## Erik Schoenmakers

List of Publications by Year in descending order

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#	Article	IF	CITATIONS
1	Mutations in the selenocysteine insertion sequence–binding protein 2 gene lead to a multisystem selenoprotein deficiency disorder in humans. Journal of Clinical Investigation, 2010, 120, 4220-4235.	8.2	268
2	Resistance to thyroid hormone caused by a mutation in thyroid hormone receptor (TR) $\hat{l}\pm 1$ and TR $\hat{l}\pm 2$ : clinical, biochemical, and genetic analyses of three related patients. Lancet Diabetes and Endocrinology,the, 2014, 2, 619-626.	11.4	100
3	Comprehensive Screening of Eight Known Causative Genes in Congenital Hypothyroidism With Gland-in-Situ. Journal of Clinical Endocrinology and Metabolism, 2016, 101, 4521-4531.	3.6	82
4	Mutation in human selenocysteine transfer RNA selectively disrupts selenoprotein synthesis. Journal of Clinical Investigation, 2016, 126, 992-996.	8.2	80
5	Homozygous loss-of-function mutations in SLC26A7 cause goitrous congenital hypothyroidism. JCI Insight, 2018, 3, .	5.0	44
6	Human Disorders Affecting the Selenocysteine Incorporation Pathway Cause Systemic Selenoprotein Deficiency. Antioxidants and Redox Signaling, 2020, 33, 481-497.	5.4	22
7	Nephrogenic syndrome of inappropriate antidiuresis secondary to an activating mutation in the arginine vasopressin receptor AVPR2. Clinical Endocrinology, 2016, 85, 306-312.	2.4	19
8	Human Genetic Disorders Resulting in Systemic Selenoprotein Deficiency. International Journal of Molecular Sciences, 2021, 22, 12927.	4.1	16
9	Liver X receptor inhibition potentiates mitotane-induced adrenotoxicity in ACC. Endocrine-Related Cancer, 2020, 27, 361-373.	3.1	15
10	Brief Report: A Novel Sodium/Iodide Symporter Mutation, S356F, Causing Congenital Hypothyroidism Thyroid, 2021, , .	4.5	2