

CITATION REPORT

List of articles citing

Characterization of human disease phenotypes associated with mutations in TREX1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, ADAR, and IFIH1

DOI: 10.1002/ajmg.a.36887

American Journal of Medical Genetics, Part A, 2015, 167A, 296-312.

Source: <https://exaly.com/paper-pdf/61735181/citation-report.pdf>

Version: 2024-04-28

This report has been generated based on the citations recorded by exaly.com for the above article. For the latest version of this publication list, visit the link given above.

The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

#	Paper	IF	Citations
393	Systemic Lupus Erythematosus--A Disease with A Dysregulated Type I Interferon System. 2015 , 82, 199-207		71
392	Professor Jean Aicardi. <i>European Journal of Paediatric Neurology</i> , 2015 , 19, 615-616	3.8	
391	SAMHD1 specifically restricts retroviruses through its RNase activity. 2015 , 12, 46		57
390	A field guide to current advances in paediatric movement disorders. 2015 , 28, 437-46		9
389	Unusual cutaneous features associated with a heterozygous gain-of-function mutation in IFIH1: overlap between Aicardi-Goutières and Singleton-Merten syndromes. 2015 , 173, 1505-13		55
388	Intrinsic host restrictions to HIV-1 and mechanisms of viral escape. 2015 , 16, 546-53		199
387	Aicardi-Goutières syndrome and the type I interferonopathies. 2015 , 15, 429-40		493
386	Isoforms of RNA-Editing Enzyme ADAR1 Independently Control Nucleic Acid Sensor MDA5-Driven Autoimmunity and Multi-organ Development. 2015 , 43, 933-44		241
385	[Type I interferonopathies]. 2015 , 142, 653-63		10
384	Translating nucleic acid-sensing pathways into therapies. 2015 , 15, 529-44		95
383	Disease specific therapies in leukodystrophies and leukoencephalopathies. <i>Molecular Genetics and Metabolism</i> , 2015 , 114, 527-36	3.7	35
382	Emerging treatments for pediatric leukodystrophies. 2015 , 62, 649-66		12
381	Human disease phenotypes associated with mutations in TREX1. <i>Journal of Clinical Immunology</i> , 2015 , 35, 235-43	5.7	108
380	RNA degradation in antiviral immunity and autoimmunity. 2015 , 36, 179-88		56
379	New monogenic autoinflammatory diseases--a clinical overview. 2015 , 37, 387-94		31
378	Inherited anomalies of innate immune receptors in pediatric-onset inflammatory diseases. 2015 , 14, 1147-53		11
377	SAMHD1: at the crossroads of cell proliferation, immune responses, and virus restriction. 2015 , 23, 680-692		59

376	Type I interferon dysregulation and neurological disease. 2015 , 11, 515-23		30
375	Reprogramming, Circular Reasoning and Self versus Non-self: One-Stop Shopping with RNA Editing. <i>Frontiers in Genetics</i> , 2016 , 7, 100	4.5	6
374	Functions of the RNA Editing Enzyme ADAR1 and Their Relevance to Human Diseases. 2016 , 7,		41
373	Genome-wide association meta-analysis in Chinese and European individuals identifies ten new loci associated with systemic lupus erythematosus. 2016 , 48, 940-946		183
372	Expression of Long Interspersed Nuclear Element 1 Retroelements and Induction of Type I Interferon in Patients With Systemic Autoimmune Disease. <i>Arthritis and Rheumatology</i> , 2016 , 68, 2686-2696	8.5	108
371	Cutaneous lupus erythematosus: updates on pathogenesis and associations with systemic lupus. 2016 , 28, 453-9		47
370	Ribonuclease H2 mutations induce a cGAS/STING-dependent innate immune response. 2016 , 35, 831-44		145
369	Clinical, radiological and possible pathological overlap of cystic leukoencephalopathy without megalencephaly and Aicardi-Goutières syndrome. <i>European Journal of Paediatric Neurology</i> , 2016 , 20, 604-10	3.8	24
368	The role of RNA editing by ADAR1 in prevention of innate immune sensing of self-RNA. 2016 , 94, 1095-1102		15
367	Loss of function of PCDH12 underlies recessive microcephaly mimicking intrauterine infection. <i>Neurology</i> , 2016 , 86, 2016-24	6.5	22
366	Insights from Mendelian Interferonopathies: Comparison of CANDLE, SAVI with AGS, Monogenic Lupus. 2016 , 94, 1111-1127		68
365	DNA sensor cGAS-mediated immune recognition. 2016 , 7, 777-791		65
364	Abnormal regulation of the antiviral response in neurological/neurodegenerative diseases. 2016 , 88, 251-258		7
363	The AIM2-like Receptors Are Dispensable for the Interferon Response to Intracellular DNA. 2016 , 45, 255-66		115
362	Systemic Lupus Erythematosus: Is It One Disease?. 2016 , 12, 274-281		1
361	Exploring Autoimmunity in a Cohort of Children with Genetically Confirmed Aicardi-Goutières Syndrome. <i>Journal of Clinical Immunology</i> , 2016 , 36, 693-9	5.7	14
360	Nucleic acid sensing and innate immunity: signaling pathways controlling viral pathogenesis and autoimmunity. 2016 , 3, 132-141		11
359	New insights into the immunopathogenesis of systemic lupus erythematosus. 2016 , 12, 716-730		541

358	The importance of chilblains as a diagnostic clue for mild Aicardi-Goutières syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2016 , 170, 3308-3312	2.5	9
357	Discriminating self from non-self in nucleic acid sensing. 2016 , 16, 566-80		253
356	Type I interferonopathies in pediatric rheumatology. 2016 , 14, 35		71
355	Type I interferon-mediated monogenic autoinflammation: The type I interferonopathies, a conceptual overview. <i>Journal of Experimental Medicine</i> , 2016 , 213, 2527-2538	16.6	243
354	Monogenic Lupus. 2016 , 18, 71		35
353	Restricting retrotransposons: a review. 2016 , 7, 16		226
352	Clinical and pathologic features of Aicardi-Goutières syndrome due to an IFIH1 mutation: A pediatric case report. <i>American Journal of Medical Genetics, Part A</i> , 2016 , 170A, 1317-24	2.5	8
351	Spondyloenchondrodysplasia Due to Mutations in ACP5: A Comprehensive Survey. <i>Journal of Clinical Immunology</i> , 2016 , 36, 220-34	5.7	48
350	DNA polymerase- β regulates the activation of type I interferons through cytosolic RNA:DNA synthesis. 2016 , 17, 495-504		83
349	RNase H2 catalytic core Aicardi-Goutières syndrome-related mutant invokes cGAS-STING innate immune-sensing pathway in mice. <i>Journal of Experimental Medicine</i> , 2016 , 213, 329-36	16.6	133
348	Systemic lupus erythematosus: Is it one disease?. 2016 , 12, 274-81		15
347	Shaping the spectrum - From autoinflammation to autoimmunity. <i>Clinical Immunology</i> , 2016 , 165, 21-8	9	47
346	In Memoriam: Professor Jean Aicardi (1926-2015). 2016 , 54, 3-4		0
345	The druggability of intracellular nucleotide-degrading enzymes. 2016 , 77, 883-93		15
344	Neuroradiologic patterns and novel imaging findings in Aicardi-Goutières syndrome. <i>Neurology</i> , 2016 , 86, 28-35	6.5	44
343	Understanding Human Autoimmunity and Autoinflammation Through Transcriptomics. 2017 , 35, 337-370		44
342	Childhood leukodystrophies: A literature review of updates on new definitions, classification, diagnostic approach and management. 2017 , 39, 369-385		26
341	A systematic approach to autoinflammatory syndromes: a spelling booklet for the beginner. 2017 , 13, 571-597		43

340	Inflammatory myopathy in a patient with Aicardi-Goutières syndrome. 2017 , 60, 154-158		10
339	Interferon-Stimulated Gene Expression as a Preferred Biomarker for Disease Activity in Aicardi-Goutières Syndrome. 2017 , 37, 147-152		5
338	Intracellular Nucleic Acid Detection in Autoimmunity. 2017 , 35, 313-336		130
337	Genetic, Phenotypic, and Interferon Biomarker Status in ADAR1-Related Neurological Disease. <i>Neuropediatrics</i> , 2017 , 48, 166-184	1.6	35
336	Discrimination of cytosolic self and non-self RNA by RIG-I-like receptors. 2017 , 292, 9000-9009		52
335	With me or against me: Tumor suppressor and drug resistance activities of SAMHD1. 2017 , 52, 32-39		23
334	RNA editing signature during myeloid leukemia cell differentiation. 2017 , 31, 2824-2832		16
333	Roles of SAMHD1 in antiviral defense, autoimmunity and cancer. 2017 , 27, e1931		13
332	Genetic and Metabolic Disorders of the White Matter. 2017 , 747-758		
331	Confirmation of five novel susceptibility loci for systemic lupus erythematosus (SLE) and integrated network analysis of 82 SLE susceptibility loci. 2017 , 26, 1205-1216		32
330	No shortcuts: new findings reinforce why nuance is the rule in genetic autoinflammatory syndromes. 2017 , 29, 506-515		5
329	Musculoskeletal Disease in MDA5-Related Type I Interferonopathy: A Mendelian Mimic of Jaccoud's Arthropathy. <i>Arthritis and Rheumatology</i> , 2017 , 69, 2081-2091	9.5	30
328	Aicardi-Goutières syndrome: unusual neuro-radiological manifestations. 2017 , 32, 679-683		7
327	Type I interferon-mediated autoimmune diseases: pathogenesis, diagnosis and targeted therapy. 2017 , 56, 1662-1675		50
326	Lupus pathobiology based on genomics. 2017 , 69, 1-12		17
325	Monogenic lupus: it's all new!. 2017 , 49, 87-95		32
324	Updates in Lupus Genetics. 2017 , 19, 68		63
323	Rare ADAR and RNASEH2B variants and a type I interferon signature in glioma and prostate carcinoma risk and tumorigenesis. 2017 , 134, 905-922		9

322	RNA editing by ADAR1 regulates innate and antiviral immune functions in primary macrophages. 2017 , 7, 13339		22
321	Lack of Trex1 Causes Systemic Autoimmunity despite the Presence of Antiretroviral Drugs. <i>Journal of Immunology</i> , 2017 , 199, 2261-2269	5.3	26
320	Chimerism for 20q11.2 microdeletion of GDF5 explains discordant phenotypes in monozygotic-diamniotic twins. <i>American Journal of Medical Genetics, Part A</i> , 2017 , 173, 3182-3188	2.5	2
319	The TREX1 Dinosaur Bites the Brain through the LINE. 2017 , 21, 287-288		4
318	SAMHD1 Mutations Are Also Responsible for Aicardi-Goutières in the Cree Population. 2017 , 44, 749-751		
317	Genomics of Systemic Lupus Erythematosus: Insights Gained by Studying Monogenic Young-Onset Systemic Lupus Erythematosus. 2017 , 43, 415-434		24
316	Neonatal detection of Aicardi Goutières Syndrome by increased C26:0 lysophosphatidylcholine and interferon signature on newborn screening blood spots. <i>Molecular Genetics and Metabolism</i> , 2017 , 122, 134-139	3.7	21
315	Late diagnosis and atypical brain imaging of Aicardi-Goutières syndrome: are we failing to diagnose Aicardi-Goutières syndrome-2?. 2017 , 59, 1307-1311		7
314	Modeling of TREX1-Dependent Autoimmune Disease using Human Stem Cells Highlights L1 Accumulation as a Source of Neuroinflammation. 2017 , 21, 319-331.e8		158
313	Adenosine Deaminases That Act on RNA (ADARs). 2017 , 41, 215-268		13
312	Epitranscriptomic profiling across cell types reveals associations between APOBEC1-mediated RNA editing, gene expression outcomes, and cellular function. 2017 , 114, 13296-13301		21
311	Type I interferon-mediated autoinflammation and autoimmunity. 2017 , 49, 96-102		52
310	Double deficiency of Trex2 and DNase1L2 nucleases leads to accumulation of DNA in lingual cornifying keratinocytes without activating inflammatory responses. 2017 , 7, 11902		10
309	Genetic interferonopathies: An overview. 2017 , 31, 441-459		20
308	Periodic fever syndromes. 2017 , 31, 596-609		54
307	Juvenile-onset systemic lupus erythematosus (jSLE) - Pathophysiological concepts and treatment options. 2017 , 31, 488-504		31
306	Toward an Inclusive, Congruent, and Precise Definition of Autoinflammatory Diseases. <i>Frontiers in Immunology</i> , 2017 , 8, 497	8.4	13
305	The Dynamic Interplay between HIV-1, SAMHD1, and the Innate Antiviral Response. <i>Frontiers in Immunology</i> , 2017 , 8, 1541	8.4	23

304	Rewriting the transcriptome: adenosine-to-inosine RNA editing by ADARs. 2017 , 18, 205		97
303	Progress towards precision medicine for lupus: the role of genetic biomarkers. 2018 , 3, 119-135		3
302	Neurodegenerative diseases have genetic hallmarks of autoinflammatory disease. 2018 , 27, R108-R118		17
301	Therapeutic Approaches to Type I Interferonopathies. 2018 , 20, 32		16
300	Identification of Significant Gene Signatures and Prognostic Biomarkers for Patients With Cervical Cancer by Integrated Bioinformatic Methods. 2018 , 17, 1533033818767455		21
299	The Twists of Pediatric Dystonia: Phenomenology, Classification, and Genetics. 2018 , 25, 65-74		9
298	Novel RNASET2 Pathogenic Variants in an East Asian Child with Delayed Psychomotor Development. 2018 , 37, 15-21		1
297	Sort Your Self Out!. 2018 , 172, 640-642		3
296	Review: Cell Death, Nucleic Acids, and Immunity: Inflammation Beyond the Grave. <i>Arthritis and Rheumatology</i> , 2018 , 70, 805-816	9.5	45
295	Bilateral striopallidodentate calcinosis associated with Sjögren's syndrome and IgD γ monoclonal gammopathy of undetermined significance. 2018 , 85, 243-245		2
294	LINE-1 retrotransposons in healthy and diseased human brain. 2018 , 78, 434-455		30
293	A central role for PI3K-AKT signaling pathway in linking SAMHD1-deficiency to the type I interferon signature. 2018 , 8, 84		20
292	Autoinflammatory phenotypes in Aicardi-Goutières syndrome with interferon upregulation and serological autoimmune features. <i>Journal of Allergy and Clinical Immunology</i> , 2018 , 141, 1135-1138	11.5	1
291	A novel IFIH1 mutation in the pincer domain underlies the clinical features of both Aicardi-Goutières and Singleton-Merten syndromes in a single patient. 2018 , 178, e111-e113		9
290	Interferonopathies in laboratory-negative suspected congenital infection. 2018 , 18, 27		
289	Pro-inflammation Associated with a Gain-of-Function Mutation (R284S) in the Innate Immune Sensor STING. 2018 , 23, 1112-1123		53
288	SAMHD1: Recurring roles in cell cycle, viral restriction, cancer, and innate immunity. 2018 , 51, 96-110		20
287	[Type I interferonopathies. Literature review]. <i>Revue De Medecine Interne</i> , 2018 , 39, 271-278	0.1	8

286	Phenotypic and Molecular Spectrum of Aicardi-Goutières Syndrome: A Study of 24 Patients. 2018 , 78, 35-40		28
285	Aicardi-Goutières syndrome with muscle involvement in early infancy. 2018 , 44, 737-742		1
284	Variable clinical phenotype in two siblings with Aicardi-Goutières syndrome type 6 and a novel mutation in the ADAR gene. <i>European Journal of Paediatric Neurology</i> , 2018 , 22, 186-189	3.8	9
283	RNASEH1 gene variants are associated with autoimmune type 1 diabetes in Colombia. 2018 , 41, 755-764		2
282	Recent developments in systemic lupus erythematosus pathogenesis and applications for therapy. 2018 , 30, 222-228		26
281	p.Arg69Trp in RNASEH2C is a founder variant in three Indian families with Aicardi-Goutières syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2018 , 176, 156-160	2.5	4
280	Adenosine-to-Inosine RNA Editing in Health and Disease. 2018 , 29, 846-863		21
279	Monogenic Autoimmune Diseases. 2018 , 25, 213		0
278	Sine causa tetraparesis: A pilot study on its possible relationship with interferon signature analysis and Aicardi Goutières syndrome related genes analysis. 2018 , 97, e13893		6
277	Monogenic Lupus: A Developing Paradigm of Disease. <i>Frontiers in Immunology</i> , 2018 , 9, 2496	8.4	54
276	The Broad-Ranging Panorama of Systemic Autoinflammatory Disorders with Specific Focus on Acute Painful Symptoms and Hematologic Manifestations in Children. 2018 , 10, e2018067		18
275	Vasculitis in Systemic Autoinflammatory Diseases. <i>Frontiers in Pediatrics</i> , 2018 , 6, 377	3.4	19
274	Combination of exome sequencing and immune testing confirms Aicardi-Goutières syndrome type 5 in a challenging pediatric neurology case. 2018 , 4,		4
273	ADAR1-mediated RNA editing is required for thymic self-tolerance and inhibition of autoimmunity. 2018 , 19,		27
272	Brain Organoids and the Study of Neurodevelopment. 2018 , 24, 982-990		44
271	Gene-level associations in suicide attempter families show overrepresentation of synaptic genes and genes differentially expressed in brain development. 2018 , 177, 774-784		13
270	[Familial chilblain lupus: Four cases spanning three generations]. 2018 , 145, 683-689		1
269	Two RNase H2 Mutants with Differential rNMP Processing Activity Reveal a Threshold of Ribonucleotide Tolerance for Embryonic Development. 2018 , 25, 1135-1145.e5		22

268	The classification, genetic diagnosis and modelling of monogenic autoinflammatory disorders. 2018 , 132, 1901-1924		14
267	L1 retrotransposition in the soma: a field jumping ahead. 2018 , 9, 22		42
266	Interferons and Proinflammatory Cytokines in Pregnancy and Fetal Development. 2018 , 49, 397-412		149
265	Dawn of Epitranscriptomic Medicine. 2018 , 11, e001927		16
264	Cellular Control of Endogenous Retroviruses and Retroelements. 2018 , 479-525		
263	Aicardi goutières syndrome is associated with pulmonary hypertension. <i>Molecular Genetics and Metabolism</i> , 2018 , 125, 351-358	3-7	20
262	Ribonucleotide Excision Repair Is Essential to Prevent Squamous Cell Carcinoma of the Skin. 2018 , 78, 5917-5926		26
261	Autosomal-dominant early-onset spastic paraparesis with brain calcification due to IFIH1 gain-of-function. 2018 , 39, 1076-1080		4
260	Dyschromatosis symmetrica hereditaria with chilblains due to a novel two-amino-acid deletion in the double-stranded RNA-binding domain of ADAR1. 2018 , 32, e394-e396		1
259	Inhibition of Cyclic GMP-AMP Synthase Using a Novel Antimalarial Drug Derivative in Trex1-Deficient Mice. <i>Arthritis and Rheumatology</i> , 2018 , 70, 1807-1819	9-5	43
258	An Update on Autoinflammatory Diseases: Interferonopathies. 2018 , 20, 38		43
257	Monogenic systemic lupus erythematosus: insights in pathophysiology. 2018 , 38, 1763-1775		11
256	Acute and chronic viral infections. 2017 , 145, 227-243		7
255	RNase H2, mutated in Aicardi-Goutières syndrome, promotes LINE-1 retrotransposition. 2018 , 37,		34
254	TREX1 Mutation Causing Autosomal Dominant Thrombotic Microangiopathy and CKD-A Novel Presentation. 2018 , 72, 895-899		3
253	RNase H2 Loss in Murine Astrocytes Results in Cellular Defects Reminiscent of Nucleic Acid-Mediated Autoinflammation. <i>Frontiers in Immunology</i> , 2018 , 9, 587	8.4	12
252	Degenerative Disorders of the Newborn. 2018 , 823-858.e11		
251	Astrocytes, an active player in Aicardi-Goutières syndrome. 2018 , 28, 399-407		18

250	Encephalopathies with intracranial calcification in children: clinical and genetic characterization. 2018 , 13, 135			12
249	SAMHD1 deficient human monocytes autonomously trigger type I interferon. 2018 , 101, 450-460			10
248	ADAR1 attenuates allogeneic graft rejection by suppressing miR-21 biogenesis in macrophages and promoting M2 polarization. 2018 , 32, 5162-5173			9
247	Constitutively Active MDA5 Proteins Are Inhibited by Paramyxovirus V Proteins. 2018 , 38, 319-332			5
246	Calcinose striopallidodentale bilatérale associée à un syndrome de Sjögren et une gammopathie monoclonale IgD de signification indéterminée. 2018 , 85, 587-590			
245	Whole Exome Sequencing of Patients from Multicase Families with Systemic Lupus Erythematosus Identifies Multiple Rare Variants. 2018 , 8, 8775			14
244	Microcephaly and intracranial calcifications: Not always TORCH infection!. 2019 , 55, 1000-1001			
243	ADAR1: "Editor-in-Chief" of Cytoplasmic Innate Immunity. <i>Frontiers in Immunology</i> , 2019 , 10, 1763	8.4		70
242	RNASEH2B Related Adult-Onset Interferonopathy. <i>Journal of Clinical Immunology</i> , 2019 , 39, 620-622	5.7		3
241	Genome-wide mutagenesis resulting from topoisomerase 1-processing of unrepaired ribonucleotides in DNA. 2019 , 84, 102641			6
240	The missing link: allostery and catalysis in the anti-viral protein SAMHD1. 2019 , 47, 1013-1027			5
239	Trypanosoma brucei ribonuclease H2A is an essential R-loop processing enzyme whose loss causes DNA damage during transcription initiation and antigenic variation. 2019 , 47, 9180-9197			17
238	Whole exome sequencing in childhood-onset lupus frequently detects single gene etiologies. 2019 , 17, 52			16
237	Autoinflammatory Diseases. 2019 , 123-133			
236	Hereditary spastic paraplegia associated with a rare mutation: a case report and literature review. 2019 , 156, 28			2
235	Characterization of six recombinant human RNase H2 bearing Aicardi-Goutières syndrome causing mutations. 2019 , 166, 537-545			3
234	Chronic Encephalopathy, Startle, and Intracranial Calcification: Think Beyond Intrauterine Infections. 2019 , 97, 78-79			
233	Prenatal presentation of Aicardi-Goutières syndrome: Nonspecific phenotype necessitates exome sequencing for definitive diagnosis. 2019 , 39, 806-810			1

232	Systemic lupus erythematosus and immunodeficiency. 2019 , 42, 1-9		20
231	Aicardi-Goutières syndrome gene Rnaseh2c is a metastasis susceptibility gene in breast cancer. 2019 , 15, e1008020		5
230	Molecular Genetics and Interferon Signature in the Italian Aicardi Goutières Syndrome Cohort: Report of 12 New Cases and Literature Review. 2019 , 8,		16
229	Systemic Lupus Erythematosus. 2019 , 1-17		
228	Management strategies and future directions for systemic lupus erythematosus in adults. 2019 , 393, 2332-2343		132
227	Ribonucleotides in mitochondrial DNA. 2019 , 593, 1554-1565		8
226	Mosaic Tetrasomy 9p Associated With Inflammatory Bowel Disease. 2019 , 13, 1474-1478		1
225	Non-self mutation: double-stranded RNA elicits antiviral pathogenic response in a Drosophila model of expanded CAG repeat neurodegenerative diseases. 2019 , 28, 3000-3012		5
224	Microglia responses to interleukin-6 and type I interferons in neuroinflammatory disease. 2019 , 67, 1821-1841	27	
223	Sources of Pathogenic Nucleic Acids in Systemic Lupus Erythematosus. <i>Frontiers in Immunology</i> , 2019 , 10, 1028	8.4	25
222	Brain organoids as a model system for human neurodevelopment and disease. 2019 , 95, 93-97		26
221	Spontaneous MRI improvement and absence of cerebral calcification in Aicardi-Goutières syndrome: Diagnostic and disease-monitoring implications. <i>Molecular Genetics and Metabolism</i> , 2019 , 126, 489-494	3.7	9
220	Aspirin meets cGAS. 2019 , 15, 254-255		3
219	Current therapies and therapeutic decision making for childhood-onset movement disorders. 2019 , 34, 637-656		9
218	Genotoxic stress increases cytoplasmic mitochondrial DNA editing by human APOBEC3 mutator enzymes at a single cell level. 2019 , 9, 3109		9
217	Neuropathological Findings in a Case of -Related Aicardi-Goutières Syndrome. 2019 , 22, 566-570		4
216	Genome instability consequences of RNase H2 Aicardi-Goutières syndrome alleles. 2019 , 84, 102614		3
215	Genetic Interferonopathies. 2019 , 433-453		1

214	Phosphoinositide Interactions Position cGAS at the Plasma Membrane to Ensure Efficient Distinction between Self- and Viral DNA. 2019 , 176, 1432-1446.e11		99
213	Acetylation Blocks cGAS Activity and Inhibits Self-DNA-Induced Autoimmunity. 2019 , 176, 1447-1460.e14		114
212	Immune Profiling and Precision Medicine in Systemic Lupus Erythematosus. <i>Cells</i> , 2019 , 8,	7.9	21
211	How common are single gene mutations as a cause for lacunar stroke? A targeted gene panel study. <i>Neurology</i> , 2019 , 93, e2007-e2020	6.5	10
210	Current State of Precision Medicine in Primary Systemic Vasculitides. <i>Frontiers in Immunology</i> , 2019 , 10, 2813	8.4	7
209	Pathogenesis and treatment of autoimmune rheumatic diseases. 2019 , 31, 307-315		18
208	Genetics of Human SLE. 2019 , 54-68		0
207	DDX58 and Classic Singleton-Merten Syndrome. <i>Journal of Clinical Immunology</i> , 2019 , 39, 75-80	5.7	21
206	Discrimination Between Self and Non-Self-Nucleic Acids by the Innate Immune System. 2019 , 344, 1-30		20
205	Systemic features of retinal vasculopathy with cerebral leukoencephalopathy and systemic manifestations: a monogenic small vessel disease. 2019 , 285, 317-332		18
204	Type I Interferons in Autoimmune Disease. 2019 , 14, 369-393		96
203	The Role of Nucleic Acid Sensing in Controlling Microbial and Autoimmune Disorders. 2019 , 345, 35-136		17
202	Treatments in Aicardi-Goutières syndrome. 2020 , 62, 42-47		32
201	An extremely severe case of Aicardi-Goutières syndrome 7 with a novel variant in IFIH1. 2020 , 63, 103646		3
200	Developmental Outcomes of Aicardi Goutières Syndrome. 2020 , 35, 7-16		15
199	239th ENMC International Workshop: Classification of dermatomyositis, Amsterdam, the Netherlands, 14-16 December 2018. 2020 , 30, 70-92		53
198	Toward a better understanding of type I interferonopathies: a brief summary, update and beyond. <i>World Journal of Pediatrics</i> , 2020 , 16, 44-51	4.6	17
197	TREX1 variants in Sjogren's syndrome related lymphomagenesis. 2020 , 132, 154781		13

196	High Prevalence and Disease Correlation of Autoantibodies Against p40 Encoded by Long Interspersed Nuclear Elements in Systemic Lupus Erythematosus. <i>Arthritis and Rheumatology</i> , 2020 , 72, 89-99	9.5	15
195	Novel and emerging treatments for Aicardi-Goutières syndrome. 2020 , 16, 189-198		16
194	An update on clinical, pathological, diagnostic, and therapeutic perspectives of childhood leukodystrophies. 2020 , 20, 65-84		17
193	The pathogenesis of systemic lupus erythematosus: Harnessing big data to understand the molecular basis of lupus. <i>Journal of Autoimmunity</i> , 2020 , 110, 102359	15.5	50
192	cGAS/STING: novel perspectives of the classic pathway.. 2020 , 1, 7		6
191	RNases H1 and H2: guardians of the stability of the nuclear genome when supply of dNTPs is limiting for DNA synthesis. 2020 , 66, 1073-1084		1
190	Endocrinopathies in Aicardi Goutières syndrome-A descriptive case series. 2020 , 8, 2181-2185		0
189	How Retroviruses and Retrotransposons in Our Genome May Contribute to Autoimmunity in Rheumatological Conditions. <i>Frontiers in Immunology</i> , 2020 , 11, 593891	8.4	5
188	Threading the Pieces Together: Integrative Perspective on SARS-CoV-2. 2020 , 9,		3
187	Movement disorders in ADAR1 disease: Insights from a comprehensive cohort. 2020 , 79, 100-104		6
186	Comment on: Diagnosis of Aicardi-Goutières Syndrome in Adults. 2020 , 7, 583-584		
185	Cardiac valve involvement in -related type I interferonopathy. 2020 , 57, 475-478		6
184	Type I Interferonopathies: from a Novel Concept to Targeted Therapeutics. 2020 , 22, 32		17
183	New Horizons in the Genetic Etiology of Systemic Lupus Erythematosus and Lupus-Like Disease: Monogenic Lupus and Beyond. 2020 , 9,		30
182	Bi-allelic LoF NRROS Variants Impairing Active TGF- β Delivery Cause a Severe Infantile-Onset Neurodegenerative Condition with Intracranial Calcification. 2020 , 106, 559-569		7
181	Catatonia in a patient with Aicardi-Goutières syndrome efficiently treated with immunoabsorption. 2020 , 222, 484-486		4
180	Protective Mechanisms Against DNA Replication Stress in the Nervous System. 2020 , 11,		4
179	Mendelian disorders of immunity related to an upregulation of type I interferon. 2020 , 751-772		1

178	Val143 of human ribonuclease H2 is not critical for, but plays a role in determining catalytic activity and substrate specificity. 2020 , 15, e0228774	0
177	Molecular and physiological consequences of faulty eukaryotic ribonucleotide excision repair. 2020 , 39, e102309	19
176	ADAR1 mediated regulation of neural crest derived melanocytes and Schwann cell development. 2020 , 11, 198	16
175	Relapsing-remitting clinical course expands the phenotype of Aicardi-Goutières syndrome. 2020 , 7, 254-258	1
174	Adenosine-to-inosine RNA editing in the immune system: friend or foe?. 2020 , 77, 2931-2948	10
173	Clinical approach to the diagnosis of autoimmune encephalitis in the pediatric patient. 2020 , 7,	74
172	Contribution of rare and predicted pathogenic gene variants to childhood-onset lupus: a large, genetic panel analysis of British and French cohorts. 2020 , 2, e99-e109	16
171	Diagnosis of Aicardi-Goutières Syndrome in Adults: A Case Series. 2020 , 7, 303-307	9
170	Aicardi-Goutieres Syndrome Presenting with Congenital Glaucoma. 2020 , 87, 652	0
169	Induction of Samhd1 by interferon gamma and lipopolysaccharide in murine macrophages requires IRF1. 2020 , 50, 1321-1334	0
168	SAMHD1 Functions and Human Diseases. 2020 , 12,	13
167	[Clinical symptoms of autoinflammatory diseases]. 2020 , 71, 342-358	
166	Development of a neurologic severity scale for Aicardi Goutières Syndrome. <i>Molecular Genetics and Metabolism</i> , 2020 , 130, 153-160	3.7 11
165	Mitochondrial DNA in inflammation and immunity. 2020 , 21, e49799	159
164	RNA/DNA sensing in SLE Toll-like receptors and beyond. 2021 , 159-170	
163	PNPT1 mutations may cause Aicardi-Goutières-Syndrome. 2021 , 43, 320-324	5
162	RNA Editing in Neurological and Neurodegenerative Disorders. 2021 , 2181, 309-330	7
161	The differing pathophysiologies that underlie COVID-19-associated perniosis and thrombotic retiform purpura: a case series. 2021 , 184, 141-150	48

160	Overview of STING-Associated Vasculopathy with Onset in Infancy (SAVI) Among 21 Patients. 2021 , 9, 803-818.e11		19
159	Signaling by cGAS-STING in Neurodegeneration, Neuroinflammation, and Aging. 2021 , 44, 83-96		21
158	Aicardi-Goutières syndrome-like encephalitis in mutant mice with constitutively active MDA5. 2021 , 33, 225-240		3
157	Late-Onset Aicardi-Goutières Syndrome: A Characterization of Presenting Clinical Features. 2021 , 115, 1-6		6
156	Dirty Fish Versus Squeaky Clean Mice: Dissecting Interspecies Differences Between Animal Models of Interferonopathy. <i>Frontiers in Immunology</i> , 2020 , 11, 623650	8.4	0
155	A zebrafish reporter line reveals immune and neuronal expression of endogenous retrovirus.		1
154	Systemic inflammation and chronic kidney disease in a patient due to the RNASEH2B defect. 2021 , 19, 9		4
153	Recognize Yourself-Innate Sensing of Non-LTR Retrotransposons. 2021 , 13,		1
152	A Nuclear Export Signal Is Required for cGAS to Sense Cytosolic DNA. 2021 , 34, 108586		19
151	Atypical phenotype? The answer's in the genotype: AGS caused by a novel RNASEH2C variant combined with XLA caused by a BTK deficiency. 2021 , 60, e240-e242		
150	Differential Expression of Interferon-Alpha Protein Provides Clues to Tissue Specificity Across Type I Interferonopathies. <i>Journal of Clinical Immunology</i> , 2021 , 41, 603-609	5.7	7
149	cGAS- Stimulator of Interferon Genes Signaling in Central Nervous System Disorders. 2021 , 12, 1658-1674		2
148	Implications of Endogenous Retroelements in the Etiopathogenesis of Systemic Lupus Erythematosus. 2021 , 10,		2
147	Anesthesia for a Patient With Aicardi-Goutières Syndrome: A First Case Report. 2021 , 15, e01410		
146	Hydroxychloroquine modulates immunological pathways activated by RNA:DNA hybrids in Aicardi-Goutières syndrome patients carrying RNASEH2 mutations. 2021 , 18, 1593-1595		1
145	The epileptology of Aicardi-Goutières syndrome: electro-clinical-radiological findings. 2021 , 86, 197-209		0
144	Type I Interferonopathies in Children: An Overview. <i>Frontiers in Pediatrics</i> , 2021 , 9, 631329	3.4	10
143	Genetics of Pediatric Immune-Mediated Diseases and Human Immunity. 2021 , 39, 227-249		2

142	Inflammation in pediatric epilepsies: Update on clinical features and treatment options. 2021 , 107959		1
141	Enhanced cGAS-STING-dependent interferon signaling associated with mutations in ATAD3A.		
140	Case Report: Novel Compound Heterozygous Mutations Cause Aicardi-Goutières Syndrome. <i>Frontiers in Immunology</i> , 2021 , 12, 672952	8.4	0
139	Sensing of transposable elements by the antiviral innate immune system. 2021 ,		8
138	Case 1: Intracranial Calcifications Associated with Hepatosplenomegaly and Thrombocytopenia. 2021 , 22, e332-e334		0
137	Childhood-Onset Dystonia Attributed to Aicardi-Goutières Syndrome and Responsive to Deep Brain Stimulation. 2021 , 8, 613-615		1
136	Systemic autoinflammatory disease in adults. 2021 , 20, 102774		7
135	Collapsing Glomerulopathy as a Complication of Type I Interferon-Mediated Glomerulopathy in a Patient With RNASEH2B-Related Aicardi-Goutières Syndrome. 2021 , 78, 750-754		2
134	RNA editing at a limited number of sites is sufficient to prevent MDA5 activation in the mouse brain. 2021 , 17, e1009516		5
133	G3BP1 Inhibition Alleviates Intracellular Nucleic Acid-Induced Autoimmune Responses. <i>Journal of Immunology</i> , 2021 , 206, 2453-2467	5.3	2
132	Case Report: Aicardi-Goutières Syndrome and Singleton-Merten Syndrome Caused by a Gain-of-Function Mutation in IFIH1. <i>Frontiers in Genetics</i> , 2021 , 12, 660953	4.5	0
131	Hypoxia-induced RNASEH2A limits activation of cGAS-STING signaling in HCC and predicts poor prognosis. 2021 , 3008916211026019		0
130	Monogenic Autoinflammatory Diseases: State of the Art and Future Perspectives. 2021 , 22,		4
129	Pathology of the neurovascular unit in leukodystrophies. 2021 , 9, 103		2
128	Severe diarrhea in a 10-year-old girl with Aicardi-Goutières syndrome due to IFIH1 gene mutation. <i>American Journal of Medical Genetics, Part A</i> , 2021 , 185, 3146-3152	2.5	0
127	Immunogenetics of the Ocular Anterior Segment: Lessons from Inherited Disorders. 2021 , 2021, 6691291		
126	Opsoclonus-myoclonus in Aicardi-Goutières syndrome. 2021 , 63, 1483-1486		0
125	The skin as a critical window in unveiling the pathophysiologic principles of COVID-19. 2021 ,		5

124	Aicardi-Goutières syndrome-associated mutation at ADAR1 gene locus activates innate immune response in mouse brain. <i>Journal of Neuroinflammation</i> , 2021 , 18, 169	10.1	0
123	Cerebrospinal fluid neopterin as a biomarker of treatment response to Janus kinase inhibition in Aicardi-Goutières syndrome. 2021 ,		2
122	ADAR1 interaction with Z-RNA promotes editing of endogenous double-stranded RNA and prevents MDA5-dependent immune activation. 2021 , 36, 109500		9
121	cGAS-STING pathway: post-translational modifications and functions in sterile inflammatory diseases. 2021 ,		5
120	Enhanced cGAS-STING-dependent interferon signaling associated with mutations in ATAD3A. <i>Journal of Experimental Medicine</i> , 2021 , 218,	16.6	8
119	Genome assembly of the tayra (<i>Eira barbara</i> , Mustelidae) and comparative genomic analysis reveal adaptive genetic variation in the subfamily Guloninae.		
118	Mutations in the adenosine deaminase ADAR1 that prevent endogenous Z-RNA binding induce Aicardi-Goutières-syndrome-like encephalopathy. 2021 , 54, 1976-1988.e7		6
117	Protein kinase R and the integrated stress response drive immunopathology caused by mutations in the RNA deaminase ADAR1. 2021 , 54, 1948-1960.e5		10
116	An ADAR1-dependent RNA editing event in the cyclin-dependent kinase CDK13 promotes thyroid cancer hallmarks. 2021 , 20, 115		1
115	Structural and functional characterization explains loss of dNTPase activity of the cancer-specific R366C/H mutant SAMHD1 proteins. 2021 , 297, 101170		1
114	ADAR RNA Modifications, the Epitranscriptome and Innate Immunity. 2021 , 46, 758-771		10
113	Factors Regulating the Activity of LINE1 Retrotransposons. 2021 , 12,		0
112	Compendium of causative genes and their encoded proteins for common monogenic disorders. 2021 ,		2
111	Glaucoma Syndromes: Insights into Glaucoma Genetics and Pathogenesis from Monogenic Syndromic Disorders. 2021 , 12,		2
110	Autoinflammatory Disorders. 2021 , 279-313		
109	Signaling Through Nucleic Acid Sensors and Their Roles in Inflammatory Diseases. <i>Frontiers in Immunology</i> , 2020 , 11, 625833	8.4	10
108	Interferonopathien bei Kindern und Jugendlichen. <i>Springer Reference Medizin</i> , 2021 , 1-9	0	
107	Genetic Basis of Myocarditis: Myth or Reality?. 2020 , 45-89		5

106	ADAR1 interaction with Z-RNA promotes editing of endogenous double-stranded RNA and prevents MDA5-dependent immune activation.	2
105	Leukodystrophies. 2018 , 24, 130-149	5
104	Brain organoids and insights on human evolution. 2019 , 8,	4
103	Monogenetic causes of chilblains, panniculitis and vasculopathy: the Type I interferonopathies. 2020 , 155, 590-598	3
102	Recurrent Encephalopathy with Spinal Cord Involvement: An Atypical Manifestation of Aicardi-Goutières Syndrome. 2019 , 22, 111-115	7
101	Le interferonopatie di tipo I. 2021 , 40, 509-514	
100	Autosomal dominant ADAR c.3019G>A (p.(G1007R)) variant is an important mimic of hereditary spastic paraplegia and cerebral palsy. 2021 ,	0
99	Genome instability independent of type I interferon signaling drives neuropathology caused by impaired ribonucleotide excision repair. 2021 ,	5
98	Deciphering the Biological Significance of ADAR1-Z-RNA Interactions. 2021 , 22,	2
97	Late onset Aicardi-Goutières syndrome case report: a rare white matter disease mimicking as pseudo-enzyme deficiency. 2021 , 1	
96	The Role of Ku70 as a Cytosolic DNA Sensor in Innate Immunity and Beyond. 2021 , 11, 761983	1
95	Molecular biology of autoinflammatory diseases. 2021 , 41, 33	1
94	The type I interferonopathies: 10 years on. 2021 ,	20
93	Aicardi Syndrome. 2016 , 126-127	
92	Autoinflammatory Disorders Showing Pernio-like Eruptions. <i>Nishinohon Journal of Dermatology</i> , 2018 , 80, 321-326	0
91	Type I Interferonopathies: Common Pathological Features Between Congenital Infections and Genetic Disorders. 2019 , 279-294	
90	Trypanosoma brucei ribonuclease H2A is an essential enzyme that resolves R-loops associated with transcription initiation and antigenic variation.	
89	Aicardi-Goutières Syndrome gene Rnaseh2c is a metastasis susceptibility gene in breast cancer.	

88	Dysregulation in nucleic acid-sensing pathway genes is associated with cancer patients' prognosis. <i>Cancer Science</i> , 2020 , 111, 2212-2222	6.9	2
87	The phenotypic spectrum of PCDH12 associated disorders - Five new cases and review of the literature. <i>European Journal of Paediatric Neurology</i> , 2021 , 36, 7-13	3.8	0
86	Case Report: The JAK-Inhibitor Ruxolitinib Use in Aicardi-Goutieres Syndrome Due to Mutation. <i>Frontiers in Pediatrics</i> , 2021 , 9, 725868	3.4	0
85	Germline variation of Ribonuclease H2 genes in ovarian cancer patients. <i>Journal of Ovarian Research</i> , 2020 , 13, 146	5.5	
84	PKR and the Integrated Stress Response drive immunopathology caused by ADAR1 mutation.		1
83	Identification of Candidate Predictors of Lupus Flare. <i>Transactions of the American Clinical and Climatological Association</i> , 2015 , 126, 184-96	0.9	1
82	Mitochondrial Nucleic Acid as a Driver of Pathogenic Type I Interferon Induction in Mendelian Disease. <i>Frontiers in Immunology</i> , 2021 , 12, 729763	8.4	2
81	[Autoinflammation-differences between children and adults]. <i>Zeitschrift Fur Rheumatologie</i> , 2021 , 1	1.9	
80	An Aicardi-Goutieres Syndrome-Causative Point Mutation in Gene Invokes Multiorgan Inflammation and Late-Onset Encephalopathy in Mice. <i>Journal of Immunology</i> , 2021 ,	5.3	0
79	The ADAR1 editome reveals drivers of editing-specificity for ADAR1-isoforms.		
78	Monogenic Systemic Autoinflammatory Diseases. 2021 ,		
77	Mitochondrial Nucleic Acid as a Driver of Pathogenic Type I Interferon Induction in Mendelian Disease. <i>Frontiers in Immunology</i> , 2021 , 12, 729763	8.4	4
76	Early-onset vascular leukoencephalopathy caused by bi-allelic NOTCH3 variants.. <i>Neuropediatrics</i> , 2022 ,	1.6	
75	Intervention of cGAS-STING signaling in sterile inflammatory diseases.. <i>Journal of Molecular Cell Biology</i> , 2022 ,	6.3	1
74	The 2021 EULAR and ACR points to consider for diagnosis and management of autoinflammatory type I interferonopathies: CANDLE/PRAAS, SAVI and AGS.. <i>Annals of the Rheumatic Diseases</i> , 2022 ,	2.4	0
73	Lung Inflammation in STING-Associated Vasculopathy with Onset in Infancy (SAVI).. <i>Cells</i> , 2022 , 11,	7.9	1
72	Systemic lupus erythematosus as a genetic disease.. <i>Clinical Immunology</i> , 2022 , 236, 108953	9	1
71	Deep White Matter Cysts in a Patient with Aicardi-Goutieres Syndrome and Variants.. <i>Molecular Syndromology</i> , 2022 , 13, 85-87	1.5	

70	A zebrafish reporter line reveals immune and neuronal expression of endogenous retrovirus.. <i>DMM Disease Models and Mechanisms</i> , 2022 ,	4.1	
69	Intracellular virus sensor MDA5 mutation develops autoimmune myocarditis and nephritis.. <i>Journal of Autoimmunity</i> , 2022 , 127, 102794	15.5	
68	Disorders of Nucleotide Metabolism. 2022 , 213-233		
67	[When to consider type I interferonopathy in adulthood?]. <i>Revue De Medecine Interne</i> , 2022 ,	0.1	
66	The neurovascular unit in leukodystrophies: towards solving the puzzle.. <i>Fluids and Barriers of the CNS</i> , 2022 , 19, 18	7	0
65	Mutations in RNU7-1 Weaken Secondary RNA Structure, Induce MCP-1 and CXCL10 in CSF, and Result in Aicardi-Goutières Syndrome with Severe End-Organ Involvement.. <i>Journal of Clinical Immunology</i> , 2022 , 1	5.7	1
64	The 2021 European Alliance of Associations for Rheumatology/American College of Rheumatology Points to Consider for Diagnosis and Management of Autoinflammatory Type I Interferonopathies: CANDLE/PRAAS, SAVI, and AGS.. <i>Arthritis and Rheumatology</i> , 2022 ,	9.5	2
63	Activation of the cGAS-STING innate immune response in cells with deficient mitochondrial topoisomerase TOP1MT.		
62	Pathogenic insights from genetic causes of autoinflammatory inflammasomopathies and interferonopathies. <i>Journal of Allergy and Clinical Immunology</i> , 2021 ,	11.5	1
61	DNA damage contributes to neurotoxic inflammation in Aicardi-Goutières syndrome astrocytes.. <i>Journal of Experimental Medicine</i> , 2022 , 219,	16.6	4
60	Self or Non-Self? It Is also a Matter of RNA Recognition and Editing by ADAR1.. <i>Biology</i> , 2022 , 11,	4.9	0
59	Aicardi-Goutières Syndrome due to a Mutation Presenting with Deep White Matter Cysts.. <i>Molecular Syndromology</i> , 2022 , 13, 132-138	1.5	0
58	Spectrum of Neuroradiologic Findings Associated with Monogenic Interferonopathies.. <i>American Journal of Neuroradiology</i> , 2021 ,	4.4	1
57	The cytokines interleukin-6 and interferon- β induce distinct microglia phenotypes.. <i>Journal of Neuroinflammation</i> , 2022 , 19, 96	10.1	1
56	data_sheet_1.xlsx. 2018 ,		
55	presentation_1.PDF. 2018 ,		
54	TREX1 plays multiple roles in human diseases.. <i>Cellular Immunology</i> , 2022 , 375, 104527	4.4	0
53	Inborn Errors of Immunity With Fetal or Perinatal Clinical Manifestations. <i>Frontiers in Pediatrics</i> , 2022 , 10,	3.4	0

52	Analysis of clinical characteristics of children with Aicardi-Goutieres syndrome in China.. <i>World Journal of Pediatrics</i> , 2022 ,	4.6	0
51	Pathophysiological functions of self-derived DNA.. <i>International Reviews of Immunology</i> , 2022 , 1-13	4.6	
50	RNA Helicases in Microsatellite Repeat Expansion Disorders and Neurodegeneration. <i>Frontiers in Genetics</i> , 2022 , 13,	4.5	
49	Genome Replication Is Associated With Release of Immunogenic DNA Waste. <i>Frontiers in Immunology</i> , 2022 , 13,	8.4	1
48	mRNA-based vaccines against SARS-CoV-2 do not stimulate interferon stimulatory gene expression in individuals affected by Aicardi Goutières Syndrome.		0
47	ZBP1 sequesters cGAS in the cytoplasm and sustains type I interferon responses to mitochondrial DNA.		0
46	Interferonopathien bei Kindern und Jugendlichen. <i>Springer Reference Medizin</i> , 2022 , 793-801		0
45	Disorders of Nucleic Acid Metabolism, tRNA Metabolism and Ribosomal Biogenesis. 2022 , 719-734		
44	Hematologic abnormalities in Aicardi Goutières Syndrome. <i>Molecular Genetics and Metabolism</i> , 2022 ,	3.7	0
43	Oral Phenotype of Singleton-Merten Syndrome: A Systematic Review Illustrated With a Case Report. <i>Frontiers in Genetics</i> , 13,	4.5	
42	Autoinflammation Unterschiede bei Kindern und Erwachsenen. <i>Rheuma Plus</i> ,		0
41	Autoimmune and autoinflammatory diseases with mucocutaneous manifestations: A pediatric rheumatology perspective. <i>International Journal of Dermatology</i> ,	1.7	
40	Child Neurology: Aicardi-Goutières Syndrome Presenting as Recurrent Ischemic Stroke. <i>Neurology</i> , 10.12101/00000000		
39	ADAR1 mutation causes ZBP1-dependent immunopathology. <i>Nature</i> ,	50.4	3
38	Mechanistic Interplay between HIV-1 Reverse Transcriptase Enzyme Kinetics and Host SAMHD1 Protein: Viral Myeloid-Cell Tropism and Genomic Mutagenesis. 2022 , 14, 1622		1
37	Loss-of-function variants in SAT1 cause X-linked childhood-onset systemic lupus erythematosus. <i>annrheumdis-2022-22</i>		
36	Association of rare variants in genes of immune regulation with pediatric autoimmune CNS diseases.		
35	Differentiating central nervous system demyelinating disorders: The role of clinical, laboratory, imaging characteristics and peripheral blood type I interferon activity. 13,		1

- 34 Polygenic autoimmune disease risk alleles impacting B cell tolerance act in concert across shared molecular networks in mouse and in humans. 13, ○
- 33 Covid-Associated Pernio is the Product of an Abortive Sars-Cov-2 Infection Resulting in the Deposition of Inflammatory Viral Rna and a Local Interferon Response. ○
- 32 A Small Subset of Cytosolic dsRNAs Must Be Edited by ADAR1 to Evade MDA5-Mediated Autoimmunity. ○
- 31 Deep Brain Stimulation in Progressive Generalized Dystonia in Childhood Associated With ADAR1 Gene Variant. 2022, 9, ○
- 30 Innate immune activation without immune cell infiltration in brains of murine models of Aicardi-Goutières Syndrome. ○
- 29 Alternative pathways driven by STING: From innate immunity to lipid metabolism. 2022, 1 ○
- 28 Periodic fever syndromes and autoinflammatory diseases. 2022, 791-828 ○
- 27 Microglia shield the murine brain from damage mediated by the cytokines IL-6 and IFN- β 13, ○
- 26 SARS-CoV-2 mRNA-based vaccines in the Aicardi Goutières Syndrome. 2022, 137, 320-327 ○
- 25 Incidence of Aicardi-Goutières syndrome and KCNT1-related epilepsy in Denmark. 2022, 33, 100924 ○
- 24 Human LINE-1 retrotransposons: impacts on the genome and regulation by host factors. 2022, ○
- 23 Characterization of Mitochondrial Alterations in Aicardi-Goutières Patients Mutated in RNASEH2A and RNASEH2B Genes. 2022, 23, 14482 ○
- 22 An AGS-associated mutation in ADAR1 catalytic domain results in early-onset and MDA5-dependent encephalopathy with IFN pathway activation in the brain. 2022, 19, ○
- 21 Research advances in cGAS β stimulator of interferon genes pathway and central nervous system diseases: Focus on new therapeutic approaches. 15, ○
- 20 The RNA-editing enzyme ADAR1: a regulatory hub that tunes multiple dsRNA-sensing pathways. ○
- 19 Case report: Pneumocystis jirovecii pneumonia in a severe case of Aicardi-Goutières syndrome with an IFIH1 gain-of-function mutation mimicking combined immunodeficiency. 13, ○
- 18 Clinical spectrum and currently available treatment of type I interferonopathy/Aicardi-Goutières syndrome. ○
- 17 Endocrinopathies in Leukodystrophy. 2022, 101351 ○

- 16 Aicardi-Goutières syndrome (AGS): recurrent fetal cardiomyopathy and pseudo-TORCH syndrome. **2022**, 15, e249192 ○
- 15 Multiplepalsgene modules control a balance between immunity and development inCaenorhabditis elegans. ○
- 14 Clinical Non-penetrance Associated with Biallelic Mutations in the RNase H2 Complex. ○
- 13 Early arteriopathy in Aicardi-Goutières syndrome 5. Case report and review of literature. 197140092311546 ○
- 12 Breaking down the cellular responses to type I interferon neurotoxicity in the brain. 14, ○
- 11 Molecular characterization of an intronic RNASEH2B variant in a patient with Aicardi-Goutières syndrome. **2023**, 66, 104731 ○
- 10 Pathogenesis of systemic lupus erythematosus: risks, mechanisms and therapeutic targets. ar-2022-223741 1
- 9 Idiopathic Inflammatory Myopathies. **2023**, 37-72 ○
- 8 A case of Aicardi-Goutières syndrome caused by TREX1 gene mutation. **2023**, 23, ○
- 7 Characterization of a mutant samhd1 zebrafish model implicates dysregulation of cholesterol biosynthesis in Aicardi-Goutières syndrome. 14, ○
- 6 Comparison between D-loop methylation and mtDNA copy number in patients with Aicardi-Goutières Syndrome. 14, ○
- 5 Altered DNA methylation and gene expression predict disease severity in patients with Aicardi-Goutières syndrome. **2023**, 249, 109299 ○
- 4 The cellular and KSHV A-to-I RNA editome in primary effusion lymphoma and its role in the viral lifecycle. **2023**, 14, ○
- 3 Genes and Microbiota Interaction in Monogenic Autoimmune Disorders. **2023**, 11, 1127 ○
- 2 Efficacy and safety of baricitinib in Japanese patients with autoinflammatory type I interferonopathies (NNS/CANDLE, SAVI, And AGS). **2023**, 21, ○
- 1 Interferonopathies de type I : dūn nouveau concept aux thēapeutiques ciblēs. **2023**, ○