Andrea Ballabio

List of Publications by Citations

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	Guidelines for the use and interpretation of assays for monitoring autophagy (3rd edition). *Autophagy*, **2016**, 12, 1-222	10.2	3838
373	Guidelines for the use and interpretation of assays for monitoring autophagy. *Autophagy*, **2012**, 8, 445-544	10.2	2783
372	Guidelines for the use and interpretation of assays for monitoring autophagy in higher eukaryotes. *Autophagy*, **2008**, 4, 151-75	10.2	1920
371	TFEB links autophagy to lysosomal biogenesis. *Science*, **2011**, 332, 1429-33	33.3	1865
370	A gene network regulating lysosomal biogenesis and function. *Science*, **2009**, 325, 473-7	33.3	1484
369	A gene from the region of the human X inactivation centre is expressed exclusively from the inactive X chromosome. *Nature*, **1991**, 349, 38-44	50.4	1164
368	A lysosome-to-nucleus signalling mechanism senses and regulates the lysosome via mTOR and TFEB. *EMBO Journal*, **2012**, 31, 1095-108	13	1156
367	Signals from the lysosome: a control centre for cellular clearance and energy metabolism. *Nature Reviews Molecular Cell Biology*, **2013**, 14, 283-96	48.7	1043
366	The tripartite motif family identifies cell compartments. *EMBO Journal*, **2001**, 20, 2140-51	13	979
365	Molecular definitions of autophagy and related processes. *EMBO Journal*, **2017**, 36, 1811-1836	13	857
364	The DNA sequence of the human X chromosome. *Nature*, **2005**, 434, 325-37	50.4	822
363	A gene deleted in Kallmann's syndrome shares homology with neural cell adhesion and axonal path-finding molecules. *Nature*, **1991**, 353, 529-36	50.4	764
362	Haploinsufficiency of ATP1A2 encoding the Na+/K+ pump alpha2 subunit associated with familial hemiplegic migraine type 2. *Nature Genetics*, **2003**, 33, 192-6	36.3	755
361	Lysosomal calcium signalling regulates autophagy through calcineurin and TFEB. *Nature Cell Biology*, **2015**, 17, 288-99	23.4	716
360	Spastic paraplegia and OXPHOS impairment caused by mutations in paraplegin, a nuclear-encoded mitochondrial metalloprotease. *Cell*, **1998**, 93, 973-83	56.2	705
359	TFEB controls cellular lipid metabolism through a starvation-induced autoregulatory loop. *Nature Cell Biology*, **2013**, 15, 647-58	23.4	599
358	Characterization of the CLEAR network reveals an integrated control of cellular clearance pathways. *Human Molecular Genetics*, **2011**, 20, 3852-66	5.6	556

357	Characterization of a murine gene expressed from the inactive X chromosome. *Nature*, **1991**, 351, 325-9	50.4	471
356	A high-resolution anatomical atlas of the transcriptome in the mouse embryo. *PLoS Biology*, **2011**, 9, e1000582	9.7	467
355	Transcriptional activation of lysosomal exocytosis promotes cellular clearance. *Developmental Cell*, **2011**, 21, 421-30	10.2	458
354	Guidelines for the use and interpretation of assays for monitoring autophagy (4th edition). *Autophagy*, **2021**, 17, 1-382	10.2	440
353	A block of autophagy in lysosomal storage disorders. *Human Molecular Genetics*, **2008**, 17, 119-29	5.6	404
352	The nicotinic receptor beta 2 subunit is mutant in nocturnal frontal lobe epilepsy. *Nature Genetics*, **2000**, 26, 275-6	36.3	386
351	Lysosomal disorders: from storage to cellular damage. *Biochimica Et Biophysica Acta - Molecular Cell Research*, **2009**, 1793, 684-96	4.9	380
350	Defective CFTR induces aggresome formation and lung inflammation in cystic fibrosis through ROS-mediated autophagy inhibition. *Nature Cell Biology*, **2010**, 12, 863-75	23.4	367
349	TFEB at a glance. *Journal of Cell Science*, **2016**, 129, 2475-81	5.3	350
348	Localization of the X inactivation centre on the human X chromosome in Xq13. *Nature*, **1991**, 349, 82-4	50.4	338
347	Lysosomes as dynamic regulators of cell and organismal homeostasis. *Nature Reviews Molecular Cell Biology*, **2020**, 21, 101-118	48.7	310
346	Opitz G/BBB syndrome, a defect of midline development, is due to mutations in a new RING finger gene on Xp22. *Nature Genetics*, **1997**, 17, 285-91	36.3	296
345	Autophagy in lysosomal storage disorders. *Autophagy*, **2012**, 8, 719-30	10.2	288
344	The multiple sulfatase deficiency gene encodes an essential and limiting factor for the activity of sulfatases. *Cell*, **2003**, 113, 445-56	56.2	281
343	A cluster of sulfatase genes on Xp22.3: mutations in chondrodysplasia punctata (CDPX) and implications for warfarin embryopathy. *Cell*, **1995**, 81, 15-25	56.2	267
342	Identification of the gene for oral-facial-digital type I syndrome. *American Journal of Human Genetics*, **2001**, 68, 569-76	11	258
341	Spastin, the protein mutated in autosomal dominant hereditary spastic paraplegia, is involved in microtubule dynamics. *Human Molecular Genetics*, **2002**, 11, 153-63	5.6	240
340	Lysosomal storage diseases: from pathophysiology to therapy. *Annual Review of Medicine*, **2015**, 66, 471-86	7.4	239

339	Non-type I cystinuria caused by mutations in SLC7A9, encoding a subunit (bo,+AT) of rBAT. *Nature Genetics*, **1999**, 23, 52-7	36.3	232
338	Selective clearance of aberrant tau proteins and rescue of neurotoxicity by transcription factor EB. *EMBO Molecular Medicine*, **2014**, 6, 1142-60	12	227
337	Contiguous gene syndromes due to deletions in the distal short arm of the human X chromosome. *Proceedings of the National Academy of Sciences of the United States of America*, **1989**, 86, 10001-5	11.5	226
336	Transcription factor EB (TFEB) is a new therapeutic target for Pompe disease. *EMBO Molecular Medicine*, **2013**, 5, 691-706	12	224
335	Axonal degeneration in paraplegin-deficient mice is associated with abnormal mitochondria and impairment of axonal transport. *Journal of Clinical Investigation*, **2004**, 113, 231-242	15.9	220
334	Loss of m-AAA protease in mitochondria causes complex I deficiency and increased sensitivity to oxidative stress in hereditary spastic paraplegia. *Journal of Cell Biology*, **2003**, 163, 777-87	7.3	212
333	A human homologue of the Drosophila melanogaster diaphanous gene is disrupted in a patient with premature ovarian failure: evidence for conserved function in oogenesis and implications for human sterility. *American Journal of Human Genetics*, **1998**, 62, 533-41	11	209
332	Cloning of the gene encoding a novel integral membrane protein, mucolipidin-and identification of the two major founder mutations causing mucolipidosis type IV. *American Journal of Human Genetics*, **2000**, 67, 1110-20	11	209
331	Transcriptional and epigenetic regulation of autophagy in aging. *Autophagy*, **2015**, 11, 867-80	10.2	207
330	SLC7A7, encoding a putative permease-related protein, is mutated in patients with lysinuric protein intolerance. *Nature Genetics*, **1999**, 21, 297-301	36.3	206
329	The Autophagy-Lysosomal Pathway in Neurodegeneration: A TFEB Perspective. *Trends in Neurosciences*, **2016**, 39, 221-234	13.3	195
328	The complex relationship between TFEB transcription factor phosphorylation and subcellular localization. *EMBO Journal*, **2018**, 37,	13	193
327	Genomic analysis of the TRIM family reveals two groups of genes with distinct evolutionary properties. *BMC Evolutionary Biology*, **2008**, 8, 225	3	190
326	Exploiting macrophage autophagy-lysosomal biogenesis as a therapy for atherosclerosis. *Nature Communications*, **2017**, 8, 15750	17.4	188
325	Functional implications of the spectrum of mutations found in 234 cases with X-linked juvenile retinoschisis. The Retinoschisis Consortium. *Human Molecular Genetics*, **1998**, 7, 1185-92	5.6	182
324	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. *Human Gene Therapy*, **2014**, 25, 506-16	4.8	177
323	Human chromosome 21 gene expression atlas in the mouse. *Nature*, **2002**, 420, 582-6	50.4	177
322	The European dimension for the mouse genome mutagenesis program. *Nature Genetics*, **2004**, 36, 925-7	36.3	176

321	Cloning of the gene for ocular albinism type 1 from the distal short arm of the X chromosome. *Nature Genetics*, **1995**, 10, 13-9	36.3	171
320	Transcription Factor EB Controls Metabolic Flexibility during Exercise. *Cell Metabolism*, **2017**, 25, 182-196	24.6	169
319	TFEB regulates autophagy: an integrated coordination of cellular degradation and recycling processes. *Autophagy*, **2011**, 7, 1379-81	10.2	166
318	Brief report: intragenic deletion of the KALIG-1 gene in Kallmann's syndrome. *New England Journal of Medicine*, **1992**, 326, 1752-5	59.2	165
317	Identification and mapping of human cDNAs homologous to Drosophila mutant genes through EST database searching. *Nature Genetics*, **1996**, 13, 167-74	36.3	163
316	Lysosomal fusion and SNARE function are impaired by cholesterol accumulation in lysosomal storage disorders. *EMBO Journal*, **2010**, 29, 3607-20	13	161
315	TFEB and TFE3 cooperate in the regulation of the innate immune response in activated macrophages. *Autophagy*, **2016**, 12, 1240-58	10.2	150
314	Induction of lysosomal biogenesis in atherosclerotic macrophages can rescue lipid-induced lysosomal dysfunction and downstream sequelae. *Arteriosclerosis, Thrombosis, and Vascular Biology*, **2014**, 34, 1942-1952	9.4	147
313	Deletions and translocations involving the distal short arm of the human X chromosome: review and hypotheses. *Human Molecular Genetics*, **1992**, 1, 221-7	5.6	147
312	Wilson disease protein ATP7B utilizes lysosomal exocytosis to maintain copper homeostasis. *Developmental Cell*, **2014**, 29, 686-700	10.2	146
311	Expression pattern of the Kallmann syndrome gene in the olfactory system suggests a role in neuronal targeting. *Nature Genetics*, **1993**, 4, 19-26	36.3	145
310	MicroRNA target prediction by expression analysis of host genes. *Genome Research*, **2009**, 19, 481-90	9.7	141
309	The sulfatase gene family. *Current Opinion in Genetics and Development*, **1997**, 7, 386-91	4.9	139
308	Systems medicine and integrated care to combat chronic noncommunicable diseases. *Genome Medicine*, **2011**, 3, 43	14.4	137
307	Sulfatases and human disease. *Annual Review of Genomics and Human Genetics*, **2005**, 6, 355-79	9.7	137
306	Autosomal recessive rolandic epilepsy with paroxysmal exercise-induced dystonia and writer's cramp: delineation of the syndrome and gene mapping to chromosome 16p12-11.2. *Annals of Neurology*, **1999**, 45, 344-52	9.4	136
305	Neuronal-Targeted TFEB Accelerates Lysosomal Degradation of APP, Reducing A Generation and Amyloid Plaque Pathogenesis. *Journal of Neuroscience*, **2015**, 35, 12137-51	6.6	134
304	Identification of microRNA-regulated gene networks by expression analysis of target genes. *Genome Research*, **2012**, 22, 1163-72	9.7	134

303	mTOR-dependent phosphorylation controls TFEB nuclear export. *Nature Communications*, **2018**, 9, 3312	17.4	133
302	A new locus for autosomal recessive hereditary spastic paraplegia maps to chromosome 16q24.3. *American Journal of Human Genetics*, **1998**, 63, 135-9	11	133
301	Sulfatases and sulfatase modifying factors: an exclusive and promiscuous relationship. *Human Molecular Genetics*, **2005**, 14, 3203-17	5.6	131
300	Lysosomal storage diseases as disorders of autophagy. *Autophagy*, **2008**, 4, 113-4	10.2	130
299	Lysosome: regulator of lipid degradation pathways. *Trends in Cell Biology*, **2014**, 24, 743-50	18.3	129
298	Identification and expression of NEU3, a novel human sialidase associated to the plasma membrane. *Biochemical Journal*, **2000**, 349, 343-351	3.8	129
297	A homeobox gene, vax2, controls the patterning of the eye dorsoventral axis. *Proceedings of the National Academy of Sciences of the United States of America*, **1999**, 96, 10729-34	11.5	129
296	Expression pattern of the Tbr2 (Eomesodermin) gene during mouse and chick brain development. *Mechanisms of Development*, **1999**, 84, 133-8	1.7	129
295	Impaired TFEB-Mediated Lysosome Biogenesis and Autophagy Promote Chronic Ethanol-Induced Liver Injury and Steatosis in Mice. *Gastroenterology*, **2018**, 155, 865-879.e12	13.3	123
294	Mapping a gene for familial situs abnormalities to human chromosome Xq24-q27.1. *Nature Genetics* , **1993**, 5, 403-7	36.3	122
293	EYA4, a novel vertebrate gene related to Drosophila eyes absent. *Human Molecular Genetics*, **1999**, 8, 11-23	5.6	120
292	A RANKL-PKC-TFEB signaling cascade is necessary for lysosomal biogenesis in osteoclasts. *Genes and Development*, **2013**, 27, 955-69	12.6	118
291	Isolation and characterization of a steroid sulfatase cDNA clone: genomic deletions in patients with X-chromosome-linked ichthyosis. *Proceedings of the National Academy of Sciences of the United States of America*, **1987**, 84, 4519-23	11.5	118
290	Gene transfer of master autophagy regulator TFEB results in clearance of toxic protein and correction of hepatic disease in alpha-1-anti-trypsin deficiency. *EMBO Molecular Medicine*, **2013**, 5, 397-412	12	116
289	Functional analysis of mutations in SLC7A9, and genotype-phenotype correlation in non-Type I cystinuria. *Human Molecular Genetics*, **2001**, 10, 305-16	5.6	115
288	Brain tyrosinase overexpression implicates age-dependent neuromelanin production in Parkinson's disease pathogenesis. *Nature Communications*, **2019**, 10, 973	17.4	114
287	A deletion map of the human Yq11 region: implications for the evolution of the Y chromosome and tentative mapping of a locus involved in spermatogenesis. *Genomics*, **1991**, 11, 443-51	4.3	112
286	Axonal degeneration in paraplegin-deficient mice is associated with abnormal mitochondria and impairment of axonal transport. *Journal of Clinical Investigation*, **2004**, 113, 231-42	15.9	112

285	Molecular cloning, expression pattern, and chromosomal localization of the human Na-Cl thiazide-sensitive cotransporter (SLC12A3). *Genomics*, **1996**, 35, 486-93	4.3	110
284	Ocular albinism: evidence for a defect in an intracellular signal transduction system. *Nature Genetics* , **1999**, 23, 108-12	36.3	107
283	Transcriptional activation of RagD GTPase controls mTORC1 and promotes cancer growth. *Science*, **2017**, 356, 1188-1192	33.3	106
282	FACL4, a new gene encoding long-chain acyl-CoA synthetase 4, is deleted in a family with Alport syndrome, elliptocytosis, and mental retardation. *Genomics*, **1998**, 47, 350-8	4.3	106
281	The awesome lysosome. *EMBO Molecular Medicine*, **2016**, 8, 73-6	12	105
280	Diversity, parental germline origin, and phenotypic spectrum of de novo HRAS missense changes in Costello syndrome. *Human Mutation*, **2007**, 28, 265-72	4.7	104
279	Identification and characterization of a novel serine-threonine kinase gene from the Xp22 region. *Genomics*, **1998**, 51, 427-33	4.3	103
278	Autophagosome-lysosome fusion triggers a lysosomal response mediated by TLR9 and controlled by OCRL. *Nature Cell Biology*, **2016**, 18, 839-850	23.4	103
277	Rox, a novel bHLHZip protein expressed in quiescent cells that heterodimerizes with Max, binds a non-canonical E box and acts as a transcriptional repressor. *EMBO Journal*, **1997**, 16, 2892-906	13	101
276	Pharmacological enhancement of mutated alpha-glucosidase activity in fibroblasts from patients with Pompe disease. *Molecular Therapy*, **2007**, 15, 508-14	11.7	101
275	A gene from the Xp22.3 region shares homology with voltage-gated chloride channels. *Human Molecular Genetics*, **1994**, 3, 547-52	5.6	99
274	Mutations of the mitochondrial holocytochrome c-type synthase in X-linked dominant microphthalmia with linear skin defects syndrome. *American Journal of Human Genetics*, **2006**, 79, 878-89	11	98
273	Functional characterization of the Opitz syndrome gene product (midin): evidence for homodimerization and association with microtubules throughout the cell cycle. *Human Molecular Genetics*, **1999**, 8, 1387-96	5.6	97
272	A novel curcumin analog binds to and activates TFEB in vitro and in vivo independent of MTOR inhibition. *Autophagy*, **2016**, 12, 1372-89	10.2	97
271	STUB1 regulates TFEB-induced autophagy-lysosome pathway. *EMBO Journal*, **2017**, 36, 2544-2552	13	96
270	Molecular cloning and characterization of NEU4, the fourth member of the human sialidase gene family. *Genomics*, **2004**, 83, 445-53	4.3	95
269	LINE-1 elements at the sites of molecular rearrangements in Alport syndrome-diffuse leiomyomatosis. *American Journal of Human Genetics*, **1999**, 64, 62-9	11	95
268	Novel molecular variants of the Na-Cl cotransporter gene are responsible for Gitelman syndrome. *American Journal of Human Genetics*, **1996**, 59, 1019-26	11	95

267	Isolation of Crb1, a mouse homologue of Drosophila crumbs, and analysis of its expression pattern in eye and brain. *Mechanisms of Development*, **2002**, 110, 203-7	1.7	94
266	A high resolution deletion map of human chromosome Xp22. *Nature Genetics*, **1993**, 4, 272-9	36.3	94
265	Molecular heterogeneity of steroid sulfatase deficiency: a multicenter study on 57 unrelated patients, at DNA and protein levels. *Genomics*, **1989**, 4, 36-40	4.3	94
264	Polyamines Control eIF5A Hypusination, TFEB Translation, and Autophagy to Reverse B Cell Senescence. *Molecular Cell*, **2019**, 76, 110-125.e9	17.6	93
263	WBSCR14, a gene mapping to the Williams--Beuren syndrome deleted region, is a new member of the Mlx transcription factor network. *Human Molecular Genetics*, **2001**, 10, 617-27	5.6	93
262	Microphthalmia with linear skin defects (MLS) syndrome: clinical, cytogenetic, and molecular characterization. *American Journal of Medical Genetics Part A*, **1994**, 49, 229-34		92
261	X-linked late-onset sensorineural deafness caused by a deletion involving OA1 and a novel gene containing WD-40 repeats. *American Journal of Human Genetics*, **1999**, 64, 1604-16	11	91
260	Functional correction of CNS lesions in an MPS-IIIA mouse model by intracerebral AAV-mediated delivery of sulfamidase and SUMF1 genes. *Human Molecular Genetics*, **2007**, 16, 2693-702	5.6	90
259	A member of a gene family on Xp22.3, VCX-A, is deleted in patients with X-linked nonspecific mental retardation. *American Journal of Human Genetics*, **2000**, 67, 563-73	11	89
258	Identification of a new locus for medullary cystic disease, on chromosome 16p12. *American Journal of Human Genetics*, **1999**, 64, 1655-60	11	89
257	Endothelin-B receptor mutations in patients with isolated Hirschsprung disease from a non-inbred population. *Human Molecular Genetics*, **1996**, 5, 351-4	5.6	89
256	Vax2inactivation in mouse determines alteration of the eye dorsal-ventral axis, misrouting of the optic fibres and eye coloboma. *Development (Cambridge)*, **2002**, 129, 805-813	6.6	89
255	Cloning and characterization of NEU2, a human gene homologous to rodent soluble sialidases. *Genomics*, **1999**, 57, 137-43	4.3	88
254	The phytoestrogen genistein modulates lysosomal metabolism and transcription factor EB (TFEB) activation. *Journal of Biological Chemistry*, **2014**, 289, 17054-69	5.4	87
253	Identification and expression of NEU3, a novel human sialidase associated to the plasma membrane. *Biochemical Journal*, **2000**, 349, 343-51	3.8	87
252	A highly secreted sulphamidase engineered to cross the blood-brain barrier corrects brain lesions of mice with mucopolysaccharidoses type IIIA. *EMBO Molecular Medicine*, **2013**, 5, 675-90	12	86
251	Filamin A is mutated in X-linked chronic idiopathic intestinal pseudo-obstruction with central nervous system involvement. *American Journal of Human Genetics*, **2007**, 80, 751-8	11	85
250	Barhl1, a gene belonging to a new subfamily of mammalian homeobox genes, is expressed in migrating neurons of the CNS. *Human Molecular Genetics*, **2000**, 9, 1443-52	5.6	84

249	X-linked ichthyosis, due to steroid sulphatase deficiency, associated with Kallmann syndrome (hypogonadotropic hypogonadism and anosmia): linkage relationships with Xg and cloned DNA sequences from the distal short arm of the X chromosome. *Human Genetics*, **1986**, 72, 237-40	6.3	84
248	Double heterozygosity for a RET substitution interfering with splicing and an EDNRB missense mutation in Hirschsprung disease. *American Journal of Human Genetics*, **1999**, 64, 1216-21	11	83
247	Astrocyte dysfunction triggers neurodegeneration in a lysosomal storage disorder. *Proceedings of the National Academy of Sciences of the United States of America*, **2012**, 109, E2334-42	11.5	81
246	The ocular albinism type 1 gene product is a membrane glycoprotein localized to melanosomes. *Proceedings of the National Academy of Sciences of the United States of America*, **1996**, 93, 9055-60	11.5	81
245	PCR test for cystic fibrosis deletion. *Nature*, **1990**, 343, 220	50.4	79
244	Autophagy in major human diseases. *EMBO Journal*, **2021**, 40, e108863	13	79
243	Familial hemiplegic migraine type 2 is linked to 0.9Mb region on chromosome 1q23. *Annals of Neurology*, **2003**, 53, 376-81	9.4	77
242	Systemic inflammation and neurodegeneration in a mouse model of multiple sulfatase deficiency. *Proceedings of the National Academy of Sciences of the United States of America*, **2007**, 104, 4506-11	11.5	76
241	X-linked Opitz syndrome: novel mutations in the MID1 gene and redefinition of the clinical spectrum **2003**, 120A, 222-8		76
240	Dysregulation of Nutrient Sensing and CLEARance in Presenilin Deficiency. *Cell Reports*, **2016**, 14, 2166-2179	10.0	75
239	Correction of Hunter syndrome in the MPSII mouse model by AAV2/8-mediated gene delivery. *Human Molecular Genetics*, **2006**, 15, 1225-36	5.6	74
238	A substrate-specific mTORC1 pathway underlies Birt-Hogg-Dub syndrome. *Nature*, **2020**, 585, 597-602	50.4	73
237	Proteoglycan desulfation determines the efficiency of chondrocyte autophagy and the extent of FGF signaling during endochondral ossification. *Genes and Development*, **2008**, 22, 2645-50	12.6	73
236	A new locus for autosomal dominant nocturnal frontal lobe epilepsy maps to chromosome 1. *Neurology*, **2000**, 55, 1467-71	6.5	73
235	Oa1 knock-out: new insights on the pathogenesis of ocular albinism type 1. *Human Molecular Genetics*, **2000**, 9, 2781-8	5.6	73
234	Analysis of the OA1 gene reveals mutations in only one-third of patients with X-linked ocular albinism. *Human Molecular Genetics*, **1995**, 4, 2319-25	5.6	73
233	An integrated physical and genetic map of a 35 Mb region on chromosome Xp22.3-Xp21.3. *Human Molecular Genetics*, **1995**, 4, 1821-7	5.6	72
232	Lysosomal adaptation: how the lysosome responds to external cues. *Cold Spring Harbor Perspectives in Biology*, **2014**, 6,	10.2	71

231	Mammalian X-chromosome inactivation and the XIST gene. *Current Opinion in Genetics and Development*, **1992**, 2, 439-47	4.9	71
230	Characterization of a human and murine gene (CLCN3) sharing similarities to voltage-gated chloride channels and to a yeast integral membrane protein. *Genomics*, **1995**, 27, 131-41	4.3	70
229	Evidence of evolutionary up-regulation of the single active X chromosome in mammals based on Clc4 expression levels in Mus spretus and Mus musculus. *Proceedings of the National Academy of Sciences of the United States of America*, **1997**, 94, 9244-8	11.5	69
228	A submicroscopic deletion in Xq26 associated with familial situs ambiguus. *American Journal of Human Genetics*, **1997**, 61, 395-401	11	68
227	The ocular albinism type 1 protein, an intracellular G protein-coupled receptor, regulates melanosome transport in pigment cells. *Human Molecular Genetics*, **2008**, 17, 3487-501	5.6	68
226	Two families of low-copy-number repeats are interspersed on Xp22.3: implications for the high frequency of deletions in this region. *Genomics*, **1990**, 8, 263-70	4.3	68
225	X-linked Alport syndrome: an SSCP-based mutation survey over all 51 exons of the COL4A5 gene. *American Journal of Human Genetics*, **1996**, 58, 1192-204	11	68
224	Evidence for interaction between human PRUNE and nm23-H1 NDPKinase. *Oncogene*, **1999**, 18, 7244-52	9.2	67
223	Fasting Imparts a Switch to Alternative Daily Pathways in Liver and Muscle. *Cell Reports*, **2018**, 25, 3299-3314.e6	8.8	67
222	Different chromosomal localization of the Clcn4 gene in Mus spretus and C57BL/6J mice. *Nature Genetics*, **1995**, 10, 466-71	36.3	66
221	Identification and characterization of AFG3L2, a novel paraplegin-related gene. *Genomics*, **1999**, 59, 51-8	4.3	65
220	Lysosome signaling controls the migration of dendritic cells. *Science Immunology*, **2017**, 2,	28	64
219	X-inactivation and human disease: X-linked dominant male-lethal disorders. *Current Opinion in Genetics and Development*, **2006**, 16, 254-9	4.9	64
218	Mlx, a new Max-like bHLHZip family member: the center stage of a novel transcription factors regulatory pathway?. *Oncogene*, **2000**, 19, 3266-77	9.2	64
217	Lysosomal calcium regulates autophagy. *Autophagy*, **2015**, 11, 970-1	10.2	63
216	Structure of the SLC7A7 gene and mutational analysis of patients affected by lysinuric protein intolerance. *American Journal of Human Genetics*, **2000**, 66, 92-9	11	63
215	Activation of the transcription factor EB rescues lysosomal abnormalities in cystinotic kidney cells. *Kidney International*, **2016**, 89, 862-73	9.9	62
214	Autophagy contributes to inflammation in patients with TNFR-associated periodic syndrome (TRAPS). *Annals of the Rheumatic Diseases*, **2013**, 72, 1044-52	2.4	62

213	The locus for a novel syndromic form of neuronal intestinal pseudoobstruction maps to Xq28. *American Journal of Human Genetics*, **1996**, 58, 743-8	11	62
212	Cystic fibrosis: a disorder with defective autophagy. *Autophagy*, **2011**, 7, 104-6	10.2	61
211	Defective autophagy in spastizin mutated patients with hereditary spastic paraparesis type 15. *Brain*, **2013**, 136, 3119-39	11.2	60
210	SUMF1 enhances sulfatase activities in vivo in five sulfatase deficiencies. *Biochemical Journal*, **2007**, 403, 305-12	3.8	60
209	The Kallmann syndrome gene homolog in C. elegans is involved in epidermal morphogenesis and neurite branching. *Development (Cambridge)*, **2002**, 129, 1283-1294	6.6	60
208	Opitz G/BBB syndrome in Xp22: mutations in the MID1 gene cluster in the carboxy-terminal domain. *American Journal of Human Genetics*, **1998**, 63, 703-10	11	58
207	mTORC1 hyperactivation arrests bone growth in lysosomal storage disorders by suppressing autophagy. *Journal of Clinical Investigation*, **2017**, 127, 3717-3729	15.9	58
206	TRPML1 links lysosomal calcium to autophagosome biogenesis through the activation of the CaMKK/VPS34 pathway. *Nature Communications*, **2019**, 10, 5630	17.4	58
205	Molecular and functional analysis of SUMF1 mutations in multiple sulfatase deficiency. *Human Mutation*, **2004**, 23, 576-81	4.7	57
204	TFE3 regulates whole-body energy metabolism in cooperation with TFEB. *EMBO Molecular Medicine* , **2017**, 9, 605-621	12	56
203	Cloning and characterization of a putative human holocytochrome c-type synthetase gene (HCCS) isolated from the critical region for microphthalmia with linear skin defects (MLS). *Genomics*, **1996**, 34, 166-72	4.3	56
202	The microphthalmia transcription factor (Mitf) controls expression of the ocular albinism type 1 gene: link between melanin synthesis and melanosome biogenesis. *Molecular and Cellular Biology*, **2004**, 24, 6550-9	4.8	55
201	Defective intracellular transport and processing of OA1 is a major cause of ocular albinism type 1. *Human Molecular Genetics*, **2000**, 9, 3011-8	5.6	55
200	Multistep, sequential control of the trafficking and function of the multiple sulfatase deficiency gene product, SUMF1 by PDI, ERGIC-53 and ERp44. *Human Molecular Genetics*, **2008**, 17, 2610-21	5.6	54
199	Definitive localization of X-linked Kallman syndrome (hypogonadotropic hypogonadism and anosmia) to Xp22.3: close linkage to the hypervariable repeat sequence CRI-S232. *American Journal of Human Genetics*, **1990**, 47, 664-9	11	54
198	Drosophila Mitf regulates the V-ATPase and the lysosomal-autophagic pathway. *Autophagy*, **2016**, 12, 484-98	10.2	53
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