Ofer Sarig

List of Publications by Year in descending order

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75 papers	1,965 citations	257357 24 h-index	276775 41 g-index
76 all docs	76 docs citations	76 times ranked	2984 citing authors

#	Article	IF	Citations
1	Desmoglein 1 deficiency results in severe dermatitis, multiple allergies and metabolic wasting. Nature Genetics, 2013, 45, 1244-1248.	9.4	289
2	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.	1.5	103
3	Variant PADI3 in Central Centrifugal Cicatricial Alopecia. New England Journal of Medicine, 2019, 380, 833-841.	13.9	102
4	Stabilizing mutations of KLHL24 ubiquitin ligase cause loss of keratin 14 and human skin fragility. Nature Genetics, 2016, 48, 1508-1516.	9.4	101
5	Population-Specific Association between a Polymorphic Variant in ST18, Encoding a Pro-Apoptotic Molecule, and Pemphigus Vulgaris. Journal of Investigative Dermatology, 2012, 132, 1798-1805.	0.3	98
6	CEDNIK syndrome results from loss-of-function mutations in SNAP29. British Journal of Dermatology, 2011, 164, no-no.	1.4	69
7	A Mutation in LIPN, Encoding Epidermal Lipase N, Causes a Late-Onset Form of Autosomal-Recessive Congenital Ichthyosis. American Journal of Human Genetics, 2011, 88, 482-487.	2.6	62
8	The Genetics of Pemphigus Vulgaris. Frontiers in Medicine, 2018, 5, 226.	1.2	60
9	Loss-of-function mutations in caspase recruitment domain-containing protein 14 (CARD14) are associated with a severe variant of atopic dermatitis. Journal of Allergy and Clinical Immunology, 2019, 143, 173-181.e10.	1.5	60
10	Short Stature, Onychodysplasia, Facial Dysmorphism, and Hypotrichosis Syndrome Is Caused by a POC1A Mutation. American Journal of Human Genetics, 2012, 91, 337-342.	2.6	59
11	Autosomal dominant inheritance of central centrifugalÂcicatricial alopecia in black South Africans. Journal of the American Academy of Dermatology, 2014, 70, 679-682.e1.	0.6	54
12	Pyoderma gangrenosum, acne and ulcerative colitis in a patient with a novel mutation in thePSTPIP1gene. Clinical and Experimental Dermatology, 2015, 40, 367-372.	0.6	53
13	Identification of a Functional Risk Variant for Pemphigus Vulgaris in the ST18 Gene. PLoS Genetics, 2016, 12, e1006008.	1.5	53
14	A phenotype combining hidradenitis suppurativa with Dowling-Degos disease caused by a founder mutation in PSENEN. British Journal of Dermatology, 2018, 178, 502-508.	1.4	48
15	Olmsted Syndrome Caused by a Homozygous Recessive Mutation in TRPV3. Journal of Investigative Dermatology, 2014, 134, 1752-1754.	0.3	44
16	Treatment of epidermolysis bullosa pruriginosaâ€associated pruritus with dupilumab. British Journal of Dermatology, 2020, 182, 1495-1497.	1.4	41
17	Infantile mitochondrial hepatopathy is a cardinal feature of MEGDEL syndrome (3â€Methylglutaconic) Tj ETQq1 1 0.784314 rgBT /Ov mutations in SERAC1. American Journal of Medical Genetics, Part A, 2013, 161, 2204-2215.	0.7	39
18	Filaggrin 2 Deficiency Results in Abnormal Cell-Cell Adhesion in the Cornified Cell Layers and Causes Peeling Skin Syndrome Type A. Journal of Investigative Dermatology, 2018, 138, 1736-1743.	0.3	37

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19	Non-syndromic autosomal recessive congenital ichthyosis in the Israeli population. Clinical and Experimental Dermatology, 2013, 38, 911-916.	0.6	34
20	Segmental basal cell naevus syndrome caused by an activating mutation in smoothened. British Journal of Dermatology, 2016, 175, 178-181.	1.4	33
21	Mutations in TSPEAR, Encoding a Regulator of Notch Signaling, Affect Tooth and Hair Follicle Morphogenesis. PLoS Genetics, 2016, 12, e1006369.	1.5	32
22	Establishment of Two Mouse Models for CEDNIK Syndrome Reveals the Pivotal Role of SNAP29 in Epidermal Differentiation. Journal of Investigative Dermatology, 2016, 136, 672-679.	0.3	31
23	Loss-of-Function Mutations in SERPINB8 Linked to Exfoliative Ichthyosis with Impaired Mechanical Stability of Intercellular Adhesions. American Journal of Human Genetics, 2016, 99, 430-436.	2.6	27
24	Analysis of CARD14 Polymorphisms in Pityriasis Rubra Pilaris: Activation of NF-Î¹B. Journal of Investigative Dermatology, 2015, 135, 1905-1908.	0.3	24
25	SAM syndrome is characterized by extensive phenotypic heterogeneity. Experimental Dermatology, 2018, 27, 787-790.	1.4	22
26	ST18 Enhances PV-IgG-Induced Loss of Keratinocyte Cohesion in Parallel to Increased ERK Activation. Frontiers in Immunology, 2019, 10, 770.	2.2	20
27	Calpain 12 Function Revealed through the Study of an Atypical Case of Autosomal Recessive Congenital Ichthyosis. Journal of Investigative Dermatology, 2017, 137, 385-393.	0.3	19
28	Treatment of hereditary hypotrichosis simplex of the scalp with topical gentamicin. British Journal of Dermatology, 2020, 183, 114-120.	1.4	19
29	<scp>RBM</scp>28, a protein deficient in <scp>ANE</scp> syndrome, regulates hair follicle growth via miRâ€203 and p63. Experimental Dermatology, 2015, 24, 618-622.	1.4	17
30	<scp>SVEP</scp>1 plays a crucial role in epidermal differentiation. Experimental Dermatology, 2017, 26, 423-430.	1.4	17
31	The Role of Desmoglein 1 in Gap Junction Turnover Revealed through the Study of SAMÂSyndrome. Journal of Investigative Dermatology, 2020, 140, 556-567.e9.	0.3	17
32	The Molecular Revolution in Cutaneous Biology: EraÂof Next-Generation Sequencing. Journal of Investigative Dermatology, 2017, 137, e79-e82.	0.3	16
33	IGFBP7 as a Potential Therapeutic Target in Psoriasis. Journal of Investigative Dermatology, 2011, 131, 1767-1770.	0.3	14
34	Loss-of-Function Variants in SERPINA12 Underlie Autosomal Recessive Palmoplantar Keratoderma. Journal of Investigative Dermatology, 2020, 140, 2178-2187.	0.3	14
35	Molecular Analysis of a Series of Israeli Families with ComÃ¨l-Netherton Syndrome. Dermatology, 2014, 228, 183-188.	0.9	12
36	Coagulation Factor XIII-A Subunit Missense Mutation in the Pathobiology of Autosomal Dominant Multiple Dermatofibromas. Journal of Investigative Dermatology, 2020, 140, 624-635.e7.	0.3	12

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37	Loss-of-function variants in C3ORF52 result in localized autosomal recessive hypotrichosis. Genetics in Medicine, 2020, 22, 1227-1234.	1.1	12
38	Papillonâ€“LefÃ¨vre syndrome: report of six patients and identification of a novel mutation. International Journal of Dermatology, 2016, 55, 898-902.	0.5	11
39	Identification of a recurrent mutation in ATP2C1 demonstrates that papular acantholytic dyskeratosis and Hailey-Hailey disease are allelic disorders. British Journal of Dermatology, 2018, 179, 1001-1002.	1.4	11
40	Semidominant Inheritance in Epidermolytic Ichthyosis. Journal of Investigative Dermatology, 2013, 133, 2626-2628.	0.3	10
41	Recessive epidermolytic ichthyosis results from loss of keratin 10 expression, regardless of the mutation location. Clinical and Experimental Dermatology, 2018, 43, 187-190.	0.6	10
42	Molecular epidemiology of nonâ€syndromic autosomal recessive congenital ichthyosis in a Middleâ€Eastern population. Experimental Dermatology, 2021, 30, 1290-1297.	1.4	10
43	Evidence for cutaneous dysbiosis in dystrophic epidermolysis bullosa. Clinical and Experimental Dermatology, 2021, 46, 1223-1229.	0.6	10
44	Extensive lentigo simplex, linear epidermolytic naevus and epidermolytic naevus comedonicus caused by a somatic mutation inKRT10. British Journal of Dermatology, 2015, 173, 293-296.	1.4	9
45	Epidermolysis bullosa simplex due to biâ€allelic DST mutations: Case series and review of the literature. Pediatric Dermatology, 2021, 38, 436-441.	0.5	9
46	Nonâ€keratinocyte <scp>SNAP</scp>29 influences epidermal differentiation and hair follicle formation in mice. Experimental Dermatology, 2016, 25, 647-649.	1.4	8
47	Striate palmoplantar keratoderma resulting from a missense mutation in DSG1. British Journal of Dermatology, 2018, 179, 755-757.	1.4	8
48	PLACK syndrome shows remarkable phenotypic homogeneity. Clinical and Experimental Dermatology, 2019, 44, 580-583.	0.6	8
49	A novel homozygous deletion inEXPH5causes a skin fragility phenotype. Clinical and Experimental Dermatology, 2016, 41, 915-918.	0.6	7
50	ST18 affects cellâ€“cell adhesion in pemphigus vulgaris in a tumour necrosis factorâ€Î±â€dependent fashion*. British Journal of Dermatology, 2021, 184, 1153-1160.	1.4	7
51	Vorinostat, a histone deacetylase inhibitor, as a potential novel treatment for psoriasis. Experimental Dermatology, 2022, 31, 567-576.	1.4	7
52	Coexistence of pachyonychia congenita and hidradenitis suppurativa: more than a coincidence. British Journal of Dermatology, 2022, 187, 392-400.	1.4	7
53	ARCI7 Revisited and Repositioned. Journal of Investigative Dermatology, 2017, 137, 970-972.	0.3	6
54	Immuneâ€regulatory genes as possible modifiers of familial pityriasis rubra pilaris â€“ lessons from a family with <scp>PRP</scp> and psoriasis. Journal of the European Academy of Dermatology and Venereology, 2018, 32, e389-e392.	1.3	6

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55	A Mutation in TP63 Causing a Mild Ectodermal Dysplasia Phenotype. Journal of Investigative Dermatology, 2014, 134, 2277-2280.	0.3	5
56	Somatic Mosaicism for a â€œLethalâ€•<scp>GJB</scp>2 Mutation Results in a Patterned Form of Spiny Hyperkeratosis without Eccrine Involvement. Pediatric Dermatology, 2016, 33, 322-326.	0.5	5
57	Punctate palmoplantar keratoderma: an unusual mutation causing an unusual phenotype. British Journal of Dermatology, 2018, 178, 1455-1457.	1.4	5
58	Palmoplantar keratoderma caused by a missense variant in CTSB encoding cathepsin B. Clinical and Experimental Dermatology, 2021, 46, 103-108.	0.6	5
59	Identification of clinically useful predictive genetic variants in pachyonychia congenita. Clinical and Experimental Dermatology, 2021, 46, 867-873.	0.6	5
60	Autosomalâ€dominant cutis laxa resulting from an intronic mutation in <scp>ELN</scp>. Experimental Dermatology, 2015, 24, 885-887.	1.4	4
61	Glutathione Sâ€transferase polymorphisms in patients with photosensitive and nonâ€photosensitive drug eruptions. Photodermatology Photoimmunology and Photomedicine, 2019, 35, 214-220.	0.7	4
62	Neonatal inflammatory skin and bowel disease type 1 caused by a complex genetic defect and responsive to combined antiâ€tumour necrosis factorâ€α and interleukinâ€12/23 blockade. British Journal of Dermatology, 2022, 186, 1026-1029.	1.4	4
63	Novel POFUT 1 mutation associated with hidradenitis suppurativaâ€“Dowlingâ€“Degos disease firm up a role for Notch signalling in the pathogenesis of this disorder: reply from the authors. British Journal of Dermatology, 2018, 178, 986-986.	1.4	3
64	Loss-of-function variants in KLF4 underlie autosomal dominant palmoplantar keratoderma. Genetics in Medicine, 2022, 24, 1085-1095.	1.1	3
65	Guanine polynucleotides are selfâ€antigens for human natural autoantibodies and are significantly reduced in the human genome. Immunology, 2015, 146, 401-410.	2.0	2
66	Phenotypic suppression of acral peeling skin syndrome in a patient with autosomal recessive congenital ichthyosis. Experimental Dermatology, 2020, 29, 742-748.	1.4	2
67	Identification of a founder mutation in <scp>KRT</scp> 14 associated with Naegeliâ€“Franceschettiâ€“Jadassohn syndrome. British Journal of Dermatology, 2020, 183, 756-757.	1.4	2
68	A unique skin phenotype resulting from a large heterozygous deletion spanning six keratin genes. British Journal of Dermatology, 2022, 187, 773-777.	1.4	2
69	A case for diagnosis. Clinical and Experimental Dermatology, 2015, 40, 697-699.	0.6	1
70	Molecular epidemiology of pachyonychia congenita in the Israeli population. Clinical and Experimental Dermatology, 2021, 46, 663-668.	0.6	1
71	Epidermolytic epidermal nevus caused by a somatic mutation in KRT2. Pediatric Dermatology, 2021, 38, 538-540.	0.5	1
72	Up-regulation of ST18 in pemphigus vulgaris drives a self-amplifying p53-dependent pathomechanism resulting in decreased desmoglein 3 expression. Scientific Reports, 2022, 12, 5958.	1.6	1

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73	Heterozygous variants in the integrin subunit beta 4 gene (ITGB4) cause autosomal dominant nail dystrophy. British Journal of Dermatology, 2022, 187, 826-828.	1.4	1
74	Atypical presentation of laryngoâ€onychoâ€cutaneous syndrome resulting from novel mutations in LAMA3A. Clinical and Experimental Dermatology, 2021, 46, 990-992.	0.6	0
75	Acute Respiratory Distress Syndrome in a Carrier of an Interleukin-36 Receptor Antagonist Mutation With Generalized Pustular Psoriasis. Journal of Psoriasis and Psoriatic Arthritis, 2022, 7, 9-12.	0.3	0