

Genetic disorders and sleep

Disease	~ Prevalence	Notes
Smith Kingsmore	Ultra rare	Majority have sleep problems
Sanfilippo	1 in 70,000	~90%
Smith Magenis	1 in 20,000	100%
Angelman's	1 in 16,000	100%
Rett	1 in 8500 females	80%, some respiratory
Genetic epilepsy	1 in 2000	Many
Down's	1 in 1000	80%, many respiratory
Autism	1 in 100	80%
DSPWD*	1 in 10	100%

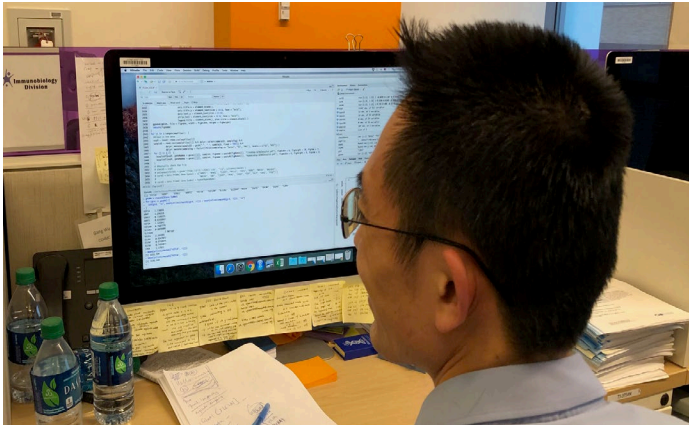
Patient	Age (years)	SGSH testing	Sleep – last visit	Sleep study	Medications
1	29	c.892T>C: c.680C>T	Sleeps 9-10 hours , occasionally wakes up at night	No	
2	8	c.1167C>A c.1272_1282del11	No sleep for 2 days during holidays – Difficulty with sleep	No	Melatonin 10 mg (Trazadone offered)
3	10	c.734G>A c.1080delC	Sleep 7 pm to 6 am, occasional awakenings and leg movements – snoring with concern for OSA -	Ordered	Melatonin 10 mg
4	14	R245H R245H	Some nights will not sleep – oxygen - 1 nap daily – recent concerns for OSA	2012	Melatonin 8 mg (Clonidine not current)
5	8	R245H Y432_R435delinsC	Poor sleep – Insomnia – Frequent arousals improvement 6 pm to 7 am – 2 naps	2015?	Melatonin 20 mg Trazodone 200 mg qhs Zyprexa 10 mg qhs
6	11	c.1080delC c.1080delC	Restless sleep –good response to clonidine, T&A for OSA and snoring	2014	Clonidine 0.1 mg
7	8	R245H c.1080delC	Restless sleep – Frequent arousals	?	Melatonin 5 mg (nightmares at 10mg)
8	13	R245H c.1295del9	Sleeps few hours at night (appears improved in chart) – History of OSA	2013	Risperdal 0.25 mg Melatonin 5 mg (discontinued)
9	17	R245H c.1026insC	Insomnia, restless sleep, frequent leg movements , snoring. Asleep at 8 pm, wakes up at 6 to 8 am, improved RLS	2018	Clonazepam 0.25 mg (Clonazepam falls)
10	22	Not determined	Insomnia		Melatonin 10 mg Zyprexa 2.5 mg Seroquel 200 mg

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Hogenesch Lab



Gang Wu



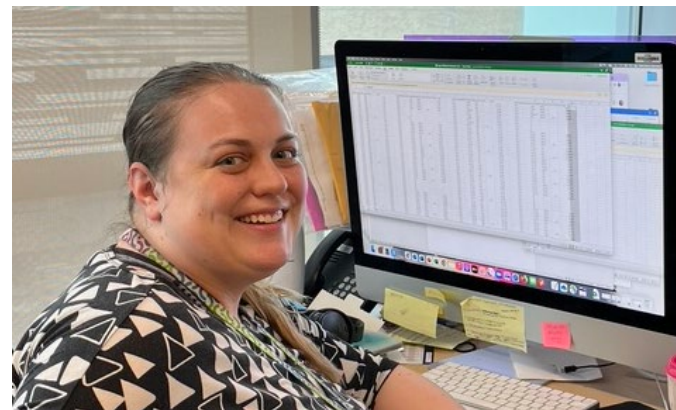
Yin Yeng Lee (YY)



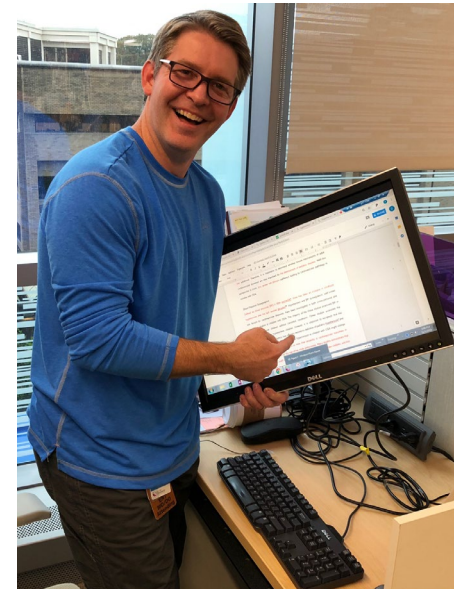
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Sam Whitacre



Dave Smith (collaborator)