# Bart Janssen

## List of Publications by Year in descending order

Source: https://exaly.com/author-pdf/11015365/publications.pdf

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|---|---|---|---|
| 33 papers | 4,104 citations | 236925 25 h-index | 395702 33 g-index |
| 33 all docs | 33 docs citations | 33 times ranked | 3811 citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Identification of the Tuberous Sclerosis Gene TSC1 on Chromosome 9q34. Science, 1997, 277, 805-808. | 12.6 | 1,550 |
| 2 | Mutations of the TGF-Î² type II receptorBMPR2 in pulmonary arterial hypertension. Human Mutation, 2006, 27, 121-132. | 2.5 | 368 |
| 3 | Carnosine as a Protective Factor in Diabetic Nephropathy. Diabetes, 2005, 54, 2320-2327. | 0.6 | 264 |
| 4 | Abnormal Pulmonary Artery Pressure Response in Asymptomatic Carriers of Primary Pulmonary Hypertension Gene. Circulation, 2000, 102, 1145-1150. | 1.6 | 235 |
| 5 | Genetic basis of pulmonary arterial hypertension. Journal of the American College of Cardiology, 2004, 43, S33-S39. | 2.8 | 227 |
| 6 | Stress Doppler Echocardiography in Relatives of Patients With Idiopathic and Familial Pulmonary Arterial Hypertension. Circulation, 2009, 119, 1747-1757. | 1.6 | 205 |
| 7 | Gene for susceptibility to diabetic nephropathy in type 2 diabetes maps to 18q22.3-23. Kidney International, 2002, 62, 2176-2183. | 5.2 | 144 |
| 8 | Sporadic Imprinting Defects in Prader-Willi Syndrome and Angelman Syndrome: Implications for Imprint-Switch Models, Genetic Counseling, and Prenatal Diagnosis. American Journal of Human Genetics, 1998, 63, 170-180. | 6.2 | 142 |
| 9 | A leucine repeat in the carnosinase gene CNDP1 is associated with diabetic end-stage renal disease in European Americans. Nephrology Dialysis Transplantation, 2007, 22, 1131-1135. | 0.7 | 111 |
| 10 | Genetic Association of the Serotonin Transporter in Pulmonary Arterial Hypertension. American Journal of Respiratory and Critical Care Medicine, 2006, 173, 793-797. | 5.6 | 88 |
| 11 | LargeBRCA1 gene deletions are found in 3% of German high-risk breast cancer families. Human Mutation, 2004, 24, 534-534. | 2.5 | 69 |
| 12 | A CTG Polymorphism in the CNDP1 Gene Determines the Secretion of Serum Carnosinase in Cos-7â€“Transfected Cells. Diabetes, 2007, 56, 2410-2413. | 0.6 | 58 |
| 13 | Quantification of the methylation status of the PWS/AS imprinted region: Comparison of two approaches based on bisulfite sequencing and methylation-sensitive MLPA. Molecular and Cellular Probes, 2007, 21, 208-215. | 2.1 | 54 |
| 14 | TSC1 and TSC2 mutations in tuberous sclerosis, the associated phenotypes and a model to explain observed TSC1/TSC2 frequency ratios. European Journal of Pediatrics, 2002, 161, 393-402. | 2.7 | 52 |
| 15 | Primary Pulmonary Hypertension in Children May Have a Different Genetic Background Than in Adults. Pediatric Research, 2004, 56, 571-578. | 2.3 | 49 |
| 16 | Anserine inhibits carnosine degradation but in human serum carnosinase (CN1) is not correlated with histidine dipeptide concentration. Clinica Chimica Acta, 2011, 412, 263-267. | 1.1 | 47 |
| 17 | Coding and small non-coding transcriptional landscape of tuberous sclerosis complex cortical tubers: implications for pathophysiology and treatment. Scientific Reports, 2017, 7, 8089. | 3.3 | 47 |
| 18 | DetectingPKD1variants in polycystic kidney disease patients by single-molecule long-read sequencing. Human Mutation, 2017, 38, 870-879. | 2.5 | 44 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | Refined localization of TSC1 by combined analysis of 9q34 and 16pl3 data in 14 tuberous sclerosis families. Human Genetics, 1994, 94, 437-440. | 3.8 | 39 |
| 20 | Association Between CNDP1 Genotype and Diabetic Nephropathy Is Sex Specific. Diabetes, 2010, 59, 1555-1559. | 0.6 | 39 |
| 21 | Relevance of allosteric conformations and homocarnosine concentration on carnosinase activity. Amino Acids, 2010, 38, 1607-1615. | 2.7 | 36 |
| 22 | Identification of a subtle t(16;19)(p13.3;p13.3) in an infant with multiple congenital abnormalities using a 12-colour multiplex FISH telomere assay, M-TEL. European Journal of Human Genetics, 2000, 8, 903-910. | 2.8 | 35 |
| 23 | N-Glycosylation of Carnosinase Influences Protein Secretion and Enzyme Activity. Diabetes, 2010, 59, 1984-1990. | 0.6 | 35 |
| 24 | Primary pulmonary hypertension may be a heterogeneous disease with a second locus on chromosome 2q31. Journal of the American College of Cardiology, 2003, 41, 2237-2244. | 2.8 | 29 |
| 25 | Sequence Variants in <i>BMPR2</i> and Genes Involved in the Serotonin and Nitric Oxide Pathways in Idiopathic Pulmonary Arterial Hypertension and Chronic Thromboembolic Pulmonary Hypertension: Relation to Clinical Parameters and Comparison with Left Heart Disease. Respiration, 2010, 79, 279-287. | 2.6 | 27 |
| 26 | Enhanced Hypoxic Pulmonary Vasoconstriction in Families of Adults or Children With Idiopathic Pulmonary Arterial Hypertension. Chest, 2005, 128, 630S-633S. | 0.8 | 25 |
| 27 | Molecular diagnosis of type 1c glycogen storage disease. Human Genetics, 1999, 104, 275-277. | 3.8 | 19 |
| 28 | Linkage Analysis in a Large Family With Primary Pulmonary Hypertension. Chest, 2002, 121, 54S-56S. | 0.8 | 18 |
| 29 | Problems in detecting mosaic DNA methylation in Angelman syndrome. European Journal of Human Genetics, 2003, 11, 913-915. | 2.8 | 12 |
| 30 | CNDP1 genotype and renal survival in pediatric nephropathies. Journal of Pediatric Endocrinology and Metabolism, 2016, 29, 827-33. | 0.9 | 10 |
| 31 | The Kruppel-like zinc-finger gene ZNF236 is alternatively spliced and excluded as susceptibility gene for diabetic nephropathy. Genomics, 2003, 82, 406-411. | 2.9 | 9 |
| 32 | Allelic variation in the CNDP1 gene and its lack of association with longevity and coronary heart disease. Mechanisms of Ageing and Development, 2006, 127, 817-820. | 4.6 | 9 |
| 33 | Genomeâ€wide characterization of 5â€hydoxymethylcytosine in melanoma reveals major differences with nevus. Genes Chromosomes and Cancer, 2020, 59, 366-374. | 2.8 | 8 |