ridge region. 2010 , 426, 197-203		31
347	Update on the genetics of movement disorders. <i>CONTINUUM Lifelong Learning in Neurology</i> , 2010 , 16, 77-95	3	
346	Current Opinion in Neurology. Current world literature. 2010 , 23, 433-44		
345	Graves' disease in a patient with Friedreich's ataxia and diabetes mellitus. 2010 , 22, 536-8		2
344	Towards a unifying, systems biology understanding of large-scale cellular death and destruction caused by poorly liganded iron: Parkinson's, Huntington's, Alzheimer's, prions, bactericides, chemical toxicology and others as examples. 2010 , 84, 825-89		292
343	Long intronic GAA repeats causing Friedreich ataxia impede transcription elongation. 2010 , 2, 120-9		92
342	A high throughput electrochemiluminescence assay for the quantification of frataxin protein levels. 2010 , 659, 129-32		29
341	Whole-body vibration alters blood flow velocity and neuromuscular activity in Friedreich's ataxia. 2011 , 31, 139-44		16
340	Understanding the molecular mechanisms of Friedreich's ataxia to develop therapeutic approaches. <i>Human Molecular Genetics</i> , 2010 , 19, R103-10	5.6	107
339	Altered lipid metabolism in a Drosophila model of Friedreich's ataxia. <i>Human Molecular Genetics</i> , 2010 , 19, 2828-40	5.6	72
338	Novel mutations in the sarsin gene in ataxia patients from Maritime Canada. 2010 , 288, 79-87		16
337	Uncommon features in Cuban families affected with Friedreich ataxia. 2010 , 472, 85-9		8
336	Tumor necrosis factor-alpha polymorphisms and expression in Guillain-Barré syndrome. 2010 , 71, 905-10		27

335	Intermediate-dose idebenone and quality of life in Friedreich ataxia. 2010 , 42, 338-42	16
334	Friedreich ataxia: molecular mechanisms, redox considerations, and therapeutic opportunities. 2010 , 13, 651-90	140
333	Frataxin and mitochondrial FeS cluster biogenesis. <i>Journal of Biological Chemistry</i> , 2010 , 285, 26737-26744	119
332	Iron redistribution as a therapeutic strategy for treating diseases of localized iron accumulation. 2010 , 88, 187-96	40
331	Spinocerebellar degenerations. 2011 , 100, 113-40	18
330	[Autosomal recessive cerebellar ataxias]. 2011 , 167, 372-84	6
329	Friedreich's ataxia: past, present and future. 2011 , 67, 311-30	89
328	What makes a prognostic biomarker in CNS diseases: strategies for targeted biomarker discovery? Part 1: acute and monophasic diseases. 2011 , 5, 333-46	8
327	Impaired inhibition of prepotent motor tendencies in Friedreich ataxia demonstrated by the Simon interference task. 2011 , 76, 140-5	20
326	Constructing and deconstructing stem cell models of neurological disease. 2011 , 70, 626-44	124
325	The Fitts task reveals impairments in planning and online control of movement in Friedreich ataxia: reduced cerebellar-cortico connectivity?. 2011 , 192, 382-90	26
324	Síndrome de Guillain-Barré associada temporal com a vacina influenza A. 2011 , 29, 685-688	3
323	Hereditary myelopathies. <i>CONTINUUM Lifelong Learning in Neurology</i> , 2011 , 17, 800-15	3 3
322	The peripheral cerebrospinal fluid outflow pathway [physiology and pathophysiology of CSF recirculation: A review and hypothesis. 2011 , 17, 51-66	20
321	ADEM as a niche variant of post infectious neurological syndromes. 2011 , 236, 123	
320	Autosomal recessive cerebellar ataxias: the current state of affairs. 2011 , 48, 651-9	34
319	The cerebellar cognitive profile. 2011 , 134, 3672-86	187
318	Ethical dilemmas in genetic testing: examples from the Cuban program for predictive diagnosis of hereditary ataxias. 2011 , 20, 241-8	9

317	Generation of induced pluripotent stem cell lines from Friedreich ataxia patients. 2011 , 7, 703-13		84
316	Superior cerebellar peduncle atrophy in Friedreich's ataxia correlates with disease symptoms. <i>Cerebellum</i> , 2011 , 10, 81-7	4.3	58
315	Utilisation of advance motor information is impaired in Friedreich ataxia. <i>Cerebellum</i> , 2011 , 10, 793-803	4.3	17
314	Multiple cranial neuropathy variant of Guillain-Barré syndrome: a case series. 2011 , 44, 252-7		8
313	Antioxidants halt axonal degeneration in a mouse model of X-adrenoleukodystrophy. 2011 , 70, 84-92		107
312	Silencing of frataxin gene expression triggers p53-dependent apoptosis in human neuron-like cells. <i>Human Molecular Genetics</i> , 2011 , 20, 2807-22	5.6	46
311	Preventing the ubiquitin-proteasome-dependent degradation of frataxin, the protein defective in Friedreich's ataxia. <i>Human Molecular Genetics</i> , 2011 , 20, 1253-61	5.6	44
310	Office assessment of gait and station. 2011 , 31, 78-84		2
309	Ultrasound-guided nerve blocks in the Charcot-Marie-Tooth disease and Friedreich's ataxia. 2012 , 108, 1042-3		9
308	Interferon gamma upregulates frataxin and corrects the functional deficits in a Friedreich ataxia model. <i>Human Molecular Genetics</i> , 2012 , 21, 2855-61	5.6	46
307	The heart in Friedreich ataxia: definition of cardiomyopathy, disease severity, and correlation with neurological symptoms. 2012 , 125, 1626-34		85
306	Clinical features of Friedreich ataxia. 2012 , 27, 1133-7		114
305	Childhood cerebellar ataxia. 2012 , 27, 1138-45		36
304	Coming into view: eukaryotic iron chaperones and intracellular iron delivery. <i>Journal of Biological Chemistry</i> , 2012 , 287, 13518-23	5.4	83
303	Clinical reasoning: a middle-aged woman with progressive symmetric weakness and a CSF pleocytosis. 2012 , 78, e88-92		
302	Friedreich ataxia: new pathways. 2012 , 27, 1204-11		22
301	Clinical monitoring in a patient with Friedreich ataxia and osteogenic sarcoma. 2012 , 27, 1159-63		3
300	Sensory neuropathy and autoimmune diseases. 2012 , 2012, 873587		26

299	Cardiac dysfunction exacerbated by endocrinopathies in Friedreich ataxia: a case series. 2012 , 27, 1316-9	3
298	Biochemical markers of autoimmune diseases of the nervous system. 2012 , 18, 4556-63	7
297	Normal left ventricular ejection fraction and mass but subclinical myocardial dysfunction in patients with Friedreich's ataxia. 2012 , 13, 346-52	28
296	Understanding the genetic and molecular pathogenesis of Friedreich's ataxia through animal and cellular models. 2012 , 5, 165-76	51
295	Atypical parkinsonism and cerebrotendinous xanthomatosis: report of a family with corticobasal syndrome and a literature review. 2012 , 27, 1769-74	23
294	[Miller-Fisher variant of Guillain-Barré syndrome in the Resuscitation Unit]. 2012 , 59, 342-3	0
293	Human adipose stem cell-conditioned medium increases survival of Friedreich's ataxia cells submitted to oxidative stress. 2012 , 21, 2817-26	17
292	Peripheral neuropathy associated with mitochondrial disease in children. 2012 , 54, 407-14	26
291	Chromatin changes in the development and pathology of the Fragile X-associated disorders and Friedreich ataxia. 2012 , 1819, 802-10	15
290	Oxidative stress induces mitochondrial fragmentation in frataxin-deficient cells. 2012 , 418, 336-41	25
289	A functional MRI study of motor dysfunction in Friedreich's ataxia. 2012 , 1471, 138-54	26
288	Recent developments and future directions in Guillain-Barré syndrome. 2012 , 17 Suppl 3, 57-70	31
287	Retrospective study of the effects of inpatient rehabilitation on improving and maintaining functional independence in people with Friedreich ataxia. 2012 , 93, 1860-3	22
286	Uses of the postural stability test for differential diagnosis of hereditary ataxias. 2012 , 316, 79-85	10
285	Inherited and Sporadic Ataxias. 2012 , 279-295	2
284	Human mesenchymal stem cells increase anti-oxidant defences in cells derived from patients with Friedreich's ataxia. <i>Cerebellum</i> , 2012 , 11, 861-71	4-3 19
283	Tandem Repeat Polymorphisms. 2012 ,	7
282	Friedreich Ataxia. 2012 , 891-896	

281	Protein stability and dynamics modulation: the case of human frataxin. <i>PLoS ONE</i> , 2012 , 7, e45743	3.7	18
280	Induced pluripotent stem cells to model and treat neurogenetic disorders. 2012 , 2012, 346053		19
279	Treatment for dysphagia (swallowing difficulties) in hereditary ataxia syndromes. 2012 ,		
278	DNA Methylation and Trinucleotide Repeat Expansion Diseases. 2012 ,		3
277	Kidney infarction in Friedreich's ataxia with dilated cardiomyopathy. 2012 , 2012,		
276	Neurodegeneration in Friedreich's ataxia is associated with a mixed activation pattern of the brain. A fMRI study. 2012 , 33, 1780-91		27
275	Differentiating profiles of speech impairments in Friedreich's ataxia: a perceptual and instrumental approach. 2012 , 47, 65-76		16
274	Altered cerebrospinal fluid index of prealbumin, fibrinogen, and haptoglobin in patients with Guillain-Barré syndrome and chronic inflammatory demyelinating polyneuropathy. 2012 , 125, 129-35		21
273	A case of anti-GA1 antibody-positive Fisher syndrome with elevated tau protein in cerebrospinal fluid. 2012 , 34, 329-32		5
272	The mismatch repair system protects against intergenerational GAA repeat instability in a Friedreich ataxia mouse model. 2012 , 46, 165-71		47
271	Impact of Friedreich's Ataxia on health-care resource utilization in the United Kingdom and Germany. 2013 , 8, 38		5
270	The role of chemokines in Guillain-Barré syndrome. 2013 , 48, 320-30		24
269	Induced pluripotent stem cells from friedreich ataxia patients fail to upregulate frataxin during in vitro differentiation to peripheral sensory neurons. 2013 , 22, 3271-82		42
268	Excessive motor overflow reveals abnormal inter-hemispheric connectivity in Friedreich ataxia. <i>Journal of Neurology</i> , 2013 , 260, 1757-64	5.5	1
267	Neuromuscular disease and anesthesia. 2013 , 48, 451-60		22
266	The impact of reactive oxygen species and genetic mitochondrial mutations in Parkinson's disease. 2013 , 532, 18-23		139
265	Friedreich's ataxia-associated GAA repeats induce replication-fork reversal and unusual molecular junctions. 2013 , 20, 486-94		65
264	Cis-silencing of PIP5K1B evidenced in Friedreich's ataxia patient cells results in cytoskeleton anomalies. <i>Human Molecular Genetics</i> , 2013 , 22, 2894-904	5.6	19

263	Elevated levels of S100B, tau and pNFH in cerebrospinal fluid are correlated with subtypes of Guillain-Barré syndrome. 2013 , 34, 655-61		11
262	Frataxin: a protein in search for a function. 2013 , 126 Suppl 1, 43-52		129
261	14-3-3 proteins, particularly of the epsilon isoform, are detectable in cerebrospinal fluids of cerebellar diseases in children. 2013 , 35, 555-60		2
260	Deferiprone and idebenone rescue frataxin depletion phenotypes in a Drosophila model of Friedreich's ataxia. 2013 , 521, 274-81		34
259	Iron-sulfur cluster synthesis, iron homeostasis and oxidative stress in Friedreich ataxia. 2013 , 55, 50-61		100
258	Metabolic remodeling in frataxin-deficient yeast is mediated by Cth2 and Adr1. 2013 , 1833, 3326-3337		14
257	A dynamic model of the proteins that form the initial iron-sulfur cluster biogenesis machinery in yeast mitochondria. 2013 , 32, 183-96		10
256	Friedreich ataxia: dysarthria profile and clinical data. <i>Cerebellum</i> , 2013 , 12, 475-84	4.3	27
255	Therapeutic strategies in Friedreich's ataxia. 2013 , 1514, 91-7		17
254	Iron metabolism in the CNS: implications for neurodegenerative diseases. <i>Nature Reviews Neuroscience</i> , 2013 , 14, 551-64	13.5	288
253	Mitochondrial pathophysiology in Friedreich's ataxia. 2013 , 126 Suppl 1, 53-64		58
252	Increased prevalence of sleep-disordered breathing in Friedreich ataxia. 2013 , 81, 46-51		15
251	Neurons and cardiomyocytes derived from induced pluripotent stem cells as a model for mitochondrial defects in Friedreich's ataxia. 2013 , 6, 608-21		111
250	Clinical features of Friedreich's ataxia: classical and atypical phenotypes. 2013 , 126 Suppl 1, 103-17		148
249	Treatment of Friedreich's ataxia. 2013 , 1, 221-234		5
248	Animal and cellular models of Friedreich ataxia. 2013 , 126 Suppl 1, 65-79		57
247	Serial cerebrospinal fluid neurofilament heavy chain levels in severe Guillain-Barré syndrome. 2013 , 48, 132-4		8
246	Missense mutations linked to Friedreich ataxia have different but synergistic effects on mitochondrial frataxin isoforms. <i>Journal of Biological Chemistry</i> , 2013 , 288, 4116-27	5.4	19

245	Rehabilitation principles in chronic neurological conditions in adults and children. 131-138		1
244	Rehabilitation of Friedreich ataxia. 185-202		0
243	Endoscopic associated iatrogenic Terson's syndrome. 2013 , 40, 265-6		3
242	Does intrathecal baclofen have a place in the treatment of painful spasms in Friedreich ataxia?. 2013 , 5, 201-3		3
241	Friedreich's Ataxia and Diseases Associated with Expansion of Non-Coding Triplets. 2013 , 227-238		
240	Posterior Spinal Fusion for Friedreich Ataxia-Related Scoliosis in Twin Girls: A Case Report. 2013 , 3, e39		
239	Stem cells from wildtype and Friedreich's ataxia mice present similar neuroprotective properties in dorsal root ganglia cells. <i>PLoS ONE</i> , 2013 , 8, e62807	3.7	10
238	Burden of Friedreich's Ataxia to the Patients and Healthcare Systems in the United States and Canada. <i>Frontiers in Pharmacology</i> , 2013 , 4, 66	5.6	5
237	Neurodegeneration in Friedreich's ataxia: from defective frataxin to oxidative stress. 2013 , 2013, 487534		44
236	. 2013 ,		4
235	Ataxia. 204-228		
234	Human Pluripotent Stem Cells Modeling Neurodegenerative Diseases. 2013 ,		2
233	Functional characterization of Friedreich ataxia iPS-derived neuronal progenitors and their integration in the adult brain. <i>PLoS ONE</i> , 2014 , 9, e101718	3.7	20
232	Mitochondrial iron-sulfur cluster dysfunction in neurodegenerative disease. <i>Frontiers in Pharmacology</i> , 2014 , 5, 29	5.6	62
231	Friedreich's Ataxia: A Neuronal Point of View on the Oxidative Stress Hypothesis. 2014 , 3, 592-603		8
230	18. Iron-sulfur proteins and human diseases.		
229	Very late-onset Friedreich ataxia with laryngeal dystonia. 2014 , 6, 287-90		2
228	Sensitivity of spatiotemporal gait parameters in measuring disease severity in Friedreich ataxia. <i>Cerebellum</i> , 2014 , 13, 677-88	4.3	22

227	Consensus clinical management guidelines for Friedreich ataxia. 2014 , 9, 184		56
226	CANVAS an update: clinical presentation, investigation and management. 2014 , 24, 465-74		46
225	Methylene blue rescues heart defects in a Drosophila model of Friedreich's ataxia. <i>Human Molecular Genetics</i> , 2014 , 23, 968-79	5.6	27
224	Dysregulation of cellular iron metabolism in Friedreich ataxia: from primary iron-sulfur cluster deficit to mitochondrial iron accumulation. <i>Frontiers in Pharmacology</i> , 2014 , 5, 130	5.6	99
223	Cerebellar pathology in Friedreich's ataxia: atrophied dentate nuclei with normal iron content. 2014 , 6, 93-9		44
222	Fixing frataxin: 'ironing out' the metabolic defect in Friedreich's ataxia. 2014 , 171, 2174-90		32
221	Longitudinal strain in Friedreich Ataxia: a potential marker for early left ventricular dysfunction. 2014 , 31, 50-7		17
220	Optical coherence tomography and visual field findings in patients with Friedreich ataxia. 2014 , 34, 118-21		15
219	Increased cerebrospinal fluid protein and motor conduction studies as prognostic markers of outcome and nerve ultrasound changes in Guillain-Barré syndrome. 2014 , 340, 37-43		18
218	Nicotinamide in Friedreich's ataxia: useful or not?. 2014 , 384, 474-5		4
217	Frataxin deficiency in neonatal rat ventricular myocytes targets mitochondria and lipid metabolism. 2014 , 73, 21-33		27
216	Cognitive deficits in Friedreich ataxia correlate with micro-structural changes in dentatorubral tract. <i>Cerebellum</i> , 2014 , 13, 187-98	4.3	30
215	Dysphagia and swallowing-related quality of life in Friedreich ataxia. <i>Journal of Neurology</i> , 2014 , 261, 392-9	5.5	17
214	Evaluating the status of antiganglioside antibodies in children with Guillain-Barré syndrome. 2014 , 21, 64-8		3
213	Diagnosis of Guillain-Barré syndrome and validation of Brighton criteria. 2014 , 137, 33-43		396
212	Deferiprone in Friedreich ataxia: a 6-month randomized controlled trial. 2014 , 76, 509-21		83
211	Low bone mineral density in Friedreich ataxia. <i>Cerebellum</i> , 2014 , 13, 549-57	4.3	10
210	Friedreich's ataxia and other hereditary ataxias in Greece: an 18-year perspective. 2014 , 336, 87-92		7

209	Myelin paucity of the superior cerebellar peduncle in individuals with Friedreich ataxia: an MRI magnetization transfer imaging study. 2014 , 343, 138-43		11
208	Brain iron homeostasis: from molecular mechanisms to clinical significance and therapeutic opportunities. 2014 , 20, 1324-63		102
207	The effect of hip and knee joint center calibration method on musculo-skeletal modeling outcomes. 2015 , 42, S46-S47		
206	Ultra-structural hair alterations in Friedreich's ataxia: A scanning electron microscopic investigation. 2015 , 78, 731-6		
205	Friedreich ataxia in Norway - an epidemiological, molecular and clinical study. 2015 , 10, 108		9
204	Friedreich Ataxia: From the Eye of a Molecular Biologist. 2015 , 20, 51-5		7
203	Iron function and dysfunction in the brain: A pediatric neurologist's perspective. 2015 , 02, 003-014		
202	Methylene Blue Partially Rescues Heart Defects in a Drosophila Model of Huntington's Disease. 2015 , 4, 173-86		5
201	Cerebellar transcriptional alterations with Purkinje cell dysfunction and loss in mice lacking PGC-1 β <i>Frontiers in Cellular Neuroscience</i> , 2014 , 8, 441	6.1	31
200	Biomarkers of Guillain-Barré Syndrome: Some Recent Progress, More Still to Be Explored. 2015 , 2015, 564098		25
199	A new cellular model to follow Friedreich's ataxia development in a time-resolved way. 2015 , 8, 711-9		13
198	Treatment for dysphagia (swallowing difficulties) in hereditary ataxia. 2015 , CD010169		15
197	A novel GAA-repeat-expansion-based mouse model of Friedreich's ataxia. 2015 , 8, 225-35		32
196	Functional and gait assessment in children with Friedreich ataxia: Comparison of quantitative and functional evaluation. 2015 , 42, S45-S46		
195	Dysarthrieprofile von Patienten mit Friedreich Ataxie und spinocerebellären Ataxien vom Typ 3 und Typ 6. 2015 , 39, 187-191		1
194	Stem Cells in Modeling Human Genetic Diseases. <i>Pancreatic Islet Biology</i> , 2015 ,		0.4
193	Unveiling a common mechanism of apoptosis in β cells and neurons in Friedreich's ataxia. <i>Human Molecular Genetics</i> , 2015 , 24, 2274-86	5.6	47
192	Milestones in Friedreich ataxia: more than a century and still learning. 2015 , 16, 151-60		36

191	Cerebrospinal Fluid in Clinical Neurology. 2015 ,	13
190	Frataxin inactivation leads to steroid deficiency in flies and human ovarian cells. <i>Human Molecular Genetics</i> , 2015 , 24, 2615-26	5.6 22
189	Genome-Engineering Tools to Establish Accurate Reporter Cell Lines That Enable Identification of Therapeutic Strategies to Treat Friedreich's Ataxia. 2015 , 20, 760-7	
188	Nasality in Friedreich ataxia. 2015 , 29, 46-58	10
187	A study of up to 12 years of follow-up of Friedreich ataxia utilising four measurement tools. 2015 , 86, 660-6	17
186	Friedreich Ataxia. 2015 , 833-843	1
185	Targeting lipid peroxidation and mitochondrial imbalance in Friedreich's ataxia. 2015 , 99, 344-50	48
184	A longitudinal study of the Friedreich Ataxia Impact Scale. 2015 , 352, 53-7	9
183	Induced Pluripotent Stem Cells (iPSCs) to Study and Treat Movement Disorders. 2015 , 159-170	
182	Association of ubiquitin carboxy-terminal hydrolase-L1 in cerebrospinal fluid with clinical severity in a cohort of patients with Guillain-Barré syndrome. 2015 , 36, 921-6	1
181	Measuring disease progression in giant axonal neuropathy: implications for clinical trial design. 2015 , 30, 741-8	5
180	Animal Models of Friedreich Ataxia. 2015 , 1017-1024	1
179	"Both Sides of the Wheelchair": The Views of Individuals with, and Parents of Individuals with Friedreich Ataxia Regarding Pre-symptomatic Testing of Minors. 2015 , 24, 732-43	3
178	Friedreich Ataxia. 2015 , 984-1002	
177	Autoantibodies against ganglioside GM3 are associated with narcolepsy-cataplexy developing after Pandemrix vaccination against 2009 pandemic H1N1 type influenza virus. 2015 , 63, 68-75	35
176	Evidence for chromosome fragility at the frataxin locus in Friedreich ataxia. 2015 , 781, 14-21	11
175	Genetics and Clinical Features of Inherited Ataxias. 2015 , 939-978	
174	Orthopedic Management. 2015 , 1053-1071	

173	Ataxia. 2015 , 33, 225-48		46
172	Mesenchymal stem cells improve motor functions and decrease neurodegeneration in ataxic mice. 2015 , 23, 130-8		28
171	Friedreich Ataxia. 2016 , 103-112		
170	Motor Speech Impairments. 2016 , 985-994		1
169	Liver Growth Factor (LGF) Upregulates Frataxin Protein Expression and Reduces Oxidative Stress in Friedreich's Ataxia Transgenic Mice. 2016 , 17,		4
168	Functional and Gait Assessment in Children and Adolescents Affected by Friedreich's Ataxia: A One-Year Longitudinal Study. <i>PLoS ONE</i> , 2016 , 11, e0162463	3-7	19
167	Human Frataxin Folds Via an Intermediate State. Role of the C-Terminal Region. 2016 , 6, 20782		13
166	Membrane Lipids in Presynaptic Function and Disease. 2016 , 90, 11-25		109
165	Time-resolved functional analysis of acute impairment of frataxin expression in an inducible cell model of Friedreich ataxia. 2016 , 5, 654-61		15
164	Deep sequencing of mitochondrial genomes reveals increased mutation load in Friedreich's ataxia. 2016 , 3, 523-36		12
163	Fronto-cerebellar dysfunction and dysconnectivity underlying cognition in friedreich ataxia: The IMAGE-FRDA study. 2016 , 37, 338-50		33
162	Cerebral and cerebellar grey matter atrophy in Friedreich ataxia: the IMAGE-FRDA study. <i>Journal of Neurology</i> , 2016 , 263, 2215-2223	5-5	36
161	Alleviating GAA Repeat Induced Transcriptional Silencing of the Friedreich's Ataxia Gene During Somatic Cell Reprogramming. 2016 , 25, 1788-1800		15
160	Lentivirus-mediated frataxin gene delivery reverses genome instability in Friedreich ataxia patient and mouse model fibroblasts. 2016 , 23, 846-856		10
159	Hereditary and metabolic myelopathies. 2016 , 136, 769-85		12
158	Characterization of frataxin gene network in Friedreich's ataxia fibroblasts using the RNA-Seq technique. 2016 , 30, 59-66		9
157	Degenerative ataxias, from genes to therapies: The 2015 Cotzias Lecture. 2016 , 86, 2284-90		20
156	Effects of genetic severity on glucose homeostasis in Friedreich ataxia. 2016 , 54, 887-894		10

155	Auditory neuropathy in Brown-Vialetto-Van Laere syndrome due to riboflavin transporter RFVT2 deficiency. 2016 , 58, 848-54		15
154	Proposed diagnostic criteria for cerebellar ataxia with neuropathy and vestibular areflexia syndrome (CANVAS). 2016 , 6, 61-68		65
153	Activating frataxin expression by repeat-targeted nucleic acids. 2016 , 7, 10606		55
152	Combined Cerebellar Proton MR Spectroscopy and DWI Study of Patients with Friedreich's Ataxia. <i>Cerebellum</i> , 2017 , 16, 82-88	4-3	10
151	Alternative mitochondrial electron transfer for the treatment of neurodegenerative diseases and cancers: Methylene blue connects the dots. 2017 , 157, 273-291		40
150	Cardiac transplantation in Friedreich Ataxia: Extended follow-up. 2017 , 375, 471-473		10
149	Measuring Inhibition and Cognitive Flexibility in Friedreich Ataxia. <i>Cerebellum</i> , 2017 , 16, 757-763	4-3	13
148	Dentate Update: Imaging Features of Entities That Affect the Dentate Nucleus. 2017 , 38, 1467-1474		29
147	Abnormal Function of Metalloproteins Underlies Most Neurodegenerative Diseases. 2017 , 415-438		1
146	Dysphagia in Friedreich Ataxia. 2017 , 32, 626-635		12
145	Historical Perspectives on Ancient Greek Derived "a" Prefixed Nomenclature for Acquired Neurocognitive Impairment. 2017 , 27, 147-157		1
144	Degenerative Ataxias: challenges in clinical research. 2017 , 4, 53-60		4
143	Deletion of the GAA repeats from the human frataxin gene using the CRISPR-Cas9 system in YG8R-derived cells and mouse models of Friedreich ataxia. 2017 , 24, 265-274		37
142	Transplantation of wild-type mouse hematopoietic stem and progenitor cells ameliorates deficits in a mouse model of Friedreich's ataxia. 2017 , 9,		31
141	Mechanisms of unexpected death and autopsy findings in Friedreich ataxia. 2017 , 57, 192-196		1
140	Combined Central and Peripheral Degenerative Vestibular Disorders: CANVAS, Idiopathic Cerebellar Ataxia with Bilateral Vestibulopathy (CABV) and Other Differential Diagnoses of the CABV Phenotype. 2017 , 5, 167-174		2
139	Circulating miR-323-3p is a biomarker for cardiomyopathy and an indicator of phenotypic variability in Friedreich's ataxia patients. 2017 , 7, 5237		15
138	Friedreich Ataxia. 2017 , 1163-1171		

137	Tumor necrosis factor- α in Guillain-Barré syndrome, friend or foe?. 2017 , 21, 103-112	9
136	hiPSC Disease Modeling of Rare Hereditary Cerebellar Ataxias: Opportunities and Future Challenges. 2017 , 23, 554-566	4
135	Voice in Friedreich Ataxia. 2017 , 31, 243.e9-243.e19	17
134	Neurobehavioral deficits in the KIKO mouse model of Friedreich's ataxia. 2017 , 316, 183-188	13
133	Friedreich Ataxia: Clinical Feature and Electrophysiological Symptoms. 2017 , 8, 691-692	
132	Diabetes Mellitus as the Presenting Feature of Friedreich's Ataxia. 2017 , 8, S117-S119	7
131	10 Iron-sulfur proteins and human diseases. 2017 ,	
130	Early VGLUT1-specific parallel fiber synaptic deficits and dysregulated cerebellar circuit in the KIKO mouse model of Friedreich ataxia. 2017 , 10, 1529-1538	17
129	Randomized, clinical trial of RT001: Early signals of efficacy in Friedreich's ataxia. 2018 , 33, 1000-1005	47
128	Activation of Frataxin Protein Expression by Antisense Oligonucleotides Targeting the Mutant Expanded Repeat. 2018 , 28, 23-33	26
127	Quantitative proteomics in Friedreich's ataxia B-lymphocytes: A valuable approach to decipher the biochemical events responsible for pathogenesis. 2018 , 1864, 997-1009	13
126	Cerebrospinal fluid findings in Guillain-Barré syndrome and chronic inflammatory demyelinating polyneuropathies. 2017 , 146, 125-138	23
125	Biophysical characterisation of the recombinant human frataxin precursor. 2018 , 8, 390-405	5
124	The role of oxidative stress in Friedreich's ataxia. 2018 , 592, 718-727	53
123	Cerebral abnormalities in Friedreich ataxia: A review. 2018 , 84, 394-406	33
122	Large Interruptions of GAA Repeat Expansion Mutations in Friedreich Ataxia Are Very Rare. <i>Frontiers in Cellular Neuroscience</i> , 2018 , 12, 443	6.1 11
121	Adding a temporal dimension to the study of Friedreich's ataxia: the effect of frataxin overexpression in a human cell model. 2018 , 11,	18
120	Rapid and Complete Reversal of Sensory Ataxia by Gene Therapy in a Novel Model of Friedreich Ataxia. 2018 , 26, 1940-1952	58

119	Main inherited neurodegenerative cerebellar ataxias, how to recognize them using magnetic resonance imaging?. 2018 , 45, 265-275		5
118	Cognitive and functional connectivity alterations in Friedreich's ataxia. 2018 , 5, 677-686		15
117	Frataxin Restoration in the Nervous System: Possibilities for Gene Therapy. 2018 , 26, 1880-1882		
116	Identification of CSF biomarkers by proteomics in Guillain-Barré syndrome. 2018 , 15, 5177-5182		4
115	Activating frataxin expression by single-stranded siRNAs targeting the GAA repeat expansion. 2018 , 28, 2850-2855		14
114	Histone Deacetylase Inhibitors: A Therapeutic Key in Neurological Disorders?. 2018 , 77, 855-870		27
113	Test-retest reliability of an instrumented electronic walkway system (GAITRite) for the measurement of spatio-temporal gait parameters in young patients with Friedreich's ataxia. 2018 , 66, 45-50		15
112	Identification of cardioprotective drugs by medium-scale pharmacological screening on a cardiac model of Friedreich's ataxia. 2018 , 11,		7
111	The use of cerebrospinal fluid in biomarker studies. 2017 , 146, 3-20		11
110	High Degree of Genetic Heterogeneity for Hereditary Cerebellar Ataxias in Australia. <i>Cerebellum</i> , 2019 , 18, 137-146	4-3	15
109	Prominent Spasticity and Hyperreflexia of the Legs in a Nepalese Patient with Friedreich Ataxia. 2019 , 58, 2865-2869		
108	Exploring iron-binding to human frataxin and to selected Friedreich ataxia mutants by means of NMR and EPR spectroscopies. 2019 , 1867, 140254		9
107	Efficient electroporation of neuronal cells using synthetic oligonucleotides: identifying duplex RNA and antisense oligonucleotide activators of human frataxin expression. 2019 , 25, 1118-1129		7
106	A Clinician's Approach to Peripheral Neuropathy. 2019 , 39, 519-530		20
105	Potential biomarker identification for Friedreich's ataxia using overlapping gene expression patterns in patient cells and mouse dorsal root ganglion. <i>PLoS ONE</i> , 2019 , 14, e0223209	3-7	3
104	Excision of the expanded GAA repeats corrects cardiomyopathy phenotypes of iPSC-derived Friedreich's ataxia cardiomyocytes. 2019 , 40, 101529		12
103	Application of Quantitative Motor Assessments in Friedreich Ataxia and Evaluation of Their Relation to Clinical Measures. <i>Cerebellum</i> , 2019 , 18, 896-909	4-3	7
102	Solubility of clopidogrel hydrogen sulfate polymorphs in ethyl acetate + 2-butanol mixtures at 283.15B13.15 K. 2019 , 139, 105846		3

101	Scoliosis in Patients With Friedreich Ataxia: Results of a Consecutive Prospective Series. 2019 , 7, 812-821		2
100	SINEUP non-coding RNAs rescue defective frataxin expression and activity in a cellular model of Friedreich's Ataxia. <i>Nucleic Acids Research</i> , 2019 , 47, 10728-10743	20.1	18
99	Advanced Technology for Gene Delivery with Homing Peptides to Spinal Cord through Systemic Circulation in Mice. <i>Molecular Therapy - Methods and Clinical Development</i> , 2019 , 13, 474-483	6.4	0
98	Patient-reported outcomes in Friedreich's ataxia after withdrawal from idebenone. 2019 , 139, 533-539		12
97	Mitochondrial dysfunction in neurodegenerative diseases and the potential countermeasure. 2019 , 25, 816-824		78
96	Early predictors of functional disability in Guillain-Barré Syndrome. 2019 , 119, 555-559		8
95	Randomized, double-blind, placebo-controlled study of interferon- 1b in Friedreich Ataxia. 2019 , 6, 546-553		18
94	A new tool to determine the cellular metabolic landscape: nanotechnology to the study of Friedreich's ataxia. 2019 , 9, 19282		5
93	100 Krankheitsbilder in der Physiotherapie. 2019 ,		
92	Correction of half the cardiomyocytes fully rescue Friedreich ataxia mitochondrial cardiomyopathy through cell-autonomous mechanisms. <i>Human Molecular Genetics</i> , 2019 , 28, 1274-1285	5.6	12
91	Transcriptional profiling of isogenic Friedreich ataxia neurons and effect of an HDAC inhibitor on disease signatures. <i>Journal of Biological Chemistry</i> , 2019 , 294, 1846-1859	5.4	15
90	Progress in understanding Friedreich's ataxia using human induced pluripotent stem cells. 2019 , 7, 81-90		4
89	Developmental and neurodegenerative damage in Friedreich's ataxia. 2019 , 26, 483-489		17
88	It is not your eyes. 2020 , 65, 487-493		
87	Trinucleotide Repeats. 2020 ,		0
86	Cerebellum and cognition in Friedreich ataxia: a voxel-based morphometry and volumetric MRI study. <i>Journal of Neurology</i> , 2020 , 267, 350-358	5.5	10
85	Exploring the Potential of Small Molecule-Based Therapeutic Approaches for Targeting Trinucleotide Repeat Disorders. 2020 , 57, 566-584		11
84	Longitudinal Increases in Cerebral Brain Activation During Working Memory Performance in Friedreich Ataxia: 24-Month Data from IMAGE-FRDA. <i>Cerebellum</i> , 2020 , 19, 182-191	4.3	5

83	Pediatric Neuromuscular Disorders. 2020 , 67, 45-57		1
82	Mitochondrial dysfunction in neurons in Friedreich's ataxia. 2020 , 102, 103419		9
81	Inhibition of the SUV4-20 H1 histone methyltransferase increases frataxin expression in Friedreich's ataxia patient cells. <i>Journal of Biological Chemistry</i> , 2020 , 295, 17973-17985	5-4	3
80	Current Status of microRNA-Based Therapeutic Approaches in Neurodegenerative Disorders. 2020 , 9,		35
79	Glia: Models for Human Neurodevelopmental and Neurodegenerative Disorders. 2020 , 21,		4
78	Central Nervous System Therapeutic Targets in Friedreich Ataxia. 2020 , 31, 1226-1236		12
77	A Drosophila model of Friedreich ataxia with CRISPR/Cas9 insertion of GAA repeats in the frataxin gene reveals in vivo protection by N-acetyl cysteine. <i>Human Molecular Genetics</i> , 2020 , 29, 2831-2844	5.6	3
76	Thioredoxin and Glutaredoxin Systems as Potential Targets for the Development of New Treatments in Friedreich's Ataxia. 2020 , 9,		12
75	Cerebrospinal fluid findings and hyponatremia in COVID-19 patients with altered mental status. 2020 , 13, 63		5
74	Glycolysis and Autoimmune Diseases: A Growing Relationship. 2020 , 14, 91-106		1
73	Hereditary Ataxia: A Focus on Heme Metabolism and Fe-S Cluster Biogenesis. 2020 , 21,		8
72	HMTase Inhibitors as a Potential Epigenetic-Based Therapeutic Approach for Friedreich's Ataxia. 2020 , 11, 584		1
71	Friedreich ataxia. 2020 , 99-112		
70	Sensory neuronopathy as a major clinical feature of mitochondrial trifunctional protein deficiency in adults. 2020 , 176, 380-386		8
69	Oxidative Stress, a Crossroad Between Rare Diseases and Neurodegeneration. 2020 , 9,		20
68	Long-term voluntary running prevents the onset of symptomatic Friedreich's ataxia in mice. 2020 , 10, 6095		3
67	Guillain-Barré syndrome spectrum associated with COVID-19: an up-to-date systematic review of 73 cases. <i>Journal of Neurology</i> , 2021 , 268, 1133-1170	5.5	150
66	The displacement of frataxin from the mitochondrial cristae correlates with abnormal respiratory supercomplexes formation and bioenergetic defects in cells of Friedreich ataxia patients. 2021 , 35, e21362		2

65	Frataxins Emerge as New Players of the Intracellular Antioxidant Machinery. 2021 , 10,		0
64	The responsiveness of gait and balance outcomes to disease progression in Friedreich ataxia.		
63	Defective palmitoylation of transferrin receptor triggers iron overload in Friedreich ataxia fibroblasts. 2021 , 137, 2090-2102		6
62	Results of a randomized double-blind study evaluating luvadaxistat in adults with Friedreich ataxia. 2021 , 8, 1343-1352		2
61	Frataxin deficiency promotes endothelial senescence in pulmonary hypertension. 2021 , 131,		8
60	Friedreich Ataxia: Multidisciplinary Clinical Care. 2021 , 14, 1645-1658		7
59	Neuro-Ophthalmological Findings in Friedreich's Ataxia. 2021 , 11,		2
58	Autosomal recessive adult-onset ataxia. <i>Journal of Neurology</i> , 2021 , 1	5.5	0
57	Characterising the neuropathology and neurobehavioural phenotype in Friedreich ataxia: a systematic review. 2012 , 769, 169-84		6
56	Primary Cultures of Pure Embryonic Dorsal Root Ganglia Sensory Neurons as a New Cellular Model for Friedreich's Ataxia. 2020 , 2056, 241-253		3
55	Frataxin Structure and Function. 2019 , 93, 393-438		8
54	Neuropathology of Ataxias. 2013 , 2327-2347		2
53	CHAPTER 2:Mechanisms of Antisense Oligonucleotides. 2019 , 22-31		1
52	Exenatide induces frataxin expression and improves mitochondrial function in Friedreich ataxia. <i>JCI Insight</i> , 2020 , 5,	9.9	23
51	Hereditary Myelopathies. <i>CONTINUUM Lifelong Learning in Neurology</i> , 2018 , 24, 523-550	3	4
50	Iron Hack - A symposium/hackathon focused on porphyrias, Friedreich's ataxia, and other rare iron-related diseases. <i>F1000Research</i> , 2019 , 8, 1135	3.6	7
49	Novel frataxin isoforms may contribute to the pathological mechanism of Friedreich ataxia. <i>PLoS ONE</i> , 2012 , 7, e47847	3.7	37
48	Friedreich ataxia patient tissues exhibit increased 5-hydroxymethylcytosine modification and decreased CTCF binding at the FXN locus. <i>PLoS ONE</i> , 2013 , 8, e74956	3.7	25

47	Cellular, molecular and functional characterisation of YAC transgenic mouse models of Friedreich ataxia. <i>PLoS ONE</i> , 2014 , 9, e107416	3.7	15
46	Somatic instability of the expanded GAA repeats in Friedreich's ataxia. <i>PLoS ONE</i> , 2017 , 12, e0189990	3.7	32
45	Ocular Involvement in Friedreich Ataxia Patients and its Relationship with Neurological Disability, a Follow-up Study. <i>Diagnostics</i> , 2020 , 10,	3.8	6
44	Demographic and clinical features and rehabilitation outcomes of patients with Friedreich ataxia: A retrospective study. <i>Turkish Journal of Physical Medicine and Rehabilitation</i> , 2018 , 64, 230-238	1.1	2
43	Targeting 3' and 5' untranslated regions with antisense oligonucleotides to stabilize frataxin mRNA and increase protein expression. <i>Nucleic Acids Research</i> , 2021 , 49, 11560-11574	20.1	0
42	Ataxias. 2012 , 3421-3444		
41	CHAPTER 5:Friedreich's Ataxia. <i>2-Oxoglutarate-Dependent Oxygenases</i> , 2013 , 98-117	1.8	
40	iPS Cells and Cardiomyopathies. <i>Pancreatic Islet Biology</i> , 2015 , 83-110	0.4	
39	Friedreich Ataxia. 2016 , 1-9		
38	Transcriptional profiling of isogenic Friedreich ataxia induced pluripotent stem cell-derived neurons.		
37	Neuropathology of Ataxias. 2020 , 1-23		
36	The neural mechanisms of manual dexterity. <i>Nature Reviews Neuroscience</i> , 2021 , 22, 741-757	13.5	6
35	Neurological Disorders in the Lower Extremity. 2020 , 115-144		
34	Cardiomyopathy as the first manifestation of Friedreich's ataxia. <i>Autopsy and Case Reports</i> , 2020 , 10, e2020204	0.6	0
33	Inhibition of the SUV4-20 H1 histone methyltransferase increases frataxin expression in Friedreich's ataxia patient cells.		
32	Molecular and clinical investigation of Iranian patients with Friedreich ataxia. <i>Iranian Biomedical Journal</i> , 2014 , 18, 28-33	2	3
31	The time dimension of neurodegeneration: the example of Friedreich's ataxia. <i>Journal of Neurology and Neuromedicine</i> , 2017 , 2, 31-34	1.5	
30	Nuclear Factor Erythroid-2-Related Factor 2 Signaling in the Neuropathophysiology of Inherited Metabolic Disorders.. <i>Frontiers in Cellular Neuroscience</i> , 2021 , 15, 785057	6.1	2

29	Neuropathology of Ataxias. 2022 , 2615-2637		
28	The Responsiveness of Gait and Balance Outcomes to Disease Progression in Friedreich Ataxia. <i>Cerebellum</i> , 2021 , 1	4.3	0
27	overexpression of frataxin causes toxicity mediated by iron-sulfur cluster deficiency.. <i>Molecular Therapy - Methods and Clinical Development</i> , 2022 , 24, 367-378	6.4	2
26	Selected Histone Deacetylase Inhibitors Reverse the Frataxin Transcriptional Defect in a Novel Friedreich's Ataxia Induced Pluripotent Stem Cell-Derived Neuronal Reporter System.. <i>Frontiers in Neuroscience</i> , 2022 , 16, 836476	5.1	0
25	Mitochondrial De Novo Assembly of IronSulfur Clusters in Mammals: Complex Matters in a Complex That Matters. <i>Inorganics</i> , 2022 , 10, 31	2.9	0
24	Difficulties translating antisense-mediated activation of Frataxin expression from cell culture to mice.. <i>RNA Biology</i> , 2022 , 19, 364-372	4.8	1
23	Modelling Protein Plasticity: The Example of Frataxin and Its Variants.. <i>Molecules</i> , 2022 , 27,	4.8	0
22	Gene therapy for Friedreich ataxia: Too much, too little, or just right?. <i>Molecular Therapy - Methods and Clinical Development</i> , 2022 , 25, 1-2	6.4	
21	Longitudinal Assessment Using Optical Coherence Tomography in Patients with Friedreich's Ataxia.. <i>Tomography</i> , 2021 , 7, 915-931	3.1	2
20	Current Drug Repurposing Strategies for Rare Neurodegenerative Disorders.. <i>Frontiers in Pharmacology</i> , 2021 , 12, 768023	5.6	2
19	Data_Sheet_1.docx. 2020 ,		
18	Table_1.DOCX. 2018 ,		
17	Posttranslational Regulation of Mitochondrial Frataxin and Identification of Compounds that Increase Frataxin Levels in Friedreich's Ataxia.. <i>Journal of Biological Chemistry</i> , 2022 , 101982	5.4	
16	Advantages and Limitations of Gene Therapy and Gene Editing for Friedreich's Ataxia. <i>Frontiers in Genome Editing</i> , 2022 , 4,	2.5	2
15	Metabolomics analysis reveals dysregulation in one carbon metabolism in Friedreich Ataxia. <i>Molecular Genetics and Metabolism</i> , 2022 ,	3.7	0
14	Gene Therapy for Mitochondrial Diseases: Current Status and Future Perspective. <i>Pharmaceutics</i> , 2022 , 14, 1287	6.4	2
13	Premature transcription termination at the expanded GAA repeats and aberrant alternative polyadenylation contributes to the Frataxin transcriptional deficit in Friedreich's ataxia. <i>Human Molecular Genetics</i> ,	5.6	0
12	Friedreich ataxia: clinical features and new developments. <i>Neurodegenerative Disease Management</i> ,	2.8	3

- 11 Frataxin deficiency disrupts mitochondrial respiration and pulmonary endothelial cell function.
- 10 Sensory neuropathies, diagnostic criteria and causes. Publish Ahead of Print, 0
- 9 Perspectives on current models of Friedreich's ataxia. 10, 0
- 8 Beyond Sarcomeric Hypertrophic Cardiomyopathy: How to Diagnose and Manage Phenocopies. 1
- 7 Prediction of the disease course in Friedreich ataxia. **2022**, 12, 1
- 6 Anesthesia Management in Scoliosis Surgery of Patients with Friedreich's Ataxia: A Report of Four Cases. 0
- 5 RNAi in Cell Nuclei: Potential for a new layer of biological regulation and a new strategy for therapeutic discovery. rna.079500.122 0
- 4 Omaveloxolone: an activator of Nrf2 for the treatment of Friedreich ataxia. **2023**, 32, 5-16 0
- 3 Development of PPAR α Agonists for the Treatment of Neuroinflammatory and Neurodegenerative Diseases: Leriglitazone as a Promising Candidate. **2023**, 24, 3201 0
- 2 Insights from yeast: Transcriptional reprogramming following metformin treatment is similar to that of deferiprone in a yeast Friedreich's ataxia model. **2023**, 40, 143-151 0
- 1 Removal of the GAA repeat in the heart of a Friedreich's ataxia mouse model using CjCas9. 0