Pierre Vabres

List of Publications by Year
in descending order

177	13,326	50276	24982
papers	citations	46 h-index	109 g-index

219	219	219	19709
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Autoantibodies against type I IFNs in patients with life-threatening COVID-19. Science, 2020, 370, .	12.6	1,983
2	Inborn errors of type I IFN immunity in patients with life-threatening COVID-19. Science, 2020, 370, .	12.6	1,749
3	Genomic rearrangement in NEMO impairs NF-Î²B activation and is a cause of incontinentia pigmenti. Nature, 2000, 405, 466-472.	27.8	709
4	Revised nomenclature and classification of inherited ichthyoses: Results of the First Ichthyosis Consensus Conference in SorÃ¨ze 2009. Journal of the American Academy of Dermatology, 2010, 63, 607-641.	1.2	610
5	A Randomized, Controlled Trial of Oral Propranolol in Infantile Hemangioma. New England Journal of Medicine, 2015, 372, 735-746.	27.0	601
6	A SUMOylation-defective MITF germline mutation predisposes to melanoma and renal carcinoma. Nature, 2011, 480, 94-98.	27.8	466
7	First-line rituximab combined with short-term prednisone versus prednisone alone for the treatment of pemphigus (Ritux 3): a prospective, multicentre, parallel-group, open-label randomised trial. Lancet, The, 2017, 389, 2031-2040.	13.7	438
8	Claudin-1 gene mutations in neonatal sclerosing cholangitis associated with ichthyosis: A tight junction disease. Gastroenterology, 2004, 127, 1386-1390.	1.3	378
9	Autoantibodies neutralizing type I IFNs are present in ~4% of uninfected individuals over 70 years old and account for ~20% of COVID-19 deaths. Science Immunology, 2021, 6, .	11.9	357
10	Features associated with germline CDKN2A mutations: a GenoMEL study of melanoma-prone families from three continents. Journal of Medical Genetics, 2006, 44, 99-106.	3.2	350
11	Lamin A and ZMPSTE24 (FACE-1) defects cause nuclear disorganization and identify restrictive dermopathy as a lethal neonatal laminopathy. Human Molecular Genetics, 2004, 13, 2493-2503.	2.9	325
12	High cumulative risks of cancer in patients withPTENhamartoma tumour syndrome. Journal of Medical Genetics, 2013, 50, 255-263.	3.2	290
13	Germline Loss-of-Function Mutations in EPHB4 Cause a Second Form of Capillary Malformation-Arteriovenous Malformation (CM-AVM2) Deregulating RAS-MAPK Signaling. Circulation, 2017, 136, 1037-1048.	1.6	204
14	Efficacy of Propranolol in Hepatic Infantile Hemangiomas with Diffuse Neonatal Hemangiomatosis. Journal of Pediatrics, 2010, 157, 340-342.	1.8	148
15	Clinical whole-exome sequencing for the diagnosis of rare disorders with congenital anomalies and/or intellectual disability: substantial interest of prospective annual reanalysis. Genetics in Medicine, 2018, 20, 645-654.	2.4	146
16	Mosaic Activating Mutations in GNA11 and GNAQ Are Associated with Phakomatosis Pigmentovascularis and Extensive Dermal Melanocytosis. Journal of Investigative Dermatology, 2016, 136, 770-778.	0.7	144
17	Segmental overgrowth, lipomatosis, arteriovenous malformation and epidermal nevus (SOLAMEN) syndrome is related to mosaic PTEN nullizygosity. European Journal of Human Genetics, 2007, 15, 767-773.	2.8	129
18	Novel FH mutations in families with hereditary leiomyomatosis and renal cell cancer (HLRCC) and patients with isolated type 2 papillary renal cell carcinoma. Journal of Medical Genetics, 2011, 48, 226-234.	3.2	116

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19	Safety and efficacy of low-dose sirolimus in the PIK3CA-related overgrowth spectrum. Genetics in Medicine, 2019, 21, 1189-1198.	2.4	115
20	Diagnostic and predictive value of skin testing in platinum salt hypersensitivity. Journal of Allergy and Clinical Immunology, 2007, 119, 726-730.	2.9	111
21	Propranolol for treatment of ulcerated infantile hemangiomas. Journal of the American Academy of Dermatology, 2011, 64, 827-832.	1.2	98
22	Lysosomal Signaling Licenses Embryonic Stem Cell Differentiation via Inactivation of Tfe3. Cell Stem Cell, 2019, 24, 257-270.e8.	11.1	97
23	In-Frame Mutations in Exon 1 of SKI Cause Dominant Shprintzen-Goldberg Syndrome. American Journal of Human Genetics, 2012, 91, 950-957.	6.2	95
24	Molecular diagnosis of PIK3CA-related overgrowth spectrum (PROS) in 162 patients and recommendations for genetic testing. Genetics in Medicine, 2017, 19, 989-997.	2.4	90
25	Prospective Multicenter Study of Pegylated Liposomal Doxorubicin Treatment in Patients With Advanced or Refractory Mycosis Fungoides or SÃ©zary Syndrome. Archives of Dermatology, 2008, 144, 727-33.	1.4	88
26	Prevalence of inherited ichthyosis in France: a study using capture-recapture method. Orphanet Journal of Rare Diseases, 2014, 9, 1.	2.7	87
27	The Gene for Bazex-DuprÃ©-Christol Syndrome Maps to Chromosome Xq. Journal of Investigative Dermatology, 1995, 105, 87-91.	0.7	83
28	A prospective study of risk for Sturge-Weber syndrome in children with upper facial port-wine stain. Journal of the American Academy of Dermatology, 2015, 72, 473-480.	1.2	77
29	Dacarbazine-Mediated Upregulation of NKG2D Ligands on Tumor Cells Activates NK and CD8 T Cells and Restrains Melanoma Growth. Journal of Investigative Dermatology, 2013, 133, 499-508.	0.7	75
30	The gene for the familial form of incontinentia pigmenti (IP2) maps to the distal part of Xq28. Human Molecular Genetics, 1994, 3, 273-278.	2.9	71
31	Clinical and Immunologic Factors Associated With Bullous Pemphigoid Relapse During the First Year of Treatment. JAMA Dermatology, 2014, 150, 25.	4.1	71
32	Segmental and nonsegmental childhood vitiligo has distinct clinical characteristics: A prospective observational study. Journal of the American Academy of Dermatology, 2010, 62, 945-949.	1.2	69
33	Efficacy and tolerability of methotrexate in severe childhood alopecia areata. British Journal of Dermatology, 2011, 165, 407-410.	1.5	69
34	Calculation of cutâ€off values based on the Autoimmune Bullous Skin Disorder Intensity Score () Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 1 for defining moderate, significant and extensive types of pemphigus. British Journal of Dermatology, 2016, 175, 142-149.	1.5	68
35	Integrating longitudinal serum IL-17 and IL-23 follow-up, along with autoantibodies variation, contributes to predict bullous pemphigoid outcome. Scientific Reports, 2016, 5, 18001.	3.3	66
36	Mutational spectrum of NSDHL in CHILD syndrome. Journal of Medical Genetics, 2005, 42, e17-e17.	3.2	65

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37	Juvenile Xanthogranuloma and Nevus Anemicus in the Diagnosis of Neurofibromatosis Type 1. JAMA Dermatology, 2014, 150, 42.	4.1	63
38	Mutations in ACTRT1 and its enhancer RNA elements lead to aberrant activation of Hedgehog signaling in inherited and sporadic basal cell carcinomas. Nature Medicine, 2017, 23, 1226-1233.	30.7	59
39	Homozygosity Mapping of a Locus for a Novel Syndromic Ichthyosis to Chromosome 3q27â€“q28. Journal of Investigative Dermatology, 2002, 119, 70-76.	0.7	58
40	Genotypes and Phenotypes of 162 Families with aGlomulinMutation. Molecular Syndromology, 2013, 4, 157-64.	0.8	55
41	Large International Validation of ABSIS and PDAI Pemphigus Severity Scores. Journal of Investigative Dermatology, 2019, 139, 31-37.	0.7	55
42	Rituximab is an effective treatment in patients with pemphigus vulgaris and demonstrates a steroidâ€sparing effect. British Journal of Dermatology, 2020, 182, 1111-1119.	1.5	55
43	Molecular apocrine differentiation is a common feature of breast cancer in patients with germline PTEN mutations. Breast Cancer Research, 2010, 12, R63.	5.0	54
44	Anetoderma. American Journal of Dermatopathology, 2002, 24, 118-129.	0.6	52
45	Sirolimus (Rapamycin) for Slow-Flow Malformations in Children. JAMA Dermatology, 2021, 157, 1289.	4.1	51
46	Reconstruction of hyperspectral cutaneous data from an artificial neural network-based multispectral imaging system. Computerized Medical Imaging and Graphics, 2011, 35, 85-88.	5.8	48
47	SPRED1, a RAS MAPK pathway inhibitor that causes Legius syndrome, is a tumour suppressor downregulated in paediatric acute myeloblastic leukaemia. Oncogene, 2015, 34, 631-638.	5.9	47
48	Mosaic abnormalities of the skin: review and guidelines from the European Reference Network for rare skin diseases. British Journal of Dermatology, 2020, 182, 552-563.	1.5	45
49	Psoriasis and obesity in French children: a case-control, multicentre study. British Journal of Dermatology, 2015, 172, 1593-1600.	1.5	41
50	Follow-Up of Patients With Complete Remission of Locally Advanced Basal Cell Carcinoma After Vismodegib Discontinuation: A Multicenter French Study of 116 Patients. Journal of Clinical Oncology, 2019, 37, 3275-3282.	1.6	41
51	Next-Generation Sequencing of Nevus Spilusâ€“Type Congenital Melanocytic Nevus: Exquisite Genotypeâ€“Phenotype Correlation in Mosaic RASopathies. Journal of Investigative Dermatology, 2014, 134, 2658-2660.	0.7	40
52	Biallelic pathogenic variants in the lanosterol synthase gene LSS involved in the cholesterol biosynthesis cause alopecia with intellectual disability, a rare recessive neuroectodermal syndrome. Genetics in Medicine, 2019, 21, 2025-2035.	2.4	40
53	Factors Associated With Short-term Relapse in Patients With Pemphigus Who Receive Rituximab as First-line Therapy. JAMA Dermatology, 2020, 156, 545.	4.1	40
54	Autosomal-recessive SASH1 variants associated with a new genodermatosis with pigmentation defects, palmoplantar keratoderma and skin carcinoma. European Journal of Human Genetics, 2015, 23, 957-962.	2.8	39

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55	Functional, structural, and genetic evaluation of 20CDKN2Agerm line mutations identified in melanoma-prone families or patients. Human Mutation, 2009, 30, 564-574.	2.5	38
56	Cutaneous B-cell lymphoblastic lymphoma in children: A rare diagnosis. Journal of the American Academy of Dermatology, 2012, 66, 51-57.	1.2	38
57	Hereditary mucoepithelial dysplasia: clinical, ultrastructural and genetic study of eight patients and literature review. British Journal of Dermatology, 2005, 153, 310-318.	1.5	37
58	Successful switch to dabrafenib after vemurafenib-induced toxic epidermal necrolysis. British Journal of Dermatology, 2015, 172, 1454-1455.	1.5	36
59	Etiologies and prognostic factors of leukocytoclastic vasculitis with skin involvement. Medicine (United States), 2016, 95, e4238.	1.0	36
60	Two unusual tumors in a patient with xeroderma pigmentosum: atypical fibroxanthoma and basosquamous carcinoma. Journal of Cutaneous Pathology, 1999, 26, 430-435.	1.3	35
61	Burden of Inherited Ichthyosis: A French National Survey. Acta Dermato-Venereologica, 2015, 95, 326-328.	1.3	34
62	Epidermolysis bullosa simplex generalized severe induces a T helper 17 response and is improved by apremilast treatment. British Journal of Dermatology, 2019, 180, 357-364.	1.5	34
63	The prognostic impact of the extent of lymph node dissection in patients with stage III melanoma. European Journal of Surgical Oncology, 2006, 32, 790-794.	1.0	33
64	Factors Associated with Impaired Quality of Life in Adult Patients Suffering from Ichthyosis. Acta Dermato-Venereologica, 2014, 94, 344-346.	1.3	33
65	Pain and quality of life evaluation in patients with localized epidermolysis bullosa simplex. Orphanet Journal of Rare Diseases, 2017, 12, 119.	2.7	31
66	Linkage of Marie-Unna hypotrichosis locus to chromosome 8p21 and exclusion of 10 genes including the hairless gene by mutation analysis. European Journal of Human Genetics, 2000, 8, 273-279.	2.8	30
67	Aplasia cutis congenita with dystrophic epidermolysis bullosa: clinical and mutational study. British Journal of Dermatology, 2014, 170, 901-906.	1.5	30
68	Reverse Phenotyping in Patients with Skin Capillary Malformations and Mosaic GNAQ or GNA11 Mutations Defines a Clinical Spectrum with Genotype-Phenotype Correlation. Journal of Investigative Dermatology, 2020, 140, 1106-1110.e2.	0.7	30
69	Treatment of port wine stains with pulsed dye laser and topical timolol: a multicenter randomized controlled trial. British Journal of Dermatology, 2014, 170, 1350-1353.	1.5	29
70	Skin elastic fibers in Williams syndrome. American Journal of Medical Genetics Part A, 1999, 87, 134-138.	2.4	28
71	Sensitivity and specificity of BP180 NC16A enzyme-linked immunosorbent assay for the diagnosis of pemphigoid gestationis. Journal of the American Academy of Dermatology, 2017, 76, 560-562.	1.2	27
72	Fixed eruption due to quinine contained in tonic water: positive patchâ€testing. Contact Dermatitis, 2009, 61, 242-244.	1.4	26

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73	Familial melanoma: Clinical factors associated with germline CDKN2A mutations according to the number of patients affected by melanoma in a family. Journal of the American Academy of Dermatology, 2012, 67, 1257-1264.e2.	1.2	26
74	Methotrexate in Severe Childhood Alopecia Areata: Long-term Follow-up. Acta Dermato-Venereologica, 2016, 96, 102-103.	1.3	26
75	The contribution of large genomic deletions at the CDKN2A locus to the burden of familial melanoma. British Journal of Cancer, 2008, 99, 364-370.	6.4	25
76	Postzygotic inactivating mutations of RHOA cause a mosaic neuroectodermal syndrome. Nature Genetics, 2019, 51, 1438-1441.	21.4	25
77	â€˜Matchstickâ€™ eyebrow hairs: a dermoscopic clue to the diagnosis of Netherton syndrome. Journal of the European Academy of Dermatology and Venereology, 2010, 24, 740-741.	2.4	24
78	Systematic molecular and cytogenetic screening of 100 patients with marfanoid syndromes and intellectual disability. Clinical Genetics, 2013, 84, 507-521.	2.0	23
79	Mosaicâ€activating FGFR2 mutation in two fetuses with papillomatous pedunculated sebaceous naevus. British Journal of Dermatology, 2017, 176, 204-208.	1.5	23
80	Microchimerism from a dizygotic twin in juvenile ulcerative lichen planus. Lancet, The, 2002, 359, 1861-1862.	13.7	22
81	Systematic search for neutropenia should be part of the first screening in patients with poikiloderma. European Journal of Medical Genetics, 2012, 55, 8-11.	1.3	22
82	Search for <scp>ReCQL4</scp> mutations in 39 patients genotyped for suspected Rothmundâ€“Thomson/Ballerâ€Gerold syndromes. Clinical Genetics, 2015, 87, 244-251.	2.0	22
83	Bazex-DuprÃ©-Christol syndrome: A possible diagnosis for basal cell carcinomas, coarse sparse hair, and milia. American Journal of Medical Genetics Part A, 1993, 45, 786-786.	2.4	21
84	Treatment of voluminous and complicated superficial slow-flow vascular malformations with sirolimus (PERFORMUS): protocol for a multicenter phase 2 trial with a randomized observational-phase design. Trials, 2018, 19, 340.	1.6	21
85	A standard of care for individuals with <scp>PIK3CA</scp>â€related disorders: An international expert consensus statement. Clinical Genetics, 2022, 101, 32-47.	2.0	21
86	Retinal angioma in a patient with Cowden disease. American Journal of Ophthalmology, 2003, 135, 400-402.	3.3	20
87	The scalp hair collar and tuft signs: A retrospective multicenter study of 78 patients with a systematic review of the literature. Journal of the American Academy of Dermatology, 2017, 76, 478-487.	1.2	19
88	A Novel Missense Mutation, A118E, in the Helix Initiation Motif of the Type II Hair Cortex Keratin hHb6, Causing Monilethrix. Human Heredity, 2000, 50, 322-324.	0.8	18
89	Bi-acromial Dimples: A Series of Seven Cases. Pediatric Dermatology, 2005, 22, 412-414.	0.9	18
90	Anetoderma of Prematurity: An Iatrogenic Consequence of Neonatal Intensive Care. Archives of Dermatology, 2010, 146, 565-7.	1.4	18

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91	Pathway-Based Analysis of a Melanoma Genome-Wide Association Study: Analysis of Genes Related to Tumour-Immunosuppression. PLoS ONE, 2011, 6, e29451.	2.5	18
92	Diversity of the clinical presentation of the MMR gene biallelic mutations. Familial Cancer, 2014, 13, 131-135.	1.9	18
93	Reliability, validity and feasibility of nail ultrasonography in psoriatic arthritis. Joint Bone Spine, 2016, 83, 539-544.	1.6	18
94	Mosaicism for a KITLG Mutation in Linear and Whorled Nevoid Hypermelanosis. Journal of Investigative Dermatology, 2017, 137, 1575-1578.	0.7	18
95	Secondary actionable findings identified by exome sequencing: expected impact on the organisation of care from the study of 700 consecutive tests. European Journal of Human Genetics, 2019, 27, 1197-1214.	2.8	18
96	A Typical Vascular and Pigmentary Dermoscopic Pattern of Capillary Malformations in Capillary Malformationâ€“Arteriovenous Malformation Syndrome: Report of Four Cases. Pediatric Dermatology, 2016, 33, e337-41.	0.9	17
97	Topical sirolimus 0.1% for treating cutaneous microcystic lymphatic malformations in children and adults (TOPICAL): protocol for a multicenter phase 2, within-person, randomized, double-blind, vehicle-controlled clinical trial. Trials, 2019, 20, 739.	1.6	17
98	Kosaki overgrowth syndrome: A novel pathogenic variant in PDGFRB and expansion of the phenotype including cerebrovascular complications. Clinical Genetics, 2020, 98, 19-31.	2.0	17
99	Active tuberculosis in psoriasis patients treated with <scp>TNF</scp> antagonists: a French nationwide retrospective study. Journal of the European Academy of Dermatology and Venereology, 2016, 30, 1336-1341.	2.4	16
100	Congenital Plaqueâ€Type Glomuvenous Malformations Associated with Fetal Pleural Effusion and Ascites. Pediatric Dermatology, 2011, 28, 528-531.	0.9	15
101	Severe Xâ€linked chondrodysplasia punctata in nine new female fetuses. Prenatal Diagnosis, 2015, 35, 675-684.	2.3	15
102	Postzygotic BRAF p.Lys601Asn Mutation in Phacomatosis Pigmentokeratotica with Woolly Hair Nevus and Focal Cortical Dysplasia. Journal of Investigative Dermatology, 2016, 136, 1060-1062.	0.7	15
103	Acute acral eruptions in children during the COVID-19 pandemic: Characteristics of 103 children and their family clusters. Annales De Dermatologie Et De Venereologie, 2021, 148, 94-100.	1.0	15
104	Dermoscopy of Longitudinal Leukonychia in Hailey-Hailey Disease. Archives of Dermatology, 2010, 146, 1204.	1.4	14
105	Quinoline Yellow dyeâ€induced fixed foodâ€andâ€drug eruption. Contact Dermatitis, 2013, 68, 187-188.	1.4	14
106	Prospective Study of the Evolution of Blood Lymphoid Immune Parameters during Dacarbazine Chemotherapy in Metastatic and Locally Advanced Melanoma Patients. PLoS ONE, 2014, 9, e105907.	2.5	14
107	2.5 yearsâ€™ experience of GeneMatcher data-sharing: a powerful tool for identifying new genes responsible for rare diseases. Genetics in Medicine, 2019, 21, 1657-1661.	2.4	14
108	Clinical spectrum of MTOR-related hypomelanosis of Ito with neurodevelopmental abnormalities. Genetics in Medicine, 2021, 23, 1484-1491.	2.4	14

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109	Oral epigallocatechin-3-gallate for treatment of dystrophic epidermolysis bullosa: a multicentre, randomized, crossover, double-blind, placebo-controlled clinical trial. Orphanet Journal of Rare Diseases, 2016, 11, 31.	2.7	13
110	Lack of interaction between NEMO and SHARPIN impairs linear ubiquitination and NF-Î²B activation and leads to incontinentia pigmenti. Journal of Allergy and Clinical Immunology, 2017, 140, 1671-1682.e2.	2.9	13
111	Medial Fronto-Facial Capillary Malformations. Journal of Pediatrics, 2011, 158, 836-841.	1.8	12
112	Clinical and neuroimaging findings in 33 patients with <scp>MCAP</scp> syndrome: A survey to evaluate relevant endpoints for future clinical trials. Clinical Genetics, 2021, 99, 650-661.	2.0	12
113	Safety and efficacy of low-dose PI3K inhibitor taselisib in adult patients with CLOVES and Klippelâ€“Trenaunay syndrome (KTS): the TOTEM trial, a phase 1/2 multicenter, open-label, single-arm study. Genetics in Medicine, 2021, 23, 2433-2442.	2.4	12
114	Incontinentia Pigmenti: Late Sequelae and Genotypic Diagnosis: A Three-Generation Study of Four Patients. Pediatric Dermatology, 1995, 12, 107-111.	0.9	11
115	Atypical Lyme borreliosis in an HIV-infected man. British Journal of Dermatology, 1997, 137, 437-439.	1.5	11
116	Polyethylenimine-mediated in vivo gene transfer of a transmembrane superantigen fusion construct inhibits B16 murine melanoma growth. Cancer Gene Therapy, 2008, 15, 742-749.	4.6	11
117	Deciphering exome sequencing data: Bringing mitochondrial DNA variants to light. Human Mutation, 2019, 40, 2430-2443.	2.5	11
118	De novo mutations in the X-linked TFE3 gene cause intellectual disability with pigmentary mosaicism and storage disorder-like features. Journal of Medical Genetics, 2020, 57, 808-819.	3.2	11
119	Atypical Lyme borreliosis in an HIV-infected man. British Journal of Dermatology, 1997, 137, 437-439.	1.5	11
120	Flexural Agminated Eruptive Nevi in Langerhans Cell Histiocytosis. JAMA Dermatology, 2013, 149, 635.	4.1	10
121	Linear and whorled nevoid hypermelanosis with bilateral giant cerebral aneurysms. American Journal of Medical Genetics Part A, 2002, 112, 95-98.	2.4	9
122	Primary cutaneous B-cell lymphoma mimicking pyoderma gangrenosum: first-line treatment with rituximab. British Journal of Dermatology, 2004, 151, 250-252.	1.5	9
123	Linear atrophoderma of moulin associated with antinuclear antibodies. Journal of the European Academy of Dermatology and Venereology, 2010, 24, 108-109.	2.4	9
124	Validation of a 2D multispectral camera: application to dermatology/cosmetology on a population covering five skin phototypes. , 2011, , .		9
125	Treatment of granuloma annulare with the 595â€nm pulsed dye laser, a multicentre retrospective study with longâ€term followâ€up. Journal of the European Academy of Dermatology and Venereology, 2013, 27, 785-788.	2.4	9
126	Extending the ALDH18A1 clinical spectrum to severe autosomal recessive fetal cutis laxa with corpus callosum agenesis. American Journal of Medical Genetics, Part A, 2018, 176, 2509-2512.	1.2	9

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127	Congenital infiltrating lipomatosis of the face with lingual mucosal neuromas associated with a PIK3CA mutation. Pediatric Dermatology, 2020, 37, 1128-1130.	0.9	9
128	Childhood Dermatosis due to Microchimerism. Dermatology, 2005, 211, 388-389.	2.1	8
129	Genetical, clinical, and functional analysis of a large international cohort of patients with autosomal recessive congenital ichthyosis due to mutations in NIPAL4. Human Mutation, 2019, 40, 2318-2333.	2.5	8
130	Compassionate use of everolimus for refractory epilepsy in a patient with MTOR mosaic mutation. European Journal of Medical Genetics, 2020, 63, 104036.	1.3	8
131	Oral erythromycin therapy in epidermolysis bullosa simplex generalized severe. British Journal of Dermatology, 2015, 173, 563-564.	1.5	7
132	Atypical dermal melanocytosis: a diagnostic clue in constitutional mismatch repair deficiency syndrome. British Journal of Dermatology, 2017, 177, e185-e186.	1.5	7
133	ARP-T1-associated Bazexâ€“DuprÃ©â€“Christol syndrome is an inherited basal cell cancer with ciliary defects characteristic of ciliopathies. Communications Biology, 2021, 4, 544.	4.4	7
134	The oculoauriculofrontonasal syndrome: Further clinical characterization and additional evidence suggesting a nontraditional mode of inheritance. American Journal of Medical Genetics, Part A, 2018, 176, 2740-2750.	1.2	6
135	Cerebriform sebaceous nevus: a subtype of organoid nevus due to specific postzygotic FGFR2 mutations. Journal of the European Academy of Dermatology and Venereology, 2021, 35, 2085-2090.	2.4	6
136	Clinical and molecular data in cases of prenatal localized overgrowth disorder: major implication of genetic variants in <scp>PI3Kâ€AKTâ€mTOR</scp> signaling pathway. Ultrasound in Obstetrics and Gynecology, 2022, 59, 532-542.	1.7	6
137	Pitfalls in Clinical Diagnosis of Female Carriers of X-linked Hypohidrotic Ectodermal Dysplasia. Archives of Dermatology, 2002, 138, 1256-1258.	1.4	6
138	Diagnostic value of nail examination in Hailey-Hailey disease. European Journal of Dermatology, 2014, 24, 628-629.	0.6	5
139	Mutations in the ERCC2 (XPD) gene associated with severe fetal ichthyosis and dysmorphic features. Prenatal Diagnosis, 2016, 36, 1276-1279.	2.3	5
140	Tongue psoriasis: Clinical aspects and analysis of epidemiological associations in 313 children, with a systematic literature review. Annales De Dermatologie Et De Venereologie, 2018, 145, 578-586.	1.0	5
141	Mosaicism due to postzygotic mutations in women with focal dermal hypoplasia. British Journal of Dermatology, 2019, 180, 657-661.	1.5	5
142	Neutralization of HSF1 in cells from PIK3CA-related overgrowth spectrum patients blocks abnormal proliferation. Biochemical and Biophysical Research Communications, 2020, 530, 520-526.	2.1	5
143	Health care transition for patients with vascular malformations: a French multicenter cross-sectional study. Orphanet Journal of Rare Diseases, 2021, 16, 352.	2.7	5
144	Multiple anaemic macules and diffuse erythrocyanosis revealing mixed cryoglobulinaemia. European Journal of Dermatology, 2011, 21, 269-270.	0.6	5

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145	Source separation on hyperspectral cube applied to dermatology. , 2010, , .		4
146	Limb overgrowth associated with a mosaic <scp>TSC2</scp> secondâ€hit in tuberous sclerosis complex. American Journal of Medical Genetics, Part A, 2020, 182, 2803-2804.	1.2	4
147	Follow-up of patients with complete remission of locally advanced basal cell carcinoma treated with vismodegib after treatment discontinuation: A retrospective multicentric French study.. Journal of Clinical Oncology, 2017, 35, 9535-9535.	1.6	4
148	Clinical and haemodynamic risk factors associated with discrepancies in lower limb length with capillary malformations: data from the national paediatric French cohort CONAPE. British Journal of Dermatology, 2018, 178, 520-526.	1.5	3
149	Search for RASA1 Variants in Capillary Malformations of the Legs in 113 Children: Results from the French National Paediatric Cohort CONAPE. Acta Dermato-Venereologica, 2018, 98, 251-255.	1.3	3
150	Severe gynaecological involvement in Proteus Syndrome. European Journal of Medical Genetics, 2019, 62, 270-272.	1.3	3
151	Absence of Lisch Nodules in Sporadic Neurofibromatosis Type 1 May Reflect Somatic Mosaicism. Archives of Dermatology, 2002, 138, 839-840.	1.4	3
152	Atypical Lyme borreliosis in an HIV-infected man. British Journal of Dermatology, 1997, 137, 437-9.	1.5	3
153	Vitamine D et soleil : risques et bÃ©nÃ©fices chez lâ€™enfant. Annales De Dermatologie Et De Venereologie, 2007, 134, 14-17.	1.0	2
154	Validation of a clinical evaluation score for irritative dermatitis: SCOREPI. Journal of the European Academy of Dermatology and Venereology, 2013, 27, 1138-1142.	2.4	2
155	Genetic Testing for Melanomaâ€"Where Are We With Moderate-Penetrance Genes?. JAMA Dermatology, 2016, 152, 375.	4.1	2
156	Skin conditions among 20th century politicians and world leaders. Journal of the European Academy of Dermatology and Venereology, 2021, 35, 2346-2348.	2.4	2
157	Fertility in McCune Albright syndrome female: A case study focusing on AMH as a marker of ovarian dysfunction and a literature review. Journal of Gynecology Obstetrics and Human Reproduction, 2021, 50, 102171.	1.3	2
158	Remembering Marc LarrÃ¨gue (1935â€“2007). Pediatric Dermatology, 2008, 25, 581-583.	0.9	1
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