Sara Cabet

List of Publications by Year
in descending order

9	114	1684188 5	1474206 9
papers	citations	h-index	g-index

9	9	9	396
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	SPEN haploinsufficiency causes a neurodevelopmental disorder overlapping proximal 1p36 deletion syndrome with an episignature of X chromosomes in females. American Journal of Human Genetics, 2021, 108, 502-516.	6.2	48
2	A novel lethal recognizable polymicrogyric syndrome caused by ATP1A2 homozygous truncating variants. Brain, 2019, 142, 3367-3374.	7.6	19
3	Two different prenatal imaging cerebral patterns of tubulinopathy. Ultrasound in Obstetrics and Gynecology, 2021, 57, 493-497.	1.7	14
4	Novel truncating and missense variants extending the spectrum of EMC1-related phenotypes, causing autism spectrum disorder, severe global development delay and visual impairment. European Journal of Medical Genetics, 2020, 63, 103897.	1.3	11
5	Prenatal diagnosis of Aicardi syndrome based on a suggestive imaging pattern: A multicenter caseâ€series. Prenatal Diagnosis, 2022, 42, 484-494.	2.3	8
6	A novel truncating variant p.(Arg297*) in the GRM1 gene causing autosomal-recessive cerebellar ataxia with juvenile-onset. European Journal of Medical Genetics, 2019, 62, 103726.	1.3	7
7	Prenatal cerebral imaging features of a new syndromic entity related to KIAA1109 pathogenic variants mimicking tubulinopathy. Prenatal Diagnosis, 2020, 40, 276-281.	2.3	4
8	Prenatal imaging features related to RAC3 pathogenic variant and differential diagnoses. Prenatal Diagnosis, 2022, 42, 478-481.	2.3	2
9	Prenatal diagnosis of congenital perineal lipoma: tip of urorectal septum malformation sequence?. Ultrasound in Obstetrics and Gynecology, 2022, 60, 139-141.	1.7	1