

# Philippe Touraine

## List of Publications by Year in descending order

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Version: 2024-02-01

65  
papers

2,748  
citations

201674

27  
h-index

189892

50  
g-index

68  
all docs

68  
docs citations

68  
times ranked

2731  
citing authors

#	ARTICLE	IF	CITATIONS
1	Congenital Adrenal Hyperplasiaâ€”Current Insights in Pathophysiology, Diagnostics, and Management. <i>Endocrine Reviews</i> , 2022, 43, 91-159.	20.1	182
2	Causal and Candidate Gene Variants in a Large Cohort of Women With Primary Ovarian Insufficiency. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2022, 107, 685-714.	3.6	13
3	Meiotic genes in premature ovarian insufficiency: variants in HROB and REC8 as likely genetic causes. <i>European Journal of Human Genetics</i> , 2022, 30, 219-228.	2.8	18
4	Long-term outcomes of lentiviral gene therapy for the $\beta^0$ -hemoglobinopathies: the HGB-205 trial. <i>Nature Medicine</i> , 2022, 28, 81-88.	30.7	53
5	Whole exome sequencing reveals copy number variants in individuals with disorders of sex development. <i>Molecular and Cellular Endocrinology</i> , 2022, 546, 111570.	3.2	4
6	Pubertal induction and transition to adult sex hormone replacement in patients with congenital pituitary or gonadal reproductive hormone deficiency: an Endo-ERN clinical practice guideline. <i>European Journal of Endocrinology</i> , 2022, 186, G9-G49.	3.7	25
7	Long-term Safety of Growth Hormone in Adults With Growth Hormone Deficiency: Overview of 15 809 GH-Treated Patients. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2022, 107, 1906-1919.	3.6	12
8	Prenatal dexamethasone treatment for classic 21-hydroxylase deficiency in Europe. <i>European Journal of Endocrinology</i> , 2022, 186, K17-K24.	3.7	7
9	High Prevalence of Early Endocrine Disorders After Childhood Brain Tumors in a Large Cohort. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2022, 107, e2156-e2166.	3.6	6
10	Safety of growth hormone replacement in survivors of cancer and intracranial and pituitary tumours: a consensus statement. <i>European Journal of Endocrinology</i> , 2022, 186, P35-P52.	3.7	42
11	Differences or Disorders of sex development in Boys: impact on fertility. <i>Annales D'Endocrinologie</i> , 2022, , .	1.4	0
12	Infertility with hypogonadotropic hypogonadism revealing a classic form of 21 hydroxylase deficiency in a 39 year-old man. <i>Annales D'Endocrinologie</i> , 2022, , .	1.4	0
13	Identification of predictive criteria for pathogenic variants of primary bilateral macronodular adrenal hyperplasia (PBMAH) gene <i>ARMC5</i> in 352 unselected patients. <i>European Journal of Endocrinology</i> , 2022, 187, 123-134.	3.7	18
14	Sperm cryopreservation in young males with congenital adrenal hyperplasia (CAH). <i>Clinical Endocrinology</i> , 2022, 97, 860-862.	2.4	2
15	Turner syndrome: French National Diagnosis and Care Protocol (NDCP; National Diagnosis and Care) Tj ETQq1 1 0.784314 rgBT / Overbo 2.7 12	2.7	12
16	Transition of young adults with endocrine and metabolic diseases: the â€”TRANSENDâ€” cohort. <i>Endocrine Connections</i> , 2021, 10, 21-28.	1.9	9
17	Effects of mitotane on testicular adrenal rest tumors in congenital adrenal hyperplasia due to 21-hydroxylase deficiency: a retrospective series of five patients. <i>European Journal of Endocrinology</i> , 2021, 184, 365-371.	3.7	8
18	Next Generation Sequencing Should Be Proposed to Every Woman With â€”Idiopathicâ€”Primary Ovarian Insufficiency. <i>Journal of the Endocrine Society</i> , 2021, 5, bvab032.	0.2	30

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19	Modified-Release Hydrocortisone in Congenital Adrenal Hyperplasia. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2021, 106, e2063-e2077.	3.6	38
20	A recessive variant in TFAM causes mtDNA depletion associated with primary ovarian insufficiency, seizures, intellectual disability and hearing loss. <i>Human Genetics</i> , 2021, 140, 1733-1751.	3.8	15
21	Age at diagnosis in patients with chronic congenital endocrine conditions: a regional cohort study from a reference center for rare diseases. <i>Orphanet Journal of Rare Diseases</i> , 2021, 16, 469.	2.7	2
22	GGPS1 Mutations Cause Muscular Dystrophy/Hearing Loss/Ovarian Insufficiency Syndrome. <i>Annals of Neurology</i> , 2020, 88, 332-347.	5.3	22
23	Genomic sequencing highlights the diverse molecular causes of Perrault syndrome: a peroxisomal disorder (PEX6), metabolic disorders (CLPP, GGPS1), and mtDNA maintenance/translation disorders (LARS2, TFAM). <i>Human Genetics</i> , 2020, 139, 1325-1343.	3.8	21
24	Effect of congenital adrenal hyperplasia treated by glucocorticoids on plasma metabolome: a machine-learning-based analysis. <i>Scientific Reports</i> , 2020, 10, 8859.	3.3	2
25	Premature ovarian insufficiency: step-by-step genetics bring new insights. <i>Fertility and Sterility</i> , 2020, 113, 767-768.	1.0	8
26	Diabetes Mellitus, Extreme Insulin Resistance, and Hypothalamic-Pituitary Langerhans Cells Histiocytosis. <i>Case Reports in Endocrinology</i> , 2019, 2019, 1-8.	0.4	1
27	Illicit Upregulation of Serotonin Signaling Pathway in Adrenals of Patients With High Plasma or Intra-Adrenal ACTH Levels. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2019, 104, 4967-4980.	3.6	15
28	TP63 truncating variants cause isolated premature ovarian insufficiency. <i>Human Mutation</i> , 2019, 40, 886-892.	2.5	29
29	Early central blood pressure elevation in adult patients with 21-hydroxylase deficiency. <i>Journal of Hypertension</i> , 2019, 37, 175-181.	0.5	10
30	Gene variants identified by whole-exome sequencing in 33 French women with premature ovarian insufficiency. <i>Journal of Assisted Reproduction and Genetics</i> , 2019, 36, 39-45.	2.5	35
31	SAT-444 Pituitary Function and the Response to GH Replacement Therapy in Patients with Histiocytosis: Analysis of the Pfizer International Metabolic Database (KIMS). <i>Journal of the Endocrine Society</i> , 2019, 3, .	0.2	0
32	A common African variant of human connexin43 is associated with Caucasian primary ovarian insufficiency and has a deleterious effect in vitro. <i>International Journal of Molecular Medicine</i> , 2018, 41, 640-648.	4.0	6
33	Complex Association of Sex Hormones on Left Ventricular Systolic Function: Insight into Sexual Dimorphism. <i>Journal of the American Society of Echocardiography</i> , 2018, 31, 231-240.e1.	2.8	13
34	Surgery is not superior to dilation for the management of vaginal agenesis in Mayer-Rokitansky-Küster-Hauser syndrome: a multicenter comparative observational study in 131 patients. <i>American Journal of Obstetrics and Gynecology</i> , 2018, 219, 281.e1-281.e9.	1.3	45
35	Hypogonadism as a Reversible Cause of Torsades de Pointes in Men. <i>Circulation</i> , 2018, 138, 110-113.	1.6	57
36	Increased long QT and torsade de pointes reporting on tamoxifen compared with aromatase inhibitors. <i>Heart</i> , 2018, 104, 1859-1863.	2.9	37

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37	<i>MCM8</i> and <i>MCM9</i> Nucleotide Variants in Women with Primary Ovarian Insufficiency. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2017, 102, jc.2016-2565.	3.6	68
38	Long-term outcome of ovarian function in women with intermittent premature ovarian insufficiency. <i>Clinical Endocrinology</i> , 2017, 86, 223-228.	2.4	42
39	MANAGEMENT OF ENDOCRINE DISEASE: Congenital adrenal hyperplasia due to 21-hydroxylase deficiency: update on the management of adult patients and prenatal treatment. <i>European Journal of Endocrinology</i> , 2017, 176, R167-R181.	3.7	49
40	Impact of transition on quality of life in patients with congenital adrenal hyperplasia diagnosed during childhood. <i>Endocrine Connections</i> , 2017, 6, 422-429.	1.9	27
41	Managing Transition in Patients Treated with Growth Hormone. <i>Frontiers in Endocrinology</i> , 2017, 8, 346.	3.5	12
42	Poor Compliance to Hormone Therapy and Decreased Bone Mineral Density in Women with Premature Ovarian Insufficiency. <i>PLoS ONE</i> , 2016, 11, e0164638.	2.5	23
43	Premature Ovarian Insufficiency: New Perspectives on Genetic Cause and Phenotypic Spectrum. <i>Endocrine Reviews</i> , 2016, 37, 609-635.	20.1	170
44	An Application of NGS for Molecular Investigations in Perrault Syndrome: Study of 14 Families and Review of the Literature. <i>Human Mutation</i> , 2016, 37, 1354-1362.	2.5	46
45	Complex Influence of Gonadotropins and Sex Steroid Hormones on QT Interval Duration. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2016, 101, 2776-2784.	3.6	46
46	Prevalence of and Risk Factors for Anal Oncogenic Human Papillomavirus Infection Among HIV-Infected Women in France in the Combination Antiretroviral Therapy Era. <i>Journal of Infectious Diseases</i> , 2016, 213, 1455-1461.	4.0	28
47	Spontaneous fertility and pregnancy outcomes amongst 480 women with Turner syndrome. <i>Human Reproduction</i> , 2016, 31, 782-788.	0.9	158
48	Postprandial GLP-1 Secretion After Bariatric Surgery in Three Cases of Severe Obesity Related to Craniopharyngiomas. <i>Obesity Surgery</i> , 2016, 26, 1133-1137.	2.1	14
49	Pregnancy in Women Previously Treated for an Adrenocortical Carcinoma. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2015, 100, 4604-4611.	3.6	19
50	Determining clinical and biological indicators for health outcomes in adult patients with childhood onset of congenital adrenal hyperplasia. <i>European Journal of Endocrinology</i> , 2015, 173, 175-184.	3.7	36
51	Gynecologic follow up of 129 women on dialysis and after kidney transplantation: a retrospective cohort study. <i>European Journal of Obstetrics, Gynecology and Reproductive Biology</i> , 2015, 187, 1-5.	1.1	24
52	Clinical Outcome, Hormonal Status, Gonadotrope Axis, and Testicular Function in 219 Adult Men Born With Classic 21-Hydroxylase Deficiency. A French National Survey. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2015, 100, 2303-2313.	3.6	94
53	Water and Electrolyte Disorders at Long-Term Post-Treatment Follow-Up in Paediatric Patients with Suprasellar Tumours Include Unexpected Persistent Cerebral Salt-Wasting Syndrome. <i>Hormone Research in Paediatrics</i> , 2014, 82, 364-371.	1.8	20
54	Monocentric study of 112 consecutive patients with childhood onset GH deficiency around and after transition. <i>European Journal of Endocrinology</i> , 2013, 169, 587-596.	3.7	19

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55	Is prolactin involved in the evolution of atherothrombotic disease?. Expert Review of Endocrinology and Metabolism, 2012, 7, 345-361.	2.4	3
56	Transition from Pediatric to Adult Healthcare: Assessment of Specific Needs of Patients with Chronic Endocrine Conditions. Hormone Research in Paediatrics, 2012, 78, 247-255.	1.8	44
57	Resumption of Ovarian Function and Pregnancies in 358 Patients with Premature Ovarian Failure. Journal of Clinical Endocrinology and Metabolism, 2011, 96, 3864-3872.	3.6	157
58	Novel NOBOX loss-of-function mutations account for 6.2% of cases in a large primary ovarian insufficiency cohort. Human Mutation, 2011, 32, 1108-1113.	2.5	94
59	Fertility in Women with Nonclassical Congenital Adrenal Hyperplasia due to 21-Hydroxylase Deficiency. Journal of Clinical Endocrinology and Metabolism, 2010, 95, 1182-1190.	3.6	180
60	Phenotyping and genetic studies of 357 consecutive patients presenting with premature ovarian failure. European Journal of Endocrinology, 2009, 161, 179-187.	3.7	125
61	Clinical and Molecular Characterization of a Cohort of 161 Unrelated Women with Nonclassical Congenital Adrenal Hyperplasia Due to 21-Hydroxylase Deficiency and 330 Family Members. Journal of Clinical Endocrinology and Metabolism, 2009, 94, 1570-1578.	3.6	186
62	Genetic investigation of four meiotic genes in women with premature ovarian failure. European Journal of Endocrinology, 2008, 158, 107-115.	3.7	111
63	Premature ovarian failure: predictability of intermittent ovarian function and response to ovulation induction agents. Current Opinion in Obstetrics and Gynecology, 2008, 20, 416-420.	2.0	46
64	Long-Term Outcome of Patients with Congenital Adrenal Hyperplasia due to 21-Hydroxylase Deficiency. Hormone Research in Paediatrics, 2007, 67, 268-276.	1.8	80
65	Ovarian Steroidogenesis and Serum Androgen Levels in Patients with Premature Ovarian Failure. Journal of Clinical Endocrinology and Metabolism, 2005, 90, 2391-2396.	3.6	26