## **Robert Gruber**

List of Publications by Year in descending order

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POREDT COURED

#	Article	IF	CITATIONS
1	Comprehensive analysis of the gene encoding filaggrin uncovers prevalent and rare mutations in ichthyosis vulgaris and atopic eczema. Nature Genetics, 2007, 39, 650-654.	21.4	598
2	Filaggrin Genotype in Ichthyosis Vulgaris Predicts Abnormalities in Epidermal Structure and Function. American Journal of Pathology, 2011, 178, 2252-2263.	3.8	213
3	Formation and functions of the corneocyte lipid envelope (CLE). Biochimica Et Biophysica Acta - Molecular and Cell Biology of Lipids, 2014, 1841, 314-318.	2.4	121
4	Diverse Regulation of Claudin-1 and Claudin-4 in Atopic Dermatitis. American Journal of Pathology, 2015, 185, 2777-2789.	3.8	105
5	Severe dermatitis, multiple allergies, and metabolic wasting syndrome caused by a novel mutation in the N-terminal plakin domain of desmoplakin. Journal of Allergy and Clinical Immunology, 2015, 136, 1268-1276.	2.9	103
6	Periodontal Ehlers-Danlos Syndrome Is Caused by Mutations in C1R and C1S , which Encode Subcomponents C1r and C1s of Complement. American Journal of Human Genetics, 2016, 99, 1005-1014.	6.2	100
7	Aggregated neutrophil extracellular traps resolve inflammation by proteolysis of cytokines and chemokines and protection from antiproteases. FASEB Journal, 2019, 33, 1401-1414.	0.5	90
8	Inherited ichthyoses/generalized Mendelian disorders of cornification. European Journal of Human Genetics, 2013, 21, 123-133.	2.8	86
9	Management of congenital ichthyoses: European guidelines of care, part one. British Journal of Dermatology, 2019, 180, 272-281.	1.5	70
10	Management of congenital ichthyoses: European guidelines of care, part two. British Journal of Dermatology, 2019, 180, 484-495.	1.5	68
11	Filaggrin mutations p.R501X and c.2282del4 in ichthyosis vulgaris. European Journal of Human Genetics, 2007, 15, 179-184.	2.8	55
12	Alterations in Epidermal Eicosanoid Metabolism Contribute to Inflammation and Impaired Late Differentiation in FLG-Mutated Atopic Dermatitis. Journal of Investigative Dermatology, 2017, 137, 706-715.	0.7	43
13	Mutations in Recessive Congenital Ichthyoses Illuminate the Origin and Functions of the CorneocyteÂLipid Envelope. Journal of Investigative Dermatology, 2019, 139, 760-768.	0.7	41
14	Revisiting the Roles of Filaggrin in Atopic Dermatitis. International Journal of Molecular Sciences, 2022, 23, 5318.	4.1	41
15	Ichthyosis vulgaris: novel <i>FLG</i> mutations in the German population and high presence of CD1a+ cells in the epidermis of the atopic subgroup. British Journal of Dermatology, 2009, 160, 771-781.	1.5	40
16	Tight Junction barriers in human hair follicles – role of claudin-1. Scientific Reports, 2018, 8, 12800.	3.3	38
17	Autosomal Recessive Keratoderma-Ichthyosis-Deafness (ARKID) Syndrome IsÂCaused by VPS33B Mutations AffectingÂRab Protein Interaction andÂCollagen Modification. Journal of Investigative Dermatology, 2017, 137, 845-854.	0.7	37
18	An appraisal of oral retinoids in the treatment of pachyonychia congenita. Journal of the American Academy of Dermatology, 2012, 66, e193-e199.	1.2	35

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19	Skin Barrier Development Depends on CGI-58 Protein Expression during Late-Stage Keratinocyte Differentiation. Journal of Investigative Dermatology, 2017, 137, 403-413.	0.7	33
20	Ustekinumab treatment in severe atopic dermatitis: Down-regulation of T-helper 2/22 expression. Journal of the American Academy of Dermatology, 2017, 76, 91-97.e3.	1.2	32
21	Distinguishing Ichthyoses by Protein Profiling. PLoS ONE, 2013, 8, e75355.	2.5	30
22	Enhanced Expression of Genes Related to Xenobiotic Metabolism in the Skin of Patients with Atopic Dermatitis but Not with Ichthyosis Vulgaris. Journal of Investigative Dermatology, 2018, 138, 98-108.	0.7	28
23	Cellular Basis of Secondary Infections and Impaired Desquamation in Certain Inherited Ichthyoses. JAMA Dermatology, 2015, 151, 285.	4.1	24
24	Transglutaminase 1 Replacement Therapy Successfully Mitigates the Autosomal Recessive Congenital Ichthyosis Phenotype in Full-Thickness Skin Disease Equivalents. Journal of Investigative Dermatology, 2019, 139, 1191-1195.	0.7	24
25	Sebaceous Gland, Hair Shaft, and Epidermal Barrier Abnormalities in Keratosis Pilaris with and without Filaggrin Deficiency. American Journal of Pathology, 2015, 185, 1012-1021.	3.8	23
26	Initial Evidence of Distinguishable Bacterial and Fungal Dysbiosis in the Skin of Patients with Atopic Dermatitis or Netherton Syndrome. Journal of Investigative Dermatology, 2021, 141, 114-123.	0.7	23
27	Increased pachyonychia congenita severity in patients with concurrent keratin and filaggrin mutations. British Journal of Dermatology, 2009, 161, 1391-1395.	1.5	21
28	Epidermal Overexpression of Xenobiotic Receptor PXR Impairs the Epidermal Barrier and Triggers Th2 Immune Response. Journal of Investigative Dermatology, 2018, 138, 109-120.	0.7	21
29	Meta-Analysis of Mutations in ALOX12B or ALOXE3 Identified in a Large Cohort of 224 Patients. Genes, 2021, 12, 80.	2.4	20
30	Epidermal barrier in hereditary ichthyoses, atopic dermatitis, and psoriasis. JDDG - Journal of the German Society of Dermatology, 2015, 13, 1119-1123.	0.8	19
31	Increased Prevalence of Filaggrin Deficiency in 51 Patients with Recessive X-Linked Ichthyosis Presenting for Dermatological Examination. Journal of Investigative Dermatology, 2018, 138, 709-711.	0.7	18
32	Peroxisomal Fatty Acid Oxidation and Glycolysis Are Triggered in Mouse Models of Lesional Atopic Dermatitis. JID Innovations, 2021, 1, 100033.	2.4	16
33	Epidermal barrier abnormalities in exfoliative ichthyosis with a novel homozygous loss-of-function mutation in <i>CSTA</i> . British Journal of Dermatology, 2015, 172, 1628-1632.	1.5	15
34	<i>Filaggrin</i> null mutations are associated with altered circulating Tregs in atopic dermatitis. Journal of Cellular and Molecular Medicine, 2019, 23, 1288-1299.	3.6	12
35	Keratinocyteâ $\in$ derived ILâ $\in$ l $\hat{1}^2$ induces PPARG downregulation and PPARD upregulation in human reconstructed epidermis following barrier impairment. Experimental Dermatology, 2021, 30, 1298-1308.	2.9	12
36	Spectrum of ichthyoses in an Austrian ichthyosis cohort from 2004 to 2017. JDDG - Journal of the German Society of Dermatology, 2020, 18, 17-25.	0.8	11

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37	Mitochondrial Activity Is Upregulated in Nonlesional Atopic Dermatitis and Amenable to Therapeutic Intervention. Journal of Investigative Dermatology, 2022, 142, 2623-2634.e12.	0.7	11
38	Lower prevalence of common filaggrin mutations in a community sample of atopic eczema: is disease severity important?. Wiener Klinische Wochenschrift, 2010, 122, 551-557.	1.9	10
39	Morphological alterations in two siblings with autosomal recessive congenital ichthyosis associated with <i>CYP4F22</i> mutations. British Journal of Dermatology, 2017, 176, 1068-1073.	1.5	10
40	Epidermale Barriere bei hereditÃ <b>¤</b> en Ichthyosen, atopischer Dermatitis und Psoriasis. JDDG - Journal of the German Society of Dermatology, 2015, 13, 1119-1124.	0.8	9
41	Unknown mutations and genotype/phenotype correlations of autosomal recessive congenital ichthyosis in patients from Saudi Arabia and Pakistan. Molecular Genetics & Genomic Medicine, 2019, 7, e539.	1.2	9
42	Successful treatment of trichothiodystrophy with dupilumab. Clinical and Experimental Dermatology, 2021, 46, 1381-1383.	1.3	9
43	Impaired epidermal barrier in autosomal recessive congenital ichthyosis (ARCI) caused by missense mutations in <i>SDR9C7</i> in two Austrian sisters. JDDG - Journal of the German Society of Dermatology, 2019, 17, 742-745.	0.8	6
44	Unbound Corneocyte Lipid Envelopes in 12R-Lipoxygenase Deficiency Support a Specific Role in Lipid-Protein Cross-Linking. American Journal of Pathology, 2021, 191, 921-929.	3.8	6
45	Concomitant juvenile xanthogranuloma and cutaneous mastocytosis in a 3â€yearâ€old Swedish girl: case report and review of the literature. International Journal of Dermatology, 2011, 50, 611-614.	1.0	5
46	A novel homozygous mutation in <i><scp>PVRL</scp>4</i> causes ectodermal dysplasiaâ€syndactyly syndrome 1. International Journal of Dermatology, 2018, 57, 223-226.	1.0	5
47	Induced pluripotent stem cell line from an atopic dermatitis patient heterozygous for c.2282del4 mutation in filaggrin: KCLi001-A. Stem Cell Research, 2018, 31, 122-126.	0.7	5
48	Induced pluripotent stem cell line heterozygous for p.R501X mutation in filaggrin: KCLi003-A. Stem Cell Research, 2019, 39, 101527.	0.7	5
49	Cystatin M/E Variant Causes Autosomal Dominant Keratosis Follicularis Spinulosa Decalvans by Dysregulating Cathepsins L and V. Frontiers in Genetics, 2021, 12, 689940.	2.3	5
50	hiPSC-Derived Epidermal Keratinocytes from Ichthyosis Patients Show Altered Expression of Cornification Markers. International Journal of Molecular Sciences, 2021, 22, 1785.	4.1	4
51	Induced pluripotent stem cell line heterozygous for p.R2447X mutation in filaggrin: KCLi002-A. Stem Cell Research, 2019, 38, 101462.	0.7	3
52	Two patients with Papillon–LefÔvre syndrome without periodontal involvement of the permanent dentition. Journal of Dermatology, 2021, 48, 537-541.	1.2	3
53	Advancing novel therapies for ichthyoses. British Journal of Dermatology, 2021, 184, 998-999.	1.5	3
54	Coincidence of Bullous Pemphigoid and Pityriasis Rubra Pilaris. Acta Dermato-Venereologica, 2022, 102, adv00674.	1.3	2

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55	Increased risk of psoriasis in individuals with childhood asthma: therapeutic implications?. British Journal of Dermatology, 2015, 173, 14-14.	1.5	1
56	Refractory Takayasu arteritis in a 65â€yearâ€old Caucasian woman. Journal of the European Academy of Dermatology and Venereology, 2009, 23, 191-193.	2.4	0
57	Slow growth of hair and nails, craniofacial abnormalities and brachyphalangy. JDDG - Journal of the German Society of Dermatology, 2013, 11, 1023-1025.	0.8	0