PI Name	Project Title	Funding Sponsor	Anticipated Direct Costs Balance as of 12/7/20 reallocations	Anticipated Total Cost Balance as of 12/7/20 reallocations	Budget Period Direct Costs	Budget Period Total Costs	Period Remaining	COMMENTS	Proj End Date	Katsanis effort FY20 avg	Davis effort FY20 avg	SMCRI Fund #	Agency #	Budget Period Start	Budget Period End
GOAL: MAINTAIN (POTENTIAL FOR RENEWAL)															
Katsanis, Nicholas	Genetic and Functional Studies of Human Ciliary Syndromes	NIH	$75,772.24	$115,957.01	$288,388.00	$441,234.00	3 years	Davis managing all current work; on track to meet all specific aims	6/30/2024	20	11.7	901570	R01 DK072301	9/14/2019	6/30/2021
Katsanis, Nicholas	Molecular Genetics of BBS	NIH	$189,700.22	$322,772.46	$284,037.00	$434,577.00	3 years	Davis managing all current work; on track to meet all specific aims	3/31/2024	11.1	10.5	901572	R01 HD042601	4/1/2020	3/21/2021
Katsanis, Nicholas	Molecular Mechanisms and Genetic Drivers of Reciprocal Genomic Disorders	Mass. Gen. Hospital	$14,271.19	$21,835.73	$139,869.00	$214,000.00	2 years	Davis has been managing project since inception; on track to meet all specific aims	6/30/2023	12.5	11.3	916022	233278 (R01 HD096326)	7/1/2019	6/30/2021
GOAL: MAINTAIN (NO POTENTIAL FOR RENEWAL)															
Katsanis, Nicholas	The Role of Basal Bodies in Wnt Signaling	NIH	$167.94	$257.12				No active experiments; end of project	7/31/2021	15	6.3	901571	R01 GM121317	8/1/2019	7/31/2021
Katsanis, Nicholas	Functional Dissection of GnRH Defects and Networks	Mass. Gen. Hospital	$25,527.51	$39,057.61				ACT-GeM not included in grant renewal submitted in 2020; Davis managing completion of existing projects	3/31/2021	15	7.6	916021	229029 (P50 HD028138)	8/1/2019	3/31/2021
GOAL: DISCONTINUE															
Katsanis, Nicholas	Gene-Environment Interactions for Cortical Development in Schizophrenia	Johns Hopkins Univ	$1,596.98	$2,444.24				No active projects, last paper published November 2020	6/30/2021	15	8.3	916020	2004443000 (P50 MH094268)	7/15/2019	6/30/2021
Katsanis, Nicholas	Proteasomal Genetic Agonists as Therapeutics for Retinal Degeneration	Research to Prevent	$11,622.93	$11,622.93				Personal Award to Katsanis, modest scientific progress	12/30/2022	1.4	0	925774	N/A	12/15/2019	12/30/2020
DAVIS FUNDS															
Davis, Erica	Functional Dissection of CNVs in Neurodevelopmental Traits	NIH	$62,112.81	$95,032.50				On track for renewal	2/28/2021	0	11.1	901580	R01 MH106826	2/15/2020	2/28/2021
Davis, Erica	Genetic and Functional Dissection of Congenital Anomalies of the Brain	Univ. North Carolina	$46,270.96	$70,949.41				On track for renewal	3/31/2021	0	13.3	916049	FEDERAL (R21 TR002770)	9/1/2019	3/31/2021
DISCRETIONARY FUNDS															
Katsanis, Nicholas	SMCRI N. Katsanis Start-Up	CHCRC						Funding 5% of genetic counselor; reagent costs for pilot projects	8/31/2024	0	0	939047	NONE		
Davis, Erica	SMCRI E. Davis Start-Up	CHCRC						Funding 5% of genetic counselor; reagent costs for pilot projects	8/31/2024	0	0	939050	NONE		
Katsanis, Nicholas	Kennedy Endowed Chair		$85,080.00	$85,080.00				Reagent costs for pilot projects	8/31/2021	0	0	498061	NONE	9/1/2020	8/31/2021

Katsanis effort FY20 avg	Davis effort FY20 avg
20	11.7
11.1	10.5
12.5	11.3
15	6.3
15	7.6
15	8.3
1.4	0
0	11.1
0	13.3
	0
	20
	0
	90

Funding Source	Theme	Projects	Brief description	Active projects	Active project/ Manuscript prep	No experiments/ Manuscript prep	No experiments/ Reagents made	Katsanis involvement	Davis involvement
R01 DK072301	Ciliopathy	6	Gene discovery, mutational burden, genetic architecture	3		3		Awarded in 2005; Long term intellectual leadership	Ongoing work since 2005. Grant preparation, publications
R01 GM121317	Ciliopathy	1	Cilia, signaling and disease				1	Awarded in 2007; Long term intellectual leadership	Ongoing work since 2007, Grant preparation, publications. F32 fellowship based on this grant.
R01 HD042601 (some overlap with RPB)	Ciliopathy	3	Molecular mechanisms of Bardet Biedl syndrome	2			1	Original career R01, awarded in 2003; Long term intellectual leadership	Ongoing work since 2005, Grant preparation, publications
Discretionary	Neurodevelopmental disorders	2	Gene discovery, functional models and therapeutic development	1	1			Initiated at Duke ~2010	Ongoing since first faculty position at Duke, 2010
P50 HD028138 (MGH sub) (submit R01 Feb 5-subcontract)	Neurodevelopmental disorders	2	Functional modeling of GnRH disorders		1		1	Awarded in 2017	Grant preparation, All progress reports, Publications
R01 MH106826	Neurodevelopmental disorders	2	Understand genes that contribute to deletion copy number variants	1		1		Conceptual involvement of original grant application, advisory role	Awarded to Golzio in 2015, transfer to Davis in 2016.
R01 HD096326	Neurodevelopmental disorders	3	Understand genes that contribute to reciprocal copy number variants			3		Subcontract awarded in 2018	Grant preparation, All progress reports, Publications
Rett Foundation	Neurodevelopmental disorders	1	Zebrafish model of Rett syndrome				1	Primary intellectual contributor	None
Discretionary	Therapeutic development	5	Use OMICs, zebrafish, and mice to develop therapies for rare pediatric conditions				5	Primary intellectual contributor	Advisory/mentorship role to trainees and staff
Murley Family Fund	Therapeutic development	1	Connectivity Mapping to identify new therapies for rare disease	1				Primary intellectual contributor	Advisory/mentorship role to trainees and staff
Discretionary	Undiagnosed rare disorders	10	Intersecting genomics, transcriptomics, and functional models to discover genes and mechanism	2		3	5	Initiated at Duke ~2010	Ongoing since first faculty position at Duke, 2010
R21 TR002770 (UNC sub) (R01 submitted Oct 2020)	Undiagnosed rare disorders	3	Gene discovery and zebrafish models in prenatal cases	3				Introduced to collaborator, advisory role.	Awarded in 2019, R01 subcontract to be reviewed in winter 2021
P50 MH094268	Psychiatric disease / cilia	0	Genetic dissection and mouse models of ciliary/basal body proteins involved in schizophrenia					Awarded subcontract PI in 2009	None

Funding Source	Theme	Project	Brief description	Status	Sharon	Kat	George	Kamal	Tanner	Marie	Farid	Carmen	Thomas	Abraham	Abby	Shiraz	Intellectual Leadership	Comments
R01 DK072301	Ciliopathy	CEP76	Test the candidacy of CEP76 as a new BBS/Joubert/intellectual disability gene	Test missense mutations in retina sections, perform AcTub staining on mutants and morphants													Erica/Nico	ACTIVE
R01 DK072301	Ciliopathy	IFT Burden	Determine whether mutational burden and discrete modular pairings of mutational burden can predict ciliopathy phenotypes and severity.	Statistical analysis is complete. Manuscript preparation.													Erica/Nico	No active experiments
R01 DK072301	Ciliopathy	IFT172	Establish pathogenicity of a novel IFT172 variant using mRNA splicing assays, zebrafish assays, and ciliated cells	test variants													Erica	ACTIVE
R01 DK072301	Ciliopathy	Joubert Burden	Determine whether mutational burden and discrete modular pairings of mutational burden can distinguish Joubert syndrome from other ciliopathies. (Collaboration: Joe Gleeson and Enza Maria Valente)	Analysis and replication cohorts. Manuscript preparation													Nico	No active experiments
R01 DK072301	Ciliopathy	let7b	Evaluate the contribution of miRNAs to ciliary defects using mammalian cells and zebrafish models	Manuscript in prep													Erica/Nico	No active experiments
R01 DK072301	Ciliopathy	Splice isoforms	Establish how splice isoforms contribute to pleiotropy in genetic disease.	Reagent generation. Eventually recontact Chris Cassa (Harvard) for reanalysis													Erica/Nico	ACTIVE
R01 GM121317	Ciliopathy	Compensatory Pathogenic Deviation	Establish BBS4 mutant cell lines; perform saturated mutagenesis, validate in zebrafish	Cell line generation													Nico	No active experiments
R01 HD042601 and RPB	Ciliopathy	BBS lead compounds	Determine whether four lead compounds identified in a cell-based compound screen can rescue BBS mouse phenotypes	Reproduce zebrafish data generated at Duke, generate exposure data, and treat Bbs4 mice													Erica/Nico	Cell based screen performed by Rescindo therapeutics. Check for IP on compounds
R01 HD042601	Ciliopathy	BBS mice for asprosin	Determine whether asprosin levels are elevated in Bbs mouse models, and if modulating asprosin levels could be a rational therapeutic for the obesity phenotype of BBS. Collaboration with Atul Chopra (Case Western Reserve University)	All samples have been sent to the Chopra lab. Awaiting results and potential for follow up.													Erica	No active experiments
R01 HD042601	Ciliopathy	Ciliopathy Mouse - Renal	Test whether double ciliopathy mutants have exacerbated phenotypes compared to single mutants; determine effects of inter- vs intra module interactions are different	New Bbs4/Ttc21b cohort, generate Bbs10/Bbs4 mouse, scRNAseq kidneys in single v doubles													Erica/Nico	ACTIVE
Discretionary	Neurodevelopmental disorders	Choremio exomes	Determine the genetic architecture of microcephaly/neurodevelopmental syndromes in proband-parent trios from Greece using whole exome sequencing (Collaboration; Jan Traeger Sinodinos, University of Athens)	December review of diagnostic families is complete. Further clinical data is required to analyze families with exome data													Erica/Nico	George: PhD thesis
Discretionary (R01 drafted for 2021 submission)	Neurodevelopmental disorders	NGLY1	Characterize mouse and zebrafish mutants. Develop conditional mice, scRNAseq of brain tissue, test CMAP	Phenotype mouse, test cmap drugs on fish, test MVA in pregnant mice, develop conditional mutants. Manuscript in prep.													Erica/Nico	ACTIVE
P50 HD028138 (MGH sub)	Neurodevelopmental disorders	RhoGap	Determine physiological relevance of ARHGAP5 and ARHGAP35 to idiopathic hypogonadotropic hypogonadism (Collaboration: Stephanie Seminara, MGH)	Manuscript draft from collaborators, phenotype stable mutants (flow sort GFP+ and RNAseq of hom vs WT if phenotype), qRT of GFP+ neurons, ISH of all 3 genes													Erica/Nico	ACTIVE
P50 HD028138 (MGH sub) (submit R01 Feb 5-subcontract)	Neurodevelopmental disorders	GnRH RNAseq	Characterize transcriptomes of GnRH disorder mutants in isolated cell types (Collaboration: Stephanie Seminara, MGH)	Characterize mutant lines													Erica/Nico	No active experiments- maintaining zebrafish lines
R01 MH106826	Neurodevelopmental disorders	SIN3B, BPTF, KCTD13, MSL2	Characterize mutant for RNAseq and Connectivity Mapping	Characterize mutant lines													Erica	ACTIVE
R01 MH106826	Neurodevelopmental disorders	SLF2-SMC5	Establish relevance of the SMC complex genes to a new microcephaly syndrome using zebrafish models. Collaboration with Grant Stewart (Birmingham), and Bertrand Isidor (Nantes University)	Manuscript in prep													Erica	No active experiments- maintaining zebrafish line
R01 MH106826 and R01 HD096326	Neurodevelopmental disorders	16p GnRH	Determine which genes contribute to the reproductive phenotypes in humans bearing the 16p11.2 deletion or duplication (Collaboration with Alex Reymond, Lausanne)	Manuscript in revision with *Nature Communications*													Erica/Nico	No active experiments
R01 MH106826 and R01 HD096326	Neurodevelopmental disorders	2q13	Determine which genes contribute to the neurological and craniofacial phenotypes in humans bearing the 2q13 deletion or duplication	Manuscript in prep													Erica/Nico	No active experiments; George: PhD thesis
R01 MH106826 and R01 HD096326	Neurodevelopmental disorders	Triplosensitive	Evaluate which genes in the human genome are sensitive to triplosensitivity, haploinsufficiency, or both using statistical predictions and zebrafish validation (Collaboration: Mike Talkowski, MGH/Harvard)	Manuscript in review at *Cell*													Erica	No active experiments
Rett Foundation	Neurodevelopmental disorders	MECP2- Rett syndrome	Characterize the transcriptome of glutamatergic and gabaergic neurons in zebrafish mecp2 models	Cross reporters for RNAseq and scRNAseq													Nico	No active experiments- maintaining zebrafish lines
Discretionary	Therapeutic development	Cyclica compound testing	Test whether chemical compounds can rescue the signaling and proteasome defects of BBS4 mutant cells (Collaboration with Cyclica)	Reporter assays- Wnt, proteasome													Nico	No active experiments
Discretionary	Therapeutic development	SLC6A1	Determine whether a zebrafish model will recapitulate human phenotypes and whether a compound library screening will identify new therapeutics	Develop zebrafish model, and test therapeutics													Nico	No active experiments
Discretionary	Therapeutic development	Toxicity testing-1,4 dioxane	Determine toxicity of a new chemical compound to treat eye disease (Collaboration with Vasillis Vasiliou, Yale)	FET testing													Nico	No active experiments
Discretionary (formerly PACS1 foundation)	Therapeutic development	PACS1	Determine whether ASOs are effective in the treatment of PACS1-associated disease symptoms	Design mouse model													Nico	No active experiments
Discretionary (SBIR award to Rescindo in 2021)	Therapeutic development	Kabuki Syndrome mouse phenotyping	Determine clinical endpoints for a KS trial in humans, and refine dosing of dabrafenib in mouse models of KS.	muscle phenotyping, immune profiling													Nico (and Rescindo Therapeutics)	No active experiments

Funding Source	Theme	Project	Brief description	Status	Sharon	Kat	George	Kamal	Tanner	Marie	Farid	Carmen	Thomas	Abraham	Abby	Shiraz	Intellectual Leadership	Comments
Murley Family Fund	Therapeutic development	C-Prime	Develop: (1) zebrafish models to isolate specific cell types; (2) treat zebrafish with and FDA-approved compound library; and (3) generate an RNAseq dataset of each treated condition for Connectivity Mapping	Generate zebrafish tg models, RNAseq samples treated with a subset of neuroactive compounds are ready to send in January 2021													Nico	ACTIVE
Discretionary	Undiagnosed rare disorders	Alveolar Capillary Dysplasia	Determine whether incomplete penetrance in an ACD. Pedigree can be attributed to variants in the FOXF1 enhancer region (Collaboration with Aaron Hamvas (Lurie) and Pawel Stankiewicz (Baylor)	Genotyping of a new region. Reporter assays taking place at Baylor.													Erica	ACTIVE
Discretionary	Undiagnosed rare disorders	ANKRD6	Determine physiological relevance of ANKRD6 to patient phenotypes.	Characterize mutant													Erica	No active experiments- maintaining zebrafish line
Discretionary	Undiagnosed rare disorders	CAPN3	Test pathogenicity of a novel variant in CAPN3 that was identified in 3 Pakistani families with limb girdle muscular dystrophy (Collaboration: Shahid Baig, NIBGE, Faisalabad)	Manuscript in prep													Erica	No active experiments
Discretionary	Undiagnosed rare disorders	Gene x Gene Interaction	Determine whether double gene morphants/mutants cause exacerbated phenotypes or hybrid syndromes	reagent development and phenotyping													Nico	No active experiments
Discretionary	Undiagnosed rare disorders	Lurie proband-parent whole exome sequencing program	Use research-based exome sequencing, re-analysis, and zebrafish models to aid gene discovery for rare congenital disorders identified in the Lurie clinics	40 families referred, 10 families enrolled, one family in progress for zebrafish modeling													Erica/Nico	ACTIVE
Discretionary	Undiagnosed rare disorders	NCAPG2	Test variants and cell characterize cell lines from newly identified cases with NCAPG2 variants	Reagent development and phenotyping													Erica/Nico	No active experiments
Discretionary	Undiagnosed rare disorders	Pakistani family - RMI1	Determine physiological relevance of different RMI isoforms to disease.	Zebrafish testing													Erica	No active experiments
Discretionary	Undiagnosed rare disorders	Pakistani family - TFAM	Establish the relevance of TFAM to ovarian phenotypes and mitochondrial abnormalities (Collaboration: Shahid Baid, NIBGE, Faisalabad)	Manuscript in prep													Erica	No active experiments; Farid: PhD thesis
Discretionary	Undiagnosed rare disorders	RNH1	Determine the gene x environment effects that lead to acute neurological disease in mouse models of the human disease	Phenotype mouse - myelination in camkII and nestin with or w/o immune challenge using LPS													Erica	No active experiments- maintaining mouse lines
Discretionary	Undiagnosed rare disorders	RPL17	Determine the molecular mechanism of Diamond Blackfan anemia caused by RPL17 mutation (Collaboration- 3 sites)	Manuscript in prep													Erica	No active experiments- maintaining zebrafish line
R21 TR002770 (R01 submitted Oct 2020)	Undiagnosed rare disorders	BECN1	Establish the physiological relevance of BECN1 loss of function to severe neurological disease with links to the autophagy pathway.	Test variants and isoforms in fish													Erica	ACTIVE
R21 TR002770 (UNC sub) (R01 submitted Oct 2020)	Undiagnosed rare disorders	CACHD1	Test the candidacy of CACHD1 as a cause of a rare prenatal urogenital and craniofacial syndrome. Collaboration with Stephane Bezieu (Nantes University)	Phenotype mutants and complete the transient work.													Erica	ACTIVE
R21 TR002770 (UNC sub) (R01 submitted Oct 2020)	Undiagnosed rare disorders	UNC fetal exomes	Establish zebrafish models for novel disease gene discovery in fetus-parent trios with ultrasound defects.	Reagent development and phenotyping													Erica	ACTIVE
					1	4	6	5	5	3	6	3	5	5	4		Nico: 10 Erica: 15 Erica/Nico: 14	