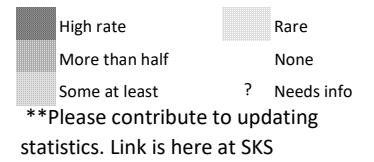


mTOR SUMMARY HEAD TO TOE

Here is a summary of what I've learned so far. In Smith Kingsmore Syndrome (and MINDS), a mutated mTOR gene is overactive although there are rare cases of under activation. First of all, I'd like to say that a gene mutation is not parents' fault at all. In our body, hundreds of gene mutate every day and get fixed everyday. One exception is not anyone's fault. Bad news is that mTOR gene controls a wide variety of important cell functions and if it's out of balance, it causes a different combination of hard-to-fix problems, that includes intellectual disability, autism, global developmental delays mainly, and also epilepsy. Most of patients will need life-time support of other people. Good news is that many of the symptoms are not progressive, some improve over time, and mTOR is one of the few rare disease genes that have been studied in various medical fields. mTOR mutation or over-active mTOR is found in cancer, autism, Alzheimer to name a few. My goal is to continue updating this chart and get help and collaboration from such medical fields, that might also benefit from understanding mTOR.



Medical Terms	In another word,	Symptoms caused	Medical Field besides Neurology/ Treatment Options (* helped Elizabeth)	My Wish List for Research Collaboration	SKS	InAutism in General	In Elizabeth	Her mTOR Variant	TSC	Other mTORpathies
mTOR IS OVERACTIVE (There are rare cases of under activation)		In simple terms, less cleaning of cells, overgrowth of number and size of brain cells. Most symptoms are hard to fix, but not regressive and some improve over time.	Neurology*, Genetics*, Gene Editing, Oncology* Molecular Biology*	Gene Editing, Autism, Alzheimer, Oncology, Other mTORpathies. Clinical trials of current and the next generation mTOR inhibitors, and treatment options of separate fuctions affected by mTOR.	High rate	High rate	High rate	High rate	High rate	High rate
Brain Malformation of Polymicrogyria	Too many wrinkles	Intellectual disability, Epilepsy			Some at least	Needs info	High rate	Some at least	Needs info	Needs info
Hypomyelination	Less white matter (Sheath of neuron)	Intellectual disability, etc.		ISRB. Vitamins, Multiple Sclerosis	Some at least	Needs info	High rate	Some at least	Needs info	Needs info
Agenesis of Corpus Callosum	Center divider of brains	Intellectual disability, etc.			Some at least	Needs info	High rate	Needs info	Needs info	Needs info

mTOR Summary Head to Toe

Medical Terms	In another word,	Symptoms caused	Medical Field besides Neurology/ Treatment Options (* helped Elizabeth)	My Wish List for Research Collaboration	SKS	ASD	Elizabeth	Th1977le	TSC	Other mTORopathy
Hydrocephalus	Water and pressure build up		Neurosurgery*/Neuro-Ophthalmology*/Ophthalmology*	Neruroptalmomy for Detection		?		?	?	?
Intellectual disabilliy	Small Brain stem	Difficulty swallowing, etc.	Feeding Therapy*			?		?	?	?
		Delays in Cognitive, Non-Verbal	ABA Therapy*				?	?		?
			ABA Therapy*, Speech Therapy			40%		?	?	?
Autism Spectrum Disorder	Autism		ABA Therapy*					?		40-50% ?
ADHD							?	?	?	?
Epilepsy	Multiple seizures		mTOR inhibitor medication &, other Anti Epileptic Drugs*, Ketogenic Diet (but my mTOR Diet* was better), Vagal Nerive Stimulator	Epilepsy, TSC (Tuberous Sclerosis Complex)		38%			80%	?
Sleep Disorder				Sleep Study		58%			?	?
	Difficulty Swallowing		Swallow Study by Feeding Therapist and X-ray video*, Feeding Therapy*, Diet adjustment*			?			?	?

mTOR Summary Head to Toe

Medical Terms	In another word,	Symptoms caused	Medical Field besides Neurology/ Treatment Options (* helped Elizabeth)	My Wish List for Research Colaboration	SKS	ASD	Elizabeth	Th1977Ile	TSC	Other mTORopathy
Autophagy Disprder	Not cleaing cells well.	Autism, Intellectual disability, Epilepsy, less receptors of GABA	mTOR Inhibitors,	Alzheimer, Autophagy enhancing medicines and supplements, Detection and Visualization of Autophagy level and resulting accumulation of Tau, beta Amylaze and other misfold proteins and junk.	■	?	?	?	?	?
Irritable Bowel Syndromes	Gastro Intestinal Issues	Diahrrea, Constipation, or both alternating, Not Digesting well.	Neuro Gastroentrogy, Gastor Intestinal Dept/ Digestive enzymes*	Neuro-Gut Relationship, Human study, Comprehensive Analysis on SKS patients	■	4 times more likely	■	■	■	?
				Comprehensive Analysis on SKS patients						