# Raphael Schiffmann

## List of Publications by Year in Descending Order



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| 205 papers | 13,095 citations | 67 h-index | 109 g-index |
| 230 ext. papers | 14,852 ext. citations | 6.7 avg, IF | 6.03 L-index |

| # | Paper | IF | Citations |
|---|---|---|---|
| 205 | Investigation of a dysmorphic facial phenotype in patients with Gaucher disease types 2 and 3. *Molecular Genetics and Metabolism*, **2021**, 134, 274-280 | 3.7 | 1 |
| 204 | Migalastat Tissue Distribution: Extrapolation From Mice to Humans Using Pharmacokinetic Modeling and Comparison With Agalsidase Beta Tissue Distribution in Mice. *Clinical Pharmacology in Drug Development*, **2021**, 10, 1075-1088 | 2.3 | 1 |
| 203 | White matter abnormalities and iron deposition in prenatal mucolipidosis IV- fetal imaging and pathology. *Metabolic Brain Disease*, **2021**, 36, 2155-2167 | 3.9 | 0 |
| 202 | Brain pathology and cerebellar purkinje cell loss in a mouse model of chronic neuronopathic Gaucher disease. *Progress in Neurobiology*, **2021**, 197, 101939 | 10.9 | 0 |
| 201 | Cerebral Microangiopathy in Leukoencephalopathy With Cerebral Calcifications and Cysts: A Pathological Description. *Journal of Child Neurology*, **2021**, 36, 133-140 | 2.5 | 0 |
| 200 | Endocrine and Growth Abnormalities in 4H Leukodystrophy Caused by Variants in POLR3A, POLR3B, and POLR1C. *Journal of Clinical Endocrinology and Metabolism*, **2021**, 106, e660-e674 | 5.6 | 9 |
| 199 | Assessment of plasma lyso-Gb for clinical monitoring of treatment response in migalastat-treated patients with Fabry disease. *Genetics in Medicine*, **2021**, 23, 192-201 | 8.1 | 6 |
| 198 | Expanded phenotype of AARS1-related white matter disease. *Genetics in Medicine*, **2021**, 23, 2352-2359 | 8.1 | 2 |
| 197 | Long-term follow-up of renal function in patients treated with migalastat for Fabry disease. *Molecular Genetics and Metabolism Reports*, **2021**, 28, 100786 | 1.8 | 3 |
| 196 | Early indicators of disease progression in Fabry disease that may indicate the need for disease-specific treatment initiation: findings from the opinion-based PREDICT-FD modified Delphi consensus initiative. *BMJ Open*, **2020**, 10, e035182 | 3 | 8 |
| 195 | Randomized Clinical Trial of First-Line Genome Sequencing in Pediatric White Matter Disorders. *Annals of Neurology*, **2020**, 88, 264-273 | 9.4 | 7 |
| 194 | Gaucher disease□euronopathic forms **2020**, 439-449 | | |
| 193 | Genome sequencing in persistently unsolved white matter disorders. *Annals of Clinical and Translational Neurology*, **2020**, 7, 144-152 | 5.3 | 13 |
| 192 | EEG abnormalities in patients with chronic neuronopathic Gaucher disease: A retrospective review. *Molecular Genetics and Metabolism*, **2020**, 131, 358-363 | 3.7 | 4 |
| 191 | Assessing the role of glycosphingolipids in the phenotype severity of Fabry disease mouse model. *Journal of Lipid Research*, **2020**, 61, 1410-1423 | 6.3 | 3 |
| 190 | Fabry disease genotype, phenotype, and migalastat amenability: Insights from a national cohort. *Journal of Inherited Metabolic Disease*, **2020**, 43, 326-333 | 5.4 | 10 |
| 189 | The definition of neuronopathic Gaucher disease. *Journal of Inherited Metabolic Disease*, **2020**, 43, 1056-1059 | 5.4 | 28 |

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| 188 | The glycosylation design space for recombinant lysosomal replacement enzymes produced in CHO cells. *Nature Communications*, **2019**, 10, 1785 | 17.4 | 32 |
| 187 | Variants Cause Spastic Paraplegia Associated with Cerebral Hypomyelination. *American Journal of Neuroradiology*, **2019**, 40, 788-791 | 4.4 | 2 |
| 186 | Leukodystrophy-associated mutations down-regulate the RNA polymerase III transcript and important regulatory RNA. *Journal of Biological Chemistry*, **2019**, 294, 7445-7459 | 5.4 | 22 |
| 185 | Pegunigalsidase alfa, a novel PEGylated enzyme replacement therapy for Fabry disease, provides sustained plasma concentrations and favorable pharmacodynamics: A 1-year Phase 1/2 clinical trial. *Journal of Inherited Metabolic Disease*, **2019**, 42, 534-544 | 5.4 | 38 |
| 184 | Bi-allelic CSF1R Mutations Cause Skeletal Dysplasia of Dysosteosclerosis-Pyle Disease Spectrum and Degenerative Encephalopathy with Brain Malformation. *American Journal of Human Genetics*, **2019**, 104, 925-935 | 11 | 56 |
| 183 | Once every 4 weeks - 2 mg/kg of pegunigalsidase alfa for treating Fabry disease Preliminary results of a phase 3 study. *Molecular Genetics and Metabolism*, **2019**, 126, S73 | 3.7 | 4 |
| 182 | Venglustat in adult Gaucher disease type 3: Preliminary safety, pharmacology, and exploratory efficacy from a phase 2 trial in combination with imiglucerase (LEAP). *Molecular Genetics and Metabolism*, **2019**, 126, S131 | 3.7 | 5 |
| 181 | The migalastat GLP-HEK assay is the gold standard for determining amenability in patients with Fabry disease. *Molecular Genetics and Metabolism Reports*, **2019**, 20, 100494 | 1.8 | 4 |
| 180 | Symptoms and Quality of Life in Patients with Fabry Disease: Results from an International Patient Survey. *Advances in Therapy*, **2019**, 36, 2866-2880 | 4.1 | 6 |
| 179 | Diagnosis, prognosis, and treatment of leukodystrophies. *Lancet Neurology, The*, **2019**, 18, 962-972 | 24.1 | 48 |
| 178 | Variation in cognitive function over time in Gaucher disease type 3. *Neurology*, **2019**, 93, e2272-e2283 | 6.5 | 10 |
| 177 | Unique molecular signature in mucolipidosis type IV microglia. *Journal of Neuroinflammation*, **2019**, 16, 276 | 10.1 | 10 |
| 176 | Dystonia in RNA Polymerase III-Related Leukodystrophy. *Movement Disorders Clinical Practice*, **2019** , 6, 155-159 | 2.2 | 7 |
| 175 | Health-Related Quality of Life for Patients With Genetically Determined Leukoencephalopathy. *Pediatric Neurology*, **2018**, 84, 21-26 | 2.9 | 4 |
| 174 | Migalastat improves diarrhea in patients with Fabry disease: clinical-biomarker correlations from the phase 3 FACETS trial. *Orphanet Journal of Rare Diseases*, **2018**, 13, 68 | 4.2 | 16 |
| 173 | Low frequency of Fabry disease in patients with common heart disease. *Genetics in Medicine*, **2018**, 20, 754-759 | 8.1 | 4 |
| 172 | Prognostic value of urinary 11-dehydro-thromboxane B for mortality: A cohort study of stable coronary artery disease patients treated with aspirin. *Catheterization and Cardiovascular Interventions*, **2018**, 92, 653-658 | 2.7 | 2 |
| 171 | Priapism in a Fabry disease mouse model is associated with upregulated penile nNOS and eNOS expression. *Journal of Inherited Metabolic Disease*, **2018**, 41, 231-238 | 5.4 | 4 |

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| 170 | A double-blind, placebo-controlled trial of triheptanoin in adult polyglucosan body disease and open-label, long-term outcome. *Journal of Inherited Metabolic Disease*, **2018**, 41, 877-883 | 5.4 | 9 |
| 169 | Migalastat for the treatment of Fabry disease. *Expert Opinion on Orphan Drugs*, **2018**, 6, 301-309 | 1.1 | 7 |
| 168 | Roscoe Owen Brady, MD: Remembrances of co-investigators and colleagues. *Molecular Genetics and Metabolism*, **2017**, 120, 1-7 | 3.7 | 2 |
| 167 | Urinary 11-Dehydro-Thromboxane B and Mortality in Patients With Stable Coronary Artery Disease. *American Journal of Cardiology*, **2017**, 119, 972-977 | 3 | 13 |
| 166 | Oral pharmacological chaperone migalastat compared with enzyme replacement therapy in Fabry disease: 18-month results from the randomised phase III ATTRACT study. *Journal of Medical Genetics*, **2017**, 54, 288-296 | 5.8 | 193 |
| 165 | Residual thromboxane activity and oxidative stress: influence on mortality in patients with stable coronary artery disease. *Coronary Artery Disease*, **2017**, 28, 287-293 | 1.4 | 6 |
| 164 | Tetrahydrobiopterin deficiency in the pathogenesis of Fabry disease. *Human Molecular Genetics*, **2017**, 26, 1182-1192 | 5.6 | 8 |
| 163 | TUBB4A mutations result in specific neuronal and oligodendrocytic defects that closely match clinically distinct phenotypes. *Human Molecular Genetics*, **2017**, 26, 4506-4518 | 5.6 | 32 |
| 162 | Oculomotor and Vestibular Findings in Gaucher Disease Type 3 and Their Correlation with Neurological Findings. *Frontiers in Neurology*, **2017**, 8, 711 | 4.1 | 16 |
| 161 | Functionally pathogenic variants in vitro may not manifest a phenotype in vivo. *Neurology: Genetics*, **2017**, 3, e162 | 3.8 | 8 |
| 160 | Genotype-phenotype evaluation of MED13L defects in the light of a novel truncating and a recurrent missense mutation. *European Journal of Medical Genetics*, **2017**, 60, 451-464 | 2.6 | 26 |
| 159 | Gaucher disease: Progress and ongoing challenges. *Molecular Genetics and Metabolism*, **2017**, 120, 8-21 | 3.7 | 72 |
| 158 | The validation of pharmacogenetics for the identification of Fabry patients to be treated with migalastat. *Genetics in Medicine*, **2017**, 19, 430-438 | 8.1 | 110 |
| 157 | Mannose receptor-mediated delivery of moss-made α-galactosidase A efficiently corrects enzyme deficiency in Fabry mice. *Journal of Inherited Metabolic Disease*, **2016**, 39, 293-303 | 5.4 | 56 |
| 156 | Update on Leukodystrophies: A Historical Perspective and Adapted Definition. *Neuropediatrics*, **2016**, 47, 349-354 | 1.6 | 72 |
| 155 | Fabry Disease: A Disorder of Childhood Onset. *Pediatric Neurology*, **2016**, 64, 10-20 | 2.9 | 25 |
| 154 | Mutations in SNORD118 cause the cerebral microangiopathy leukoencephalopathy with calcifications and cysts. *Nature Genetics*, **2016**, 48, 1185-92 | 36.3 | 74 |
| 153 | Molecular basis for globotriaosylceramide regulation and enzyme uptake in immortalized aortic endothelial cells from Fabry mice. *Journal of Inherited Metabolic Disease*, **2016**, 39, 447-455 | 5.4 | 1 |

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| 152 | Triheptanoin dramatically reduces paroxysmal motor disorder in patients with GLUT1 deficiency. *Journal of Neurology, Neurosurgery and Psychiatry*, **2016**, 87, 550-3 | 5.5 | 60 |
| 151 | A Founder Mutation in VPS11 Causes an Autosomal Recessive Leukoencephalopathy Linked to Autophagic Defects. *PLoS Genetics*, **2016**, 12, e1005848 | 6 | 41 |
| 150 | An open-label clinical trial of agalsidase alfa enzyme replacement therapy in children with Fabry disease who are naïve to enzyme replacement therapy. *Drug Design, Development and Therapy*, **2016**, 10, 1771-81 | 4.4 | 12 |
| 149 | Whole exome sequencing in patients with white matter abnormalities. *Annals of Neurology*, **2016**, 79, 1031-1037 | 9.4 | 86 |
| 148 | Oxidative stress reflected by increased F-isoprostanes is associated with increasing urinary 11-dehydro thromboxane B levels in patients with coronary artery disease. *Thrombosis Research*, **2016**, 148, 85-88 | 8.2 | 8 |
| 147 | Is it Fabry disease?. *Genetics in Medicine*, **2016**, 18, 1181-1185 | 8.1 | 45 |
| 146 | Long-term follow-up and sudden unexpected death in Gaucher disease type 3 in Egypt. *Neurology: Genetics*, **2016**, 2, e55 | 3.8 | 13 |
| 145 | Treatment of Fabry® Disease with the Pharmacologic Chaperone Migalastat. *New England Journal of Medicine*, **2016**, 375, 545-55 | 59.2 | 254 |
| 144 | Deep intronic GBE1 mutation in manifesting heterozygous patients with adult polyglucosan body disease. *JAMA Neurology*, **2015**, 72, 441-5 | 17.2 | 25 |
| 143 | A prospective 10-year study of individualized, intensified enzyme replacement therapy in advanced Fabry disease. *Journal of Inherited Metabolic Disease*, **2015**, 38, 1129-36 | 5.4 | 29 |
| 142 | The consequences of genetic and pharmacologic reduction in sphingolipid synthesis. *Journal of Inherited Metabolic Disease*, **2015**, 38, 77-84 | 5.4 | 8 |
| 141 | Neurogenic bladder and neuroendocrine abnormalities in Pol III-related leukodystrophy. *BMC Neurology*, **2015**, 15, 22 | 3.1 | 3 |
| 140 | Blocking hyperactive androgen receptor signaling ameliorates cardiac and renal hypertrophy in Fabry mice. *Human Molecular Genetics*, **2015**, 24, 3181-91 | 5.6 | 17 |
| 139 | Gaucher Disease: Neuronopathic Forms **2015**, 301-311 | | |
| 138 | Fabry disease. *Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn*, **2015**, 132, 231-48 | 3 | 42 |
| 137 | Identification of a biomarker in cerebrospinal fluid for neuronopathic forms of Gaucher disease. *PLoS ONE*, **2015**, 10, e0120194 | 3.7 | 36 |
| 136 | A genetic form of achlorhydria and gastritis. *American Journal of Clinical Nutrition*, **2015**, 102, 1615 | 7 | |
| 135 | Disease specific therapies in leukodystrophies and leukoencephalopathies. *Molecular Genetics and Metabolism*, **2015**, 114, 527-36 | 3.7 | 35 |

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| 134 | Case definition and classification of leukodystrophies and leukoencephalopathies. *Molecular Genetics and Metabolism*, **2015**, 114, 494-500 | 3.7 | 137 |
| 133 | CSF and Blood Levels of GFAP in Alexander Disease. *ENeuro*, **2015**, 2, | 3.9 | 22 |
| 132 | Clinical spectrum of 4H leukodystrophy caused by POLR3A and POLR3B mutations. *Neurology*, **2014** , 83, 1898-905 | 6.5 | 118 |
| 131 | Hypomyelination with atrophy of the basal ganglia and cerebellum: further delineation of the phenotype and genotype-phenotype correlation. *Brain*, **2014**, 137, 1921-30 | 11.2 | 72 |
| 130 | Agalsidase alfa in pediatric patients with Fabry disease: a 6.5-year open-label follow-up study. *Orphanet Journal of Rare Diseases*, **2014**, 9, 169 | 4.2 | 27 |
| 129 | Risk of death in heart disease is associated with elevated urinary globotriaosylceramide. *Journal of the American Heart Association*, **2014**, 3, e000394 | 6 | 18 |
| 128 | Quantitative neuroimaging in mucolipidosis type IV. *Molecular Genetics and Metabolism*, **2014**, 111, 147-51 | 3.7 | 18 |
| 127 | Abnormal glycogen in astrocytes is sufficient to cause adult polyglucosan body disease. *Gene*, **2013**, 515, 376-9 | 3.8 | 17 |
| 126 | A de novo mutation in the β-tubulin gene TUBB4A results in the leukoencephalopathy hypomyelination with atrophy of the basal ganglia and cerebellum. *American Journal of Human Genetics*, **2013**, 92, 767-73 | 11 | 133 |
| 125 | Neuronopathic phenotypes of Gaucher disease **2013**, 12-24 | | 1 |
| 124 | Auditory analysis of xeroderma pigmentosum 1971-2012: hearing function, sun sensitivity and DNA repair predict neurological degeneration. *Brain*, **2013**, 136, 194-208 | 11.2 | 39 |
| 123 | Reply: To PMID 23034915. *Annals of Neurology*, **2013**, 73, 318 | 9.4 | |
| 122 | Changes in plasma and urine globotriaosylceramide levels do not predict Fabry disease progression over 1 year of agalsidase alfa. *Genetics in Medicine*, **2013**, 15, 983-9 | 8.1 | 15 |
| 121 | HIV Tat Domain Improves Cross-correction of Human Galactocerebrosidase in a Gene- and Flanking Sequence-dependent Manner. *Molecular Therapy - Nucleic Acids*, **2013**, 2, e130 | 10.7 | 7 |
| 120 | More than hypomyelination in Pol-III disorder. *Journal of Neuropathology and Experimental Neurology*, **2013**, 72, 67-75 | 3.1 | 18 |
| 119 | Characterization of Transferrin Glycopeptide Structures in Human Cerebrospinal Fluid. *International Journal of Mass Spectrometry*, **2012**, 312, 97-106 | 1.9 | 15 |
| 118 | 4H syndrome with late-onset growth hormone deficiency caused by POLR3A mutations. *Archives of Neurology*, **2012**, 69, 920-3 | | 37 |
| 117 | Adult polyglucosan body disease: Natural History and Key Magnetic Resonance Imaging Findings. *Annals of Neurology*, **2012**, 72, 433-41 | 9.4 | 88 |

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| 116 | Mutations in CTC1, encoding conserved telomere maintenance component 1, cause Coats plus. *Nature Genetics*, **2012**, 44, 338-42 | 36.3 | 186 |
| 115 | Developmental splicing deregulation in leukodystrophies related to EIF2B mutations. *PLoS ONE*, **2012**, 7, e38264 | 3.7 | 12 |
| 114 | Early alterations of brain cellular energy homeostasis in Huntington disease models. *Journal of Biological Chemistry*, **2012**, 287, 1361-70 | 5.4 | 92 |
| 113 | TACH leukodystrophy: locus refinement to chromosome 10q22.3-23.1. *Canadian Journal of Neurological Sciences*, **2012**, 39, 122-3 | 1 | 4 |
| 112 | Skin ultrastructural findings in type 2 Gaucher disease: diagnostic implications. *Molecular Genetics and Metabolism*, **2011**, 104, 631-6 | 3.7 | 25 |
| 111 | Altered dopamine and serotonin metabolism in motorically asymptomatic R6/2 mice. *PLoS ONE*, **2011**, 6, e18336 | 3.7 | 41 |
| 110 | Mutations of POLR3A encoding a catalytic subunit of RNA polymerase Pol III cause a recessive hypomyelinating leukodystrophy. *American Journal of Human Genetics*, **2011**, 89, 415-23 | 11 | 163 |
| 109 | Recessive mutations in POLR3B, encoding the second largest subunit of Pol III, cause a rare hypomyelinating leukodystrophy. *American Journal of Human Genetics*, **2011**, 89, 652-5 | 11 | 107 |
| 108 | A pharmacogenetic approach to identify mutant forms of Ɑ-galactosidase A that respond to a pharmacological chaperone for Fabry disease. *Human Mutation*, **2011**, 32, 965-77 | 4.7 | 69 |
| 107 | GnRH-deficient phenotypes in humans and mice with heterozygous variants in KISS1/Kiss1. *Journal of Clinical Endocrinology and Metabolism*, **2011**, 96, E1771-81 | 5.6 | 51 |
| 106 | Sex differences of urinary and kidney globotriaosylceramide and lyso-globotriaosylceramide in Fabry mice. *Journal of Lipid Research*, **2011**, 52, 1742-6 | 6.3 | 15 |
| 105 | The saccadic and neurological deficits in type 3 Gaucher disease. *PLoS ONE*, **2011**, 6, e22410 | 3.7 | 52 |
| 104 | The pharmacological chaperone isofagomine increases the activity of the Gaucher disease L444P mutant form of beta-glucosidase. *FEBS Journal*, **2010**, 277, 1618-38 | 5.7 | 117 |
| 103 | Use of lissamine rhodamine ceramide trihexoside as a functional assay for alpha-galactosidase A in intact cells. *Journal of Lipid Research*, **2010**, 51, 2808-17 | 6.3 | 5 |
| 102 | Biomarkers of Fabry disease nephropathy. *Clinical Journal of the American Society of Nephrology: CJASN*, **2010**, 5, 360-4 | 6.9 | 46 |
| 101 | Therapeutic approaches for neuronopathic lysosomal storage disorders. *Journal of Inherited Metabolic Disease*, **2010**, 33, 373-9 | 5.4 | 27 |
| 100 | Four-year prospective clinical trial of agalsidase alfa in children with Fabry disease. *Journal of Pediatrics*, **2010**, 156, 832-7, 837.e1 | 3.6 | 46 |
| 99 | Unexpected occurrence of xeroderma pigmentosum in an uncle and nephew. *Archives of Dermatology*, **2009**, 145, 1285-91 | | 9 |

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| 98 | Agalsidase alfa and kidney dysfunction in Fabry disease. *Journal of the American Society of Nephrology: JASN*, **2009**, 20, 1132-9 | 12.7 | 127 |
| 97 | Fabry disease. *Pharmacology & Therapeutics*, **2009**, 122, 65-77 | 13.9 | 169 |
| 96 | Effect of agalsidase alfa replacement therapy on Fabry disease-related hypertrophic cardiomyopathy: a 12- to 36-month, retrospective, blinded echocardiographic pooled analysis. *Clinical Therapeutics*, **2009**, 31, 1966-76 | 3.5 | 32 |
| 95 | XPC initiation codon mutation in xeroderma pigmentosum patients with and without neurological symptoms. *DNA Repair*, **2009**, 8, 114-25 | 4.3 | 29 |
| 94 | Fabry disease: progression of nephropathy, and prevalence of cardiac and cerebrovascular events before enzyme replacement therapy. *Nephrology Dialysis Transplantation*, **2009**, 24, 2102-11 | 4.3 | 252 |
| 93 | Falsely elevated urinary Gb3 (globotriaosylceramide, CTH, GL3). *Molecular Genetics and Metabolism*, **2009**, 97, 91 | 3.7 | 10 |
| 92 | Invited article: an MRI-based approach to the diagnosis of white matter disorders. *Neurology*, **2009**, 72, 750-9 | 6.5 | 395 |
| 91 | Eukaryotic initiation factor 2B (eIF2B) GEF activity as a diagnostic tool for EIF2B-related disorders. *PLoS ONE*, **2009**, 4, e8318 | 3.7 | 37 |
| 90 | Time series proteome profiling to study endoplasmic reticulum stress response. *Journal of Proteome Research*, **2008**, 7, 2435-44 | 5.6 | 34 |
| 89 | Globotriaosylceramide induces oxidative stress and up-regulates cell adhesion molecule expression in Fabry disease endothelial cells. *Molecular Genetics and Metabolism*, **2008**, 95, 163-8 | 3.7 | 155 |
| 88 | Genetic and clinical heterogeneity in eIF2B-related disorder. *Journal of Child Neurology*, **2008**, 23, 205-15 | 2.5 | 42 |
| 87 | The sub-cellular localization globotriaosylceramide in Fabry disease. *Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin*, **2008**, 452, 707-708 | 5.1 | 2 |
| 86 | Randomized, controlled trial of miglustat in Gaucher's disease type 3. *Annals of Neurology*, **2008**, 64, 514-22 | 9.4 | 177 |
| 85 | The cerebral vasculopathy of Fabry disease. *Journal of the Neurological Sciences*, **2007**, 257, 258-63 | 3.2 | 109 |
| 84 | Mitochondrial aspartyl-tRNA synthetase deficiency causes leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation. *Nature Genetics*, **2007**, 39, 534-9 | 36.3 | 363 |
| 83 | Cellular and tissue localization of globotriaosylceramide in Fabry disease. *Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin*, **2007**, 451, 823-34 | 5.1 | 81 |
| 82 | The pharmacology of multiple regimens of agalsidase alfa enzyme replacement therapy for Fabry disease. *Genetics in Medicine*, **2007**, 9, 504-9 | 8.1 | 32 |
| 81 | Weekly enzyme replacement therapy may slow decline of renal function in patients with Fabry disease who are on long-term biweekly dosing. *Journal of the American Society of Nephrology: JASN*, **2007**, 18, 1576-83 | 12.7 | 101 |

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| 80 | Enzyme replacement in Fabry disease: pharmacokinetics and pharmacodynamics of agalsidase alpha in children and adolescents. *Journal of Clinical Pharmacology*, **2007**, 47, 1222-30 | 2.9 | 32 |
| 79 | Proteomics of specific treatment-related alterations in Fabry disease: a strategy to identify biological abnormalities. *Proceedings of the National Academy of Sciences of the United States of America*, **2007**, 104, 2873-8 | 11.5 | 44 |
| 78 | Elevated endothelial microparticles in Fabry children decreased after enzyme replacement therapy. *Arteriosclerosis, Thrombosis, and Vascular Biology*, **2007**, 27, e138-9 | 9.4 | 20 |
| 77 | Genomic abnormalities of the murine model of Fabry disease after disease-related perturbation, a systems biology approach. *Proceedings of the National Academy of Sciences of the United States of America*, **2007**, 104, 8065-70 | 11.5 | 14 |
| 76 | Cellular and tissue distribution of intravenously administered agalsidase alfa. *Molecular Genetics and Metabolism*, **2007**, 90, 307-12 | 3.7 | 30 |
| 75 | Establishment and characterization of Fabry disease endothelial cells with an extended lifespan. *Molecular Genetics and Metabolism*, **2007**, 92, 137-44 | 3.7 | 21 |
| 74 | Screening for pharmacological chaperones in Fabry disease. *Biochemical and Biophysical Research Communications*, **2007**, 359, 168-73 | 3.4 | 42 |
| 73 | Clinical and molecular phenotype of Aicardi-Goutieres syndrome. *American Journal of Human Genetics*, **2007**, 81, 713-25 | 11 | 310 |
| 72 | Enzyme replacement therapy and intraepidermal innervation density in Fabry disease. *Muscle and Nerve*, **2006**, 34, 53-6 | 3.4 | 77 |
| 71 | Quantitative dysmorphology assessment in Fabry disease. *Genetics in Medicine*, **2006**, 8, 96-101 | 8.1 | 33 |
| 70 | Long-term therapy with agalsidase alfa for Fabry disease: safety and effects on renal function in a home infusion setting. *Nephrology Dialysis Transplantation*, **2006**, 21, 345-54 | 4.3 | 207 |
| 69 | Enzyme-replacement therapy with agalsidase alfa in children with Fabry disease. *Pediatrics*, **2006**, 118, 924-32 | 7.4 | 140 |
| 68 | Childhood ataxia with CNS hypomyelination/vanishing white matter disease--a common leukodystrophy caused by abnormal control of protein synthesis. *Molecular Genetics and Metabolism*, **2006**, 88, 7-15 | 3.7 | 57 |
| 67 | Lamin B1 duplications cause autosomal dominant leukodystrophy. *Nature Genetics*, **2006**, 38, 1114-23 | 36.3 | 312 |
| 66 | PLP1 and GPM6B intragenic copy number analysis by MAPH in 262 patients with hypomyelinating leukodystrophies: Identification of one partial triplication and two partial deletions of PLP1. *Neurogenetics*, **2006**, 7, 31-7 | 3 | 20 |
| 65 | Pathological findings in a patient with Fabry disease who died after 2.5 years of enzyme replacement. *Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin*, **2006**, 448, 337-43 | 5.1 | 67 |
| 64 | The significance of lysosomal inclusions in Fabry disease. *Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin*, **2006**, 449, 134 | 5.1 | 1 |
| 63 | Elevated Counts of Circulating Endothelial Microparticles in Pediatric Fabry Patients Decreased after Enzyme Replacement Therapy.. *Blood*, **2006**, 108, 1818-1818 | 2.2 | |

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| 62 | Neuropathy and Fabry disease: pathogenesis and enzyme replacement therapy. *Acta Neurologica Belgica*, **2006**, 106, 61-5 | 1.5 | 26 |
| 61 | Effect of genetic modifiers on cerebral lesions in Fabry disease. *Neurology*, **2005**, 64, 2148-50 | 6.5 | 79 |
| 60 | The relationship of vascular glycolipid storage to clinical manifestations of Fabry disease: a cross-sectional study of a large cohort of clinically affected heterozygous women. *Medicine (United States)*, **2005**, 84, 261-268 | 1.8 | 91 |
| 59 | Fabry disease--an important risk factor for stroke. *Lancet, The*, **2005**, 366, 1754-6 | 40 | 32 |
| 58 | Enhanced calcium release in the acute neuronopathic form of Gaucher disease. *Neurobiology of Disease*, **2005**, 18, 83-8 | 7.5 | 125 |
| 57 | Characterization of tiger-tail banding and hair shaft abnormalities in trichothiodystrophy. *Journal of the American Academy of Dermatology*, **2005**, 52, 224-32 | 4.5 | 59 |
| 56 | Improved intracellular delivery of glucocerebrosidase mediated by the HIV-1 TAT protein transduction domain. *Biochemical and Biophysical Research Communications*, **2005**, 337, 701-7 | 3.4 | 21 |
| 55 | Gaucher mutation N188S is associated with myoclonic epilepsy. *Human Mutation*, **2005**, 26, 271-3; author reply 274-5 | 4.7 | 17 |
| 54 | Heightened stress response in primary fibroblasts expressing mutant eIF2B genes from CACH/VWM leukodystrophy patients. *Human Genetics*, **2005**, 118, 99-106 | 6.3 | 69 |
| 53 | Decreased asialotransferrin in cerebrospinal fluid of patients with childhood-onset ataxia and central nervous system hypomyelination/vanishing white matter disease. *Clinical Chemistry*, **2005**, 51, 2031-42 | 5.5 | 40 |
| 52 | Pediatric Fabry disease. *Pediatrics*, **2005**, 115, e344-55 | 7.4 | 144 |
| 51 | Fabry disease: angiokeratoma, biomarker, and the effect of enzyme replacement therapy on kidney function. *Archives of Dermatology*, **2005**, 141, 904-5; author reply 905-6 | | 7 |
| 50 | Parapelvic kidney cysts: a distinguishing feature with high prevalence in Fabry disease. *Kidney International*, **2004**, 66, 978-82 | 9.9 | 28 |
| 49 | Decreased guanine nucleotide exchange factor activity in eIF2B-mutated patients. *European Journal of Human Genetics*, **2004**, 12, 561-6 | 5.3 | 75 |
| 48 | Enzyme-replacement therapy for metabolic storage disorders. *Lancet Neurology, The*, **2004**, 3, 752-6 | 24.1 | 75 |
| 47 | Transfer of a mitochondrial DNA fragment to MCOLN1 causes an inherited case of mucolipidosis IV. *Human Mutation*, **2004**, 24, 460-5 | 4.7 | 61 |
| 46 | Ascorbate decreases Fabry cerebral hyperperfusion suggesting a reactive oxygen species abnormality: an arterial spin tagging study. *Journal of Magnetic Resonance Imaging*, **2004**, 20, 674-83 | 5.6 | 61 |
| 45 | Neuropathology provides clues to the pathophysiology of Gaucher disease. *Molecular Genetics and Metabolism*, **2004**, 82, 192-207 | 3.7 | 360 |

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|---|---|---|---|
| 44 | The latest on leukodystrophies. *Current Opinion in Neurology*, **2004**, 17, 187-92 | 7.1 | 73 |
| 43 | Myoclonic epilepsy in Gaucher disease: genotype-phenotype insights from a rare patient subgroup. *Pediatric Research*, **2003**, 53, 387-95 | 3.2 | 85 |
| 42 | Images in clinical medicine. Fabry's disease. *New England Journal of Medicine*, **2003**, 349, e20 | 59.2 | |
| 41 | Enzyme replacement therapy improves peripheral nerve and sweat function in Fabry disease. *Muscle and Nerve*, **2003**, 28, 703-10 | 3.4 | 177 |
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