

# Robert Gruber

## List of Publications by Year in descending order

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Version: 2024-02-01

57  
papers

2,387  
citations

279798

23  
h-index

214800

47  
g-index

62  
all docs

62  
docs citations

62  
times ranked

3092  
citing authors

| #  | ARTICLE   | IF  | CITATIONS |
|----|---|-----|-----------|
| 1  | Coincidence of Bullous Pemphigoid and Pityriasis Rubra Pilaris. <i>Acta Dermato-Venereologica</i> , 2022, 102, adv00674.  | 1.3 | 2         |
| 2  | Mitochondrial Activity Is Upregulated in Nonlesional Atopic Dermatitis and Amenable to Therapeutic Intervention. <i>Journal of Investigative Dermatology</i> , 2022, 142, 2623-2634.e12.  | 0.7 | 11        |
| 3  | Revisiting the Roles of Filaggrin in Atopic Dermatitis. <i>International Journal of Molecular Sciences</i> , 2022, 23, 5318.  | 4.1 | 41        |
| 4  | Initial Evidence of Distinguishable Bacterial and Fungal Dysbiosis in the Skin of Patients with Atopic Dermatitis or Netherton Syndrome. <i>Journal of Investigative Dermatology</i> , 2021, 141, 114-123.                            | 0.7 | 23        |
| 5  | Meta-Analysis of Mutations in ALOX12B or ALOXE3 Identified in a Large Cohort of 224 Patients. <i>Genes</i> , 2021, 12, 80.  | 2.4 | 20        |
| 6  | Two patients with Papillon-Lefèvre syndrome without periodontal involvement of the permanent dentition. <i>Journal of Dermatology</i> , 2021, 48, 537-541.  | 1.2 | 3         |
| 7  | hiPSC-Derived Epidermal Keratinocytes from Ichthyosis Patients Show Altered Expression of Cornification Markers. <i>International Journal of Molecular Sciences</i> , 2021, 22, 1785.   | 4.1 | 4         |
| 8  | Keratinocyte-derived IL-1 $\beta$ induces PPAR $\gamma$ downregulation and PPAR $\delta$ upregulation in human reconstructed epidermis following barrier impairment. <i>Experimental Dermatology</i> , 2021, 30, 1298-1308.           | 2.9 | 12        |
| 9  | Successful treatment of trichothiodystrophy with dupilumab. <i>Clinical and Experimental Dermatology</i> , 2021, 46, 1381-1383.   | 1.3 | 9         |
| 10 | Unbound Corneocyte Lipid Envelopes in 12R-Lipoxygenase Deficiency Support a Specific Role in Lipid-Protein Cross-Linking. <i>American Journal of Pathology</i> , 2021, 191, 921-929.  | 3.8 | 6         |
| 11 | Cystatin M/E Variant Causes Autosomal Dominant Keratosis Follicularis Spinulosa Decalvans by Dysregulating Cathepsins L and V. <i>Frontiers in Genetics</i> , 2021, 12, 689940.   | 2.3 | 5         |
| 12 | Peroxisomal Fatty Acid Oxidation and Glycolysis Are Triggered in Mouse Models of Lesional Atopic Dermatitis. <i>JID Innovations</i> , 2021, 1, 100033.  | 2.4 | 16        |
| 13 | Advancing novel therapies for ichthyoses. <i>British Journal of Dermatology</i> , 2021, 184, 998-999.   | 1.5 | 3         |
| 14 | Spectrum of ichthyoses in an Austrian ichthyosis cohort from 2004 to 2017. <i>JDDG - Journal of the German Society of Dermatology</i> , 2020, 18, 17-25.  | 0.8 | 11        |
| 15 | Management of congenital ichthyoses: European guidelines of care, part two. <i>British Journal of Dermatology</i> , 2019, 180, 484-495.   | 1.5 | 68        |
| 16 | Aggregated neutrophil extracellular traps resolve inflammation by proteolysis of cytokines and chemokines and protection from antiproteases. <i>FASEB Journal</i> , 2019, 33, 1401-1414.  | 0.5 | 90        |
| 17 | Induced pluripotent stem cell line heterozygous for p.R501X mutation in filaggrin: KCLi003-A. <i>Stem Cell Research</i> , 2019, 39, 101527.   | 0.7 | 5         |
| 18 | Transglutaminase 1 Replacement Therapy Successfully Mitigates the Autosomal Recessive Congenital Ichthyosis Phenotype in Full-Thickness Skin Disease Equivalents. <i>Journal of Investigative Dermatology</i> , 2019, 139, 1191-1195. | 0.7 | 24        |

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|----|---|-----|-----------|
| 19 | Impaired epidermal barrier in autosomal recessive congenital ichthyosis (ARCI) caused by missense mutations in <i>SDR9C7</i> in two Austrian sisters. <i>JDDG - Journal of the German Society of Dermatology</i> , 2019, 17, 742-745. | 0.8 | 6         |
| 20 | Induced pluripotent stem cell line heterozygous for p.R2447X mutation in filaggrin: KCLi002-A. <i>Stem Cell Research</i> , 2019, 38, 101462.  | 0.7 | 3         |
| 21 | Mutations in Recessive Congenital Ichthyoses Illuminate the Origin and Functions of the Corneocyte Lipid Envelope. <i>Journal of Investigative Dermatology</i> , 2019, 139, 760-768.  | 0.7 | 41        |
| 22 | Unknown mutations and genotype/phenotype correlations of autosomal recessive congenital ichthyosis in patients from Saudi Arabia and Pakistan. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2019, 7, e539.                      | 1.2 | 9         |
| 23 | Management of congenital ichthyoses: European guidelines of care, part one. <i>British Journal of Dermatology</i> , 2019, 180, 272-281.   | 1.5 | 70        |
| 24 | <i>Filaggrin</i> null mutations are associated with altered circulating Tregs in atopic dermatitis. <i>Journal of Cellular and Molecular Medicine</i> , 2019, 23, 1288-1299.  | 3.6 | 12        |
| 25 | A novel homozygous mutation in <i>PVRL4</i> causes ectodermal dysplasia-ectrodactyly syndrome 1. <i>International Journal of Dermatology</i> , 2018, 57, 223-226.   | 1.0 | 5         |
| 26 | Epidermal Overexpression of Xenobiotic Receptor PXR Impairs the Epidermal Barrier and Triggers Th2 Immune Response. <i>Journal of Investigative Dermatology</i> , 2018, 138, 109-120.   | 0.7 | 21        |
| 27 | Increased Prevalence of Filaggrin Deficiency in 51 Patients with Recessive X-Linked Ichthyosis Presenting for Dermatological Examination. <i>Journal of Investigative Dermatology</i> , 2018, 138, 709-711.                           | 0.7 | 18        |
| 28 | Enhanced Expression of Genes Related to Xenobiotic Metabolism in the Skin of Patients with Atopic Dermatitis but Not with Ichthyosis Vulgaris. <i>Journal of Investigative Dermatology</i> , 2018, 138, 98-108.                       | 0.7 | 28        |
| 29 | Induced pluripotent stem cell line from an atopic dermatitis patient heterozygous for c.2282del4 mutation in filaggrin: KCLi001-A. <i>Stem Cell Research</i> , 2018, 31, 122-126.   | 0.7 | 5         |
| 30 | Tight Junction barriers in human hair follicles – role of claudin-1. <i>Scientific Reports</i> , 2018, 8, 12800.  | 3.3 | 38        |
| 31 | Skin Barrier Development Depends on CGI-58 Protein Expression during Late-Stage Keratinocyte Differentiation. <i>Journal of Investigative Dermatology</i> , 2017, 137, 403-413.   | 0.7 | 33        |
| 32 | Autosomal Recessive Keratoderma-Ichthyosis-Deafness (ARKID) Syndrome Is Caused by VPS33B Mutations Affecting Rab Protein Interaction and Collagen Modification. <i>Journal of Investigative Dermatology</i> , 2017, 137, 845-854.     | 0.7 | 37        |
| 33 | Ustekinumab treatment in severe atopic dermatitis: Down-regulation of T-helper 2/22 expression. <i>Journal of the American Academy of Dermatology</i> , 2017, 76, 91-97.e3.   | 1.2 | 32        |
| 34 | Alterations in Epidermal Eicosanoid Metabolism Contribute to Inflammation and Impaired Late Differentiation in FLG-Mutated Atopic Dermatitis. <i>Journal of Investigative Dermatology</i> , 2017, 137, 706-715.                       | 0.7 | 43        |
| 35 | Morphological alterations in two siblings with autosomal recessive congenital ichthyosis associated with <i>CYP4F22</i> mutations. <i>British Journal of Dermatology</i> , 2017, 176, 1068-1073.                                      | 1.5 | 10        |
| 36 | Periodontal Ehlers-Danlos Syndrome Is Caused by Mutations in C1R and C1S, which Encode Subcomponents C1r and C1s of Complement. <i>American Journal of Human Genetics</i> , 2016, 99, 1005-1014.                                      | 6.2 | 100       |

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|----|---|-----|-----------|
| 37 | Epidermal barrier in hereditary ichthyoses, atopic dermatitis, and psoriasis. <i>JDDG - Journal of the German Society of Dermatology</i> , 2015, 13, 1119-1123.   | 0.8 | 19        |
| 38 | Epidermale Barriere bei hereditären Ichthyosen, atopischer Dermatitis und Psoriasis. <i>JDDG - Journal of the German Society of Dermatology</i> , 2015, 13, 1119-1124.  | 0.8 | 9         |
| 39 | Epidermal barrier abnormalities in exfoliative ichthyosis with a novel homozygous loss-of-function mutation in <i>CSTA</i> . <i>British Journal of Dermatology</i> , 2015, 172, 1628-1632.                                  | 1.5 | 15        |
| 40 | Increased risk of psoriasis in individuals with childhood asthma: therapeutic implications?. <i>British Journal of Dermatology</i> , 2015, 173, 14-14.  | 1.5 | 1         |
| 41 | Cellular Basis of Secondary Infections and Impaired Desquamation in Certain Inherited Ichthyoses. <i>JAMA Dermatology</i> , 2015, 151, 285.   | 4.1 | 24        |
| 42 | Severe dermatitis, multiple allergies, and metabolic wasting syndrome caused by a novel mutation in the N-terminal plaklin domain of desmoplakin. <i>Journal of Allergy and Clinical Immunology</i> , 2015, 136, 1268-1276. | 2.9 | 103       |
| 43 | Sebaceous Gland, Hair Shaft, and Epidermal Barrier Abnormalities in Keratosis Pilaris with and without Filaggrin Deficiency. <i>American Journal of Pathology</i> , 2015, 185, 1012-1021.                                   | 3.8 | 23        |
| 44 | Diverse Regulation of Claudin-1 and Claudin-4 in Atopic Dermatitis. <i>American Journal of Pathology</i> , 2015, 185, 2777-2789.  | 3.8 | 105       |
| 45 | Formation and functions of the corneocyte lipid envelope (CLE). <i>Biochimica Et Biophysica Acta - Molecular and Cell Biology of Lipids</i> , 2014, 1841, 314-318.  | 2.4 | 121       |
| 46 | Inherited ichthyoses/generalized Mendelian disorders of cornification. <i>European Journal of Human Genetics</i> , 2013, 21, 123-133.   | 2.8 | 86        |
| 47 | Slow growth of hair and nails, craniofacial abnormalities and brachyphalangy. <i>JDDG - Journal of the German Society of Dermatology</i> , 2013, 11, 1023-1025.   | 0.8 | 0         |
| 48 | Distinguishing Ichthyoses by Protein Profiling. <i>PLoS ONE</i> , 2013, 8, e75355.  | 2.5 | 30        |
| 49 | An appraisal of oral retinoids in the treatment of pachyonychia congenita. <i>Journal of the American Academy of Dermatology</i> , 2012, 66, e193-e199.   | 1.2 | 35        |
| 50 | Filaggrin Genotype in Ichthyosis Vulgaris Predicts Abnormalities in Epidermal Structure and Function. <i>American Journal of Pathology</i> , 2011, 178, 2252-2263.  | 3.8 | 213       |
| 51 | Concomitant juvenile xanthogranuloma and cutaneous mastocytosis in a 3-year-old Swedish girl: case report and review of the literature. <i>International Journal of Dermatology</i> , 2011, 50, 611-614.                    | 1.0 | 5         |
| 52 | Lower prevalence of common filaggrin mutations in a community sample of atopic eczema: is disease severity important?. <i>Wiener Klinische Wochenschrift</i> , 2010, 122, 551-557.  | 1.9 | 10        |
| 53 | Ichthyosis vulgaris: novel <i>FLG</i> mutations in the German population and high presence of CD1a+ cells in the epidermis of the atopic subgroup. <i>British Journal of Dermatology</i> , 2009, 160, 771-781.              | 1.5 | 40        |
| 54 | Increased pachyonychia congenita severity in patients with concurrent keratin and filaggrin mutations. <i>British Journal of Dermatology</i> , 2009, 161, 1391-1395.  | 1.5 | 21        |

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|----|---|------|-----------|
| 55 | Refractory Takayasu arteritis in a 65-year-old Caucasian woman. <i>Journal of the European Academy of Dermatology and Venereology</i> , 2009, 23, 191-193.                        | 2.4  | 0         |
| 56 | Comprehensive analysis of the gene encoding filaggrin uncovers prevalent and rare mutations in ichthyosis vulgaris and atopic eczema. <i>Nature Genetics</i> , 2007, 39, 650-654. | 21.4 | 598       |
| 57 | Filaggrin mutations p.R501X and c.2282del4 in ichthyosis vulgaris. <i>European Journal of Human Genetics</i> , 2007, 15, 179-184.   | 2.8  | 55        |