

# Andrea Ballabio

## List of Publications by Year in Descending Order

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

374  
papers

48,159  
citations

99  
h-index

213  
g-index

394  
ext. papers

55,330  
ext. citations

11.7  
avg, IF

7.22  
L-index

#	Paper	IF	Citations
374	A gene toolbox for monitoring autophagy transcription. <i>Cell Death and Disease</i> , <b>2021</b> , 12, 1044	9.8	2
373	A lysosome-targeted DNA nanodevice selectively targets macrophages to attenuate tumours. <i>Nature Nanotechnology</i> , <b>2021</b> ,	28.7	8
372	Loss of Hepatic Transcription Factor EB Attenuates Alcohol-Associated Liver Carcinogenesis. <i>American Journal of Pathology</i> , <b>2021</b> ,	5.8	1
371	TFEB links MYC signaling to epigenetic control of myeloid differentiation and acute myeloid leukemia. <i>Blood Cancer Discovery</i> , <b>2021</b> , 2, 162-185	7	6
370	Cancer of unknown primary stem-like cells model multi-organ metastasis and unveil liability to MEK inhibition. <i>Nature Communications</i> , <b>2021</b> , 12, 2498	17.4	4
369	TFEB deficiency attenuates mitochondrial degradation upon brown adipose tissue whitening at thermoneutrality. <i>Molecular Metabolism</i> , <b>2021</b> , 47, 101173	8.8	1
368	Altered heparan sulfate metabolism during development triggers dopamine-dependent autistic-behaviours in models of lysosomal storage disorders. <i>Nature Communications</i> , <b>2021</b> , 12, 3495	17.4	4
367	Autophagy in major human diseases. <i>EMBO Journal</i> , <b>2021</b> , 40, e108863	13	79
366	Repurposing of tamoxifen ameliorates CLN3 and CLN7 disease phenotype. <i>EMBO Molecular Medicine</i> , <b>2021</b> , 13, e13742	12	4
365	GABARAP sequesters the FLCN-FNIP tumor suppressor complex to couple autophagy with lysosomal biogenesis. <i>Science Advances</i> , <b>2021</b> , 7, eabj2485	14.3	5
364	The rapidly evolving view of lysosomal storage diseases. <i>EMBO Molecular Medicine</i> , <b>2021</b> , 13, e12836	12	35
363	Guidelines for the use and interpretation of assays for monitoring autophagy (4th edition). <i>Autophagy</i> , <b>2021</b> , 17, 1-382	10.2	440
362	Multiple Mechanisms Converging on Transcription Factor EB Activation by the Natural Phenol Pterostilbene.. <i>Oxidative Medicine and Cellular Longevity</i> , <b>2021</b> , 2021, 7658501	6.7	
361	TFEB regulates lysosomal exocytosis of tau and its loss of function exacerbates tau pathology and spreading. <i>Molecular Psychiatry</i> , <b>2020</b> ,	15.1	20
360	Loss of function of transcription factor EB remodels lipid metabolism and cell death pathways in the cardiomyocyte. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , <b>2020</b> , 1866, 165832	6.9	11
359	TFEB regulates murine liver cell fate during development and regeneration. <i>Nature Communications</i> , <b>2020</b> , 11, 2461	17.4	14
358	TFEB Modulates p21/WAF1/CIP1 during the DNA Damage Response. <i>Cells</i> , <b>2020</b> , 9,	7.9	5

357	Potential of rifampin activity in a mouse model of tuberculosis by activation of host transcription factor EB. <i>PLoS Pathogens</i> , <b>2020</b> , 16, e1008567	7.6	3
356	A substrate-specific mTORC1 pathway underlies Birt-Hogg-Dubé syndrome. <i>Nature</i> , <b>2020</b> , 585, 597-602	50.4	73
355	HDAC6-dependent ciliophagy is involved in ciliary loss and cholangiocarcinoma growth in human cells and murine models. <i>American Journal of Physiology - Renal Physiology</i> , <b>2020</b> , 318, G1022-G1033	5.1	13
354	LC3 lipidation is essential for TFEB activation during the lysosomal damage response to kidney injury. <i>Nature Cell Biology</i> , <b>2020</b> , 22, 1252-1263	23.4	42
353	Lysosomes as dynamic regulators of cell and organismal homeostasis. <i>Nature Reviews Molecular Cell Biology</i> , <b>2020</b> , 21, 101-118	48.7	310
352	Mit/TFE factors control ER-phagy via transcriptional regulation of FAM134B. <i>EMBO Journal</i> , <b>2020</b> , 39, e105696	13	23
351	c-Abl Inhibition Activates TFEB and Promotes Cellular Clearance in a Lysosomal Disorder. <i>iScience</i> , <b>2020</b> , 23, 101691	6.1	10
350	GADD34 is a modulator of autophagy during starvation. <i>Science Advances</i> , <b>2020</b> , 6,	14.3	10
349	Light-responsive microRNA miR-211 targets Ezrin to modulate lysosomal biogenesis and retinal cell clearance. <i>EMBO Journal</i> , <b>2020</b> , 39, e102468	13	13
348	Polyamines Control eIF5A Hypusination, TFEB Translation, and Autophagy to Reverse B Cell Senescence. <i>Molecular Cell</i> , <b>2019</b> , 76, 110-125.e9	17.6	93
347	Nutrient-sensitive transcription factors TFEB and TFE3 couple autophagy and metabolism to the peripheral clock. <i>EMBO Journal</i> , <b>2019</b> , 38,	13	34
346	The lysosomal disease caused by mutant VPS33A. <i>Human Molecular Genetics</i> , <b>2019</b> , 28, 2514-2530	5.6	14
345	Impaired TFEB-mediated lysosomal biogenesis promotes the development of pancreatitis in mice and is associated with human pancreatitis. <i>Autophagy</i> , <b>2019</b> , 15, 1954-1969	10.2	27
344	Brain tyrosinase overexpression implicates age-dependent neuromelanin production in Parkinson's disease pathogenesis. <i>Nature Communications</i> , <b>2019</b> , 10, 973	17.4	114
343	TFEB-driven endocytosis coordinates mTORC1 signaling and autophagy. <i>Autophagy</i> , <b>2019</b> , 15, 151-164	10.2	48
342	Keeping the autophagy tempo. <i>Autophagy</i> , <b>2019</b> , 15, 1854-1856	10.2	3
341	TFEB controls retromer expression in response to nutrient availability. <i>Journal of Cell Biology</i> , <b>2019</b> , 218, 3954-3966	7.3	8
340	A transcriptomic study of Williams-Beuren syndrome associated genes in mouse embryonic stem cells. <i>Scientific Data</i> , <b>2019</b> , 6, 262	8.2	1

339	TFEB activation in macrophages attenuates postmyocardial infarction ventricular dysfunction independently of ATG5-mediated autophagy. <i>JCI Insight</i> , <b>2019</b> , 4,	9.9	15
338	Transcription factor EB overexpression prevents neurodegeneration in experimental synucleinopathies. <i>JCI Insight</i> , <b>2019</b> , 4,	9.9	30
337	Enhanced lysosomal degradation maintains the quiescent state of neural stem cells. <i>Nature Communications</i> , <b>2019</b> , 10, 5446	17.4	32
336	TRPML1 links lysosomal calcium to autophagosome biogenesis through the activation of the CaMKK $\beta$ /VPS34 pathway. <i>Nature Communications</i> , <b>2019</b> , 10, 5630	17.4	58
335	Histone methyl-transferases and demethylases in the autophagy regulatory network: the emerging role of KDM1A/LSD1 demethylase. <i>Autophagy</i> , <b>2019</b> , 15, 187-196	10.2	15
334	TFEB controls vascular development by regulating the proliferation of endothelial cells. <i>EMBO Journal</i> , <b>2019</b> , 38,	13	28
333	MiT/TFE Family of Transcription Factors, Lysosomes, and Cancer. <i>Annual Review of Cancer Biology</i> , <b>2019</b> , 3, 203-222	13.3	47
332	Complex care of individuals with multiple sulfatase deficiency: Clinical cases and consensus statement. <i>Molecular Genetics and Metabolism</i> , <b>2018</b> , 123, 337-346	3.7	19
331	Enhancement of hepatic autophagy increases ureagenesis and protects against hyperammonemia. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2018</b> , 115, 391-396	11.5	23
330	Overexpression of TFEB Drives a Pleiotropic Neurotrophic Effect and Prevents Parkinson's Disease-Related Neurodegeneration. <i>Molecular Therapy</i> , <b>2018</b> , 26, 1552-1567	11.7	38
329	Impaired TFEB-Mediated Lysosome Biogenesis and Autophagy Promote Chronic Ethanol-Induced Liver Injury and Steatosis in Mice. <i>Gastroenterology</i> , <b>2018</b> , 155, 865-879.e12	13.3	123
328	mTOR-dependent phosphorylation controls TFEB nuclear export. <i>Nature Communications</i> , <b>2018</b> , 9, 3312	17.4	133
327	Excessive dietary lipid intake provokes an acquired form of lysosomal lipid storage disease in the kidney. <i>Journal of Pathology</i> , <b>2018</b> , 246, 470-484	9.4	16
326	Fasting Imparts a Switch to Alternative Daily Pathways in Liver and Muscle. <i>Cell Reports</i> , <b>2018</b> , 25, 3299-3314.e67	11.4	67
325	The complex relationship between TFEB transcription factor phosphorylation and subcellular localization. <i>EMBO Journal</i> , <b>2018</b> , 37,	13	193
324	Activation of the c-Jun N-terminal kinase pathway aggravates proteotoxicity of hepatic mutant Z alpha1-antitrypsin. <i>Hepatology</i> , <b>2017</b> , 65, 1865-1874	11.2	23
323	TFEB-mediated increase in peripheral lysosomes regulates store-operated calcium entry. <i>Scientific Reports</i> , <b>2017</b> , 7, 40797	4.9	28
322	Next-Generation Sequencing Approaches to Define the Role of the Autophagy Lysosomal Pathway in Human Disease: The Example of LysoPlex. <i>Methods in Molecular Biology</i> , <b>2017</b> , 1594, 227-241	1.4	3

321	Transcriptional activation of RagD GTPase controls mTORC1 and promotes cancer growth. <i>Science</i> , <b>2017</b> , 356, 1188-1192	33.3	106
320	Exploiting macrophage autophagy-lysosomal biogenesis as a therapy for atherosclerosis. <i>Nature Communications</i> , <b>2017</b> , 8, 15750	17.4	188
319	Molecular definitions of autophagy and related processes. <i>EMBO Journal</i> , <b>2017</b> , 36, 1811-1836	13	857
318	TFE3 regulates whole-body energy metabolism in cooperation with TFEB. <i>EMBO Molecular Medicine</i> , <b>2017</b> , 9, 605-621	12	56
317	Abnormal cell-clearance and accumulation of autophagic vesicles in lymphocytes from patients affected with Ataxia-Teleangiectasia. <i>Clinical Immunology</i> , <b>2017</b> , 175, 16-25	9	16
316	Transcription Factor EB Controls Metabolic Flexibility during Exercise. <i>Cell Metabolism</i> , <b>2017</b> , 25, 182-196	4.6	169
315	Lysosome signaling controls the migration of dendritic cells. <i>Science Immunology</i> , <b>2017</b> , 2,	28	64
314	Methods to Monitor and Manipulate TFEB Activity During Autophagy. <i>Methods in Enzymology</i> , <b>2017</b> , 588, 61-78	1.7	8
313	STUB1 regulates TFEB-induced autophagy-lysosome pathway. <i>EMBO Journal</i> , <b>2017</b> , 36, 2544-2552	13	96
312	AAV-mediated transcription factor EB (TFEB) gene delivery ameliorates muscle pathology and function in the murine model of Pompe Disease. <i>Scientific Reports</i> , <b>2017</b> , 7, 15089	4.9	29
311	mTORC1 hyperactivation arrests bone growth in lysosomal storage disorders by suppressing autophagy. <i>Journal of Clinical Investigation</i> , <b>2017</b> , 127, 3717-3729	15.9	58
310	TFEB-mTORC1 feedback loop in metabolism and cancer. <i>Cell Stress</i> , <b>2017</b> , 1, 7-10	5.5	2
309	TFEB at a glance. <i>Journal of Cell Science</i> , <b>2016</b> , 129, 2475-81	5.3	350
308	Drosophila Mitf regulates the V-ATPase and the lysosomal-autophagic pathway. <i>Autophagy</i> , <b>2016</b> , 12, 484-98	10.2	53
307	Guidelines for the use and interpretation of assays for monitoring autophagy (3rd edition). <i>Autophagy</i> , <b>2016</b> , 12, 1-222	10.2	3838
306	Activation of the transcription factor EB rescues lysosomal abnormalities in cystinotic kidney cells. <i>Kidney International</i> , <b>2016</b> , 89, 862-73	9.9	62
305	The Autophagy-Lysosomal Pathway in Neurodegeneration: A TFEB Perspective. <i>Trends in Neurosciences</i> , <b>2016</b> , 39, 221-234	13.3	195
304	Dysregulation of Nutrient Sensing and CLEARance in Presenilin Deficiency. <i>Cell Reports</i> , <b>2016</b> , 14, 2166-2170	7.0	75

303	Modelling TFE renal cell carcinoma in mice reveals a critical role of WNT signaling. <i>ELife</i> , <b>2016</b> , 5,	8.9	45
302	Autophagosome-lysosome fusion triggers a lysosomal response mediated by TLR9 and controlled by OCRL. <i>Nature Cell Biology</i> , <b>2016</b> , 18, 839-850	23.4	103
301	The awesome lysosome. <i>EMBO Molecular Medicine</i> , <b>2016</b> , 8, 73-6	12	105
300	TFEB, Master Regulator of Cellular Clearance <b>2016</b> , 101-114		1
299	A novel curcumin analog binds to and activates TFEB in vitro and in vivo independent of MTOR inhibition. <i>Autophagy</i> , <b>2016</b> , 12, 1372-89	10.2	97
298	TFEB and TFE3 cooperate in the regulation of the innate immune response in activated macrophages. <i>Autophagy</i> , <b>2016</b> , 12, 1240-58	10.2	150
297	Lysosomal calcium signalling regulates autophagy through calcineurin and TFEB. <i>Nature Cell Biology</i> , <b>2015</b> , 17, 288-99	23.4	716
296	Lysoplex: An efficient toolkit to detect DNA sequence variations in the autophagy-lysosomal pathway. <i>Autophagy</i> , <b>2015</b> , 11, 928-38	10.2	38
295	Fighting rare diseases: the model of the telethon research institutes in Italy. <i>Human Gene Therapy</i> , <b>2015</b> , 26, 183-5	4.8	2
294	Transcriptional and epigenetic regulation of autophagy in aging. <i>Autophagy</i> , <b>2015</b> , 11, 867-80	10.2	207
293	Neuronal-Targeted TFEB Accelerates Lysosomal Degradation of APP, Reducing A $\beta$ Generation and Amyloid Plaque Pathogenesis. <i>Journal of Neuroscience</i> , <b>2015</b> , 35, 12137-51	6.6	134
292	Lysosomal calcium regulates autophagy. <i>Autophagy</i> , <b>2015</b> , 11, 970-1	10.2	63
291	Lysosomal storage diseases: from pathophysiology to therapy. <i>Annual Review of Medicine</i> , <b>2015</b> , 66, 471-86	16.4	239
290	Lysosomal adaptation: how the lysosome responds to external cues. <i>Cold Spring Harbor Perspectives in Biology</i> , <b>2014</b> , 6,	10.2	71
289	Cell metabolism: autophagy transcribed. <i>Nature</i> , <b>2014</b> , 516, 40-1	50.4	19
288	Intracerebral administration of adeno-associated viral vector serotype rh.10 carrying human SGSH and SUMF1 cDNAs in children with mucopolysaccharidosis type IIIA disease: results of a phase I/II trial. <i>Human Gene Therapy</i> , <b>2014</b> , 25, 506-16	4.8	177
287	Phosphorylation-regulated degradation of the tumor-suppressor form of PED by chaperone-mediated autophagy in lung cancer cells. <i>Journal of Cellular Physiology</i> , <b>2014</b> , 229, 1359-68	7	38
286	Lysosome: regulator of lipid degradation pathways. <i>Trends in Cell Biology</i> , <b>2014</b> , 24, 743-50	18.3	129

285	Induction of lysosomal biogenesis in atherosclerotic macrophages can rescue lipid-induced lysosomal dysfunction and downstream sequelae. <i>Arteriosclerosis, Thrombosis, and Vascular Biology</i> , <b>2014</b> , 34, 1942-1952	9.4	147
284	Wilson disease protein ATP7B utilizes lysosomal exocytosis to maintain copper homeostasis. <i>Developmental Cell</i> , <b>2014</b> , 29, 686-700	10.2	146
283	Multiple sulfatase deficiency with neonatal manifestation. <i>Italian Journal of Pediatrics</i> , <b>2014</b> , 40, 86	3.2	10
282	A non-conserved miRNA regulates lysosomal function and impacts on a human lysosomal storage disorder. <i>Nature Communications</i> , <b>2014</b> , 5, 5840	17.4	30
281	Selective clearance of aberrant tau proteins and rescue of neurotoxicity by transcription factor EB. <i>EMBO Molecular Medicine</i> , <b>2014</b> , 6, 1142-60	12	227
280	The phytoestrogen genistein modulates lysosomal metabolism and transcription factor EB (TFEB) activation. <i>Journal of Biological Chemistry</i> , <b>2014</b> , 289, 17054-69	5.4	87
279	Neonatal multiple sulfatase deficiency with a novel mutation and review of the literature. <i>Turkish Journal of Pediatrics</i> , <b>2014</b> , 56, 418-22	0.7	5
278	Transcription factor EB (TFEB) is a new therapeutic target for Pompe disease. <i>EMBO Molecular Medicine</i> , <b>2013</b> , 5, 691-706	12	224
277	Signals from the lysosome: a control centre for cellular clearance and energy metabolism. <i>Nature Reviews Molecular Cell Biology</i> , <b>2013</b> , 14, 283-96	48.7	1043
276	TFEB controls cellular lipid metabolism through a starvation-induced autoregulatory loop. <i>Nature Cell Biology</i> , <b>2013</b> , 15, 647-58	23.4	599
275	Defective autophagy in spastizin mutated patients with hereditary spastic paraparesis type 15. <i>Brain</i> , <b>2013</b> , 136, 3119-39	11.2	60
274	A RANKL-PKC $\beta$ -TFEB signaling cascade is necessary for lysosomal biogenesis in osteoclasts. <i>Genes and Development</i> , <b>2013</b> , 27, 955-69	12.6	118
273	Autophagy contributes to inflammation in patients with TNFR-associated periodic syndrome (TRAPS). <i>Annals of the Rheumatic Diseases</i> , <b>2013</b> , 72, 1044-52	2.4	62
272	New targets for old diseases: lessons from mucopolipidosis type II. <i>EMBO Molecular Medicine</i> , <b>2013</b> , 5, 1801-13	11.2	118
271	Gene transfer of master autophagy regulator TFEB results in clearance of toxic protein and correction of hepatic disease in alpha-1-anti-trypsin deficiency. <i>EMBO Molecular Medicine</i> , <b>2013</b> , 5, 397-412	11.2	116
270	A highly secreted sulphamidase engineered to cross the blood-brain barrier corrects brain lesions of mice with mucopolysaccharidoses type IIIA. <i>EMBO Molecular Medicine</i> , <b>2013</b> , 5, 675-90	12	86
269	What else is in store for autophagy? Exocytosis of autolysosomes as a mechanism of TFEB-mediated cellular clearance in Pompe disease. <i>Autophagy</i> , <b>2013</b> , 9, 1117-8	10.2	28
268	Autophagy master regulator TFEB induces clearance of toxic SERPINA1/ $\alpha$ 1-antitrypsin polymers. <i>Autophagy</i> , <b>2013</b> , 9, 1094-6	10.2	35

267	A lysosome-to-nucleus signalling mechanism senses and regulates the lysosome via mTOR and TFEB. <i>EMBO Journal</i> , <b>2012</b> , 31, 1095-108	13	1156
266	Identification of microRNA-regulated gene networks by expression analysis of target genes. <i>Genome Research</i> , <b>2012</b> , 22, 1163-72	9.7	134
265	Sulfatases are determinants of alveolar formation. <i>Matrix Biology</i> , <b>2012</b> , 31, 253-60	11.4	7
264	Guidelines for the use and interpretation of assays for monitoring autophagy. <i>Autophagy</i> , <b>2012</b> , 8, 445-544.2	11.2	2783
263	Autophagy in astrocytes: a novel culprit in lysosomal storage disorders. <i>Autophagy</i> , <b>2012</b> , 8, 1871-2	10.2	26
262	Autophagy in lysosomal storage disorders. <i>Autophagy</i> , <b>2012</b> , 8, 719-30	10.2	288
261	Astrocyte dysfunction triggers neurodegeneration in a lysosomal storage disorder. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2012</b> , 109, E2334-42	11.5	81
260	Impaired parkin-mediated mitochondrial targeting to autophagosomes differentially contributes to tissue pathology in lysosomal storage diseases. <i>Human Molecular Genetics</i> , <b>2012</b> , 21, 1770-81	5.6	42
259	TFEB regulates autophagy: an integrated coordination of cellular degradation and recycling processes. <i>Autophagy</i> , <b>2011</b> , 7, 1379-81	10.2	166
258	TFEB links autophagy to lysosomal biogenesis. <i>Science</i> , <b>2011</b> , 332, 1429-33	33.3	1865
257	Transcriptional activation of lysosomal exocytosis promotes cellular clearance. <i>Developmental Cell</i> , <b>2011</b> , 21, 421-30	10.2	458
256	Cystic fibrosis: a disorder with defective autophagy. <i>Autophagy</i> , <b>2011</b> , 7, 104-6	10.2	61
255	Systems medicine and integrated care to combat chronic noncommunicable diseases. <i>Genome Medicine</i> , <b>2011</b> , 3, 43	14.4	137
254	Characterization of the CLEAR network reveals an integrated control of cellular clearance pathways. <i>Human Molecular Genetics</i> , <b>2011</b> , 20, 3852-66	5.6	556
253	Efficacy of a combined intracerebral and systemic gene delivery approach for the treatment of a severe lysosomal storage disorder. <i>Molecular Therapy</i> , <b>2011</b> , 19, 860-9	11.7	35
252	A high-resolution anatomical atlas of the transcriptome in the mouse embryo. <i>PLoS Biology</i> , <b>2011</b> , 9, e1000582	9.7	467
251	Lysosomal fusion and SNARE function are impaired by cholesterol accumulation in lysosomal storage disorders. <i>EMBO Journal</i> , <b>2010</b> , 29, 3607-20	13	161
250	Defective CFTR induces aggresome formation and lung inflammation in cystic fibrosis through ROS-mediated autophagy inhibition. <i>Nature Cell Biology</i> , <b>2010</b> , 12, 863-75	23.4	367



249	Lack of sik1 in mouse embryonic stem cells impairs cardiomyogenesis by down-regulating the cyclin-dependent kinase inhibitor p57kip2. <i>PLoS ONE</i> , <b>2010</b> , 5, e9029	3.7	21
248	A mouse embryonic stem cell bank for inducible overexpression of human chromosome 21 genes. <i>Genome Biology</i> , <b>2010</b> , 11, R64	18.3	12
247	Self-eating in skeletal development: implications for lysosomal storage disorders. <i>Autophagy</i> , <b>2009</b> , 5, 228-9	10.2	12
246	Lysosomal disorders: from storage to cellular damage. <i>Biochimica Et Biophysica Acta - Molecular Cell Research</i> , <b>2009</b> , 1793, 684-96	4.9	380
245	A gene network regulating lysosomal biogenesis and function. <i>Science</i> , <b>2009</b> , 325, 473-7	33.3	1484
244	MicroRNA target prediction by expression analysis of host genes. <i>Genome Research</i> , <b>2009</b> , 19, 481-90	9.7	141
243	Disease pathogenesis explained by basic science: lysosomal storage diseases as autophagocytic disorders. <i>International Journal of Clinical Pharmacology and Therapeutics</i> , <b>2009</b> , 47 Suppl 1, S34-8	2	18
242	Genomic analysis of the TRIM family reveals two groups of genes with distinct evolutionary properties. <i>BMC Evolutionary Biology</i> , <b>2008</b> , 8, 225	3	190
241	Multiple sulfatase deficiency in a Turkish family resulting from a novel mutation. <i>Brain and Development</i> , <b>2008</b> , 30, 374-7	2.2	10
240	Lysosomal storage diseases as disorders of autophagy. <i>Autophagy</i> , <b>2008</b> , 4, 113-4	10.2	130
239	The ocular albinism type 1 protein, an intracellular G protein-coupled receptor, regulates melanosome transport in pigment cells. <i>Human Molecular Genetics</i> , <b>2008</b> , 17, 3487-501	5.6	68
238	A block of autophagy in lysosomal storage disorders. <i>Human Molecular Genetics</i> , <b>2008</b> , 17, 119-29	5.6	404
237	Multistep, sequential control of the trafficking and function of the multiple sulfatase deficiency gene product, SUMF1 by PDI, ERGIC-53 and ERp44. <i>Human Molecular Genetics</i> , <b>2008</b> , 17, 2610-21	5.6	54
236	Guidelines for the use and interpretation of assays for monitoring autophagy in higher eukaryotes. <i>Autophagy</i> , <b>2008</b> , 4, 151-75	10.2	1920
235	Proteoglycan desulfation determines the efficiency of chondrocyte autophagy and the extent of FGF signaling during endochondral ossification. <i>Genes and Development</i> , <b>2008</b> , 22, 2645-50	12.6	73
234	Safety of arylsulfatase A overexpression for gene therapy of metachromatic leukodystrophy. <i>Human Gene Therapy</i> , <b>2007</b> , 18, 821-36	4.8	41
233	Diversity, parental germline origin, and phenotypic spectrum of de novo HRAS missense changes in Costello syndrome. <i>Human Mutation</i> , <b>2007</b> , 28, 265-72	4.7	104
232	Multiple sulfatase deficiency is due to hypomorphic mutations of the SUMF1 gene. <i>Human Mutation</i> , <b>2007</b> , 28, 928	4.7	35

231	Sulfatase modifying factor 1 trafficking through the cells: from endoplasmic reticulum to the endoplasmic reticulum. <i>EMBO Journal</i> , <b>2007</b> , 26, 2443-53	13	39
230	Mutational analysis of the HGSNAT gene in Italian patients with mucopolysaccharidosis IIIC (Sanfilippo C syndrome). Mutation in brief #959. Online. <i>Human Mutation</i> , <b>2007</b> , 28, 523	4.7	47
229	Slc7a7 disruption causes fetal growth retardation by downregulating Igf1 in the mouse model of lysinuric protein intolerance. <i>American Journal of Physiology - Cell Physiology</i> , <b>2007</b> , 293, C191-8	5.4	38
228	Pharmacological enhancement of mutated alpha-glucosidase activity in fibroblasts from patients with Pompe disease. <i>Molecular Therapy</i> , <b>2007</b> , 15, 508-14	11.7	101
227	Functional correction of CNS lesions in an MPS-IIIA mouse model by intracerebral AAV-mediated delivery of sulfamidase and SUMF1 genes. <i>Human Molecular Genetics</i> , <b>2007</b> , 16, 2693-702	5.6	90
226	Systemic inflammation and neurodegeneration in a mouse model of multiple sulfatase deficiency. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2007</b> , 104, 4506-11	11.5	76
225	SUMF1 enhances sulfatase activities in vivo in five sulfatase deficiencies. <i>Biochemical Journal</i> , <b>2007</b> , 403, 305-12	3.8	60
224	New mutations identified in the ocular albinism type 1 gene. <i>Gene</i> , <b>2007</b> , 402, 20-7	3.8	5
223	Filamin A is mutated in X-linked chronic idiopathic intestinal pseudo-obstruction with central nervous system involvement. <i>American Journal of Human Genetics</i> , <b>2007</b> , 80, 751-8	11	85
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