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## Grzegorz M Burzynski

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Web of Science ResearcherID: D-2800-2009

#### Current affiliation:

- Johns Hopkins Medicine until present

### **Publications**

#### **PUBLICATION METRICS**

For manuscripts published from date range January 2001 - January 2023

TOTAL TIMES CITED H-INDEX PUBLICATIONS

1373 | 13 | 21

**CORE COLLECTION DOCUMENTS** 

21

#### For all time

TOTAL TIMES CITED	H-INDEX	PUBLICATIONS

1373 | 13 | 21

**CORE COLLECTION DOCUMENTS** 

21

#### **MANUSCRIPTS PUBLISHED (21)**

From date range January 2001 - January 2023

TIMES CITED (ALL TIME)

4

# Integration of genomic and functional approaches reveals enhancers at LMX1A and LMX1B

Authors (5): Burzynski, Grzegorz M.; Reed, Xylena ... McCallion, Andrew S. Published in Molecular Genetics and Genomics

Systematic elucidation and in vivo validation of sequences enriched in hindbrain transcriptional control Authors (7): Burzynski, Grzegorz M.; Reed, Xylena McCallion, Andrew S. Published in Genome Research	13
A Rare Myelin Protein Zero (MPZ) Variant Alters Enhancer Activity In Vitro and In Vivo Authors (14): Anthony Antonellis; Megan Y. Dennis Eric D. Green Published in Plos One	10
KBP interacts with SCG10, linking Goldberg-Shprintzen syndrome to microtubule dynamics and neuronal differentiation  Authors (13): Alves, Maria M.; Burzynski, Grzegorz Hofstra, Robert M. W.  Published in Human Molecular Genetics	29
Differential Contributions of Rare and Common, Coding and Noncoding Ret Mutations to Multifactorial Hirschsprung Disease Liability Authors (25): Emison, Eileen Sproat; Garcia-Barcelo, Merce Chakravarti, Aravinda Published in The American Journal of Human Genetics	190
Pericentrosomal targeting of Rab6 secretory vesicles by Bicaudal-D-related protein 1 (BICDR-1) regulates neuritogenesis  Published in The EMBO Journal	100
Fine mapping of the 9q31 Hirschsprung's disease locus Authors (16): Tang, C. S.; Sribudiani, Y Garcia-Barcelo, M. M. Published in Human Genetics	19
Characterization of spatial and temporal expression pattern of SCG10 during zebrafish development Published in Gene Expression Patterns	7
Genetic model system studies of the development of the enteric nervous system, gut motility and Hirschsprung's disease Published in Neurogastroenterology and Motility	58
Goldberg-Shprintzen syndrome and microtubules Published in Neurogastroenterology and Motility	0
Microarray-based identification of differentially expressed genes in the intestines of Zebrafish ENS mutant lessen  Published in Neurogastroenterology and Motility	0
Characterization of the Stathmin SCG10 and its interactions with Kinesin Binding Protein (KBP) in zebrafish Published in Neurogastroenterology and Motility	0

Hirschsprung disease, associated syndromes and genetics: a review Authors (24): Amiel, J.; Sproat-Emison, E Fernandez, R. Published in Journal of Medical Genetics	576
Single worldwide origin for a common low-penetrance RET mutation in Hirschsprung disease (HSCR) Authors (17): Lantieri, F.; Amiel, J Chakravarti, A. Published in Genetic Epidemiology	0
Current concepts in RET-related genetics, signaling and therapeutics Published in Trends in Genetics	83
A novel susceptibility locus for Hirschsprung's disease maps to 4q31.3-q32.3 Authors (10): Brooks, AS; Leegwater, PA Bertoli-Avella, AM Published in Journal of Medical Genetics	23
Homozygous nonsense mutations in KIAA1279 are associated with malformations of the central and enteric nervous systems Authors (17): Brooks, AS; Bertoli-Avella, AM Hofstra, RMW Published in The American Journal of Human Genetics	110
Identifying Candidate Hirschsprung Disease—Associated RET Variants Published in The American Journal of Human Genetics	42
RET-Familial Medullary Thyroid Carcinoma Mutants Y791F and S891A Activate a Src/JAK/STAT3 Pathway, Independent of Glial Cell Line—Derived Neurotrophic Factor Published in Cancer Research	70
Localizing a putative mutation as the major contributor to the development of sporadic Hirschsprung disease to the RET genomic sequence between the promoter region and exon 2  Authors (11): Burzynski, GM; Nolte, IM Hofstra, RMW  Published in European Journal of Human Genetics	
Mutation in the RET genomic sequence between the transcription start site and exon 2 as a major contributor to the development of Hirschsprung disease  Authors (10): Hofstra, RMW; Burzynski, G Burys, CHCM  Published in The American Journal of Human Genetics	0