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### **Oncology**

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Our team is focused on using integrative approaches to genomic data to identify genetic biomarkers for tumor progression and response to therapy. While many genomic analyses in cancer have focused on a single type of genomic data, we are interested in integrating somatic mutations and gene expression profiles with germline genetic information to build rigorous predictive models. For example, variants (germline or somatic) associated with the immune state of the tumor microenvironment may inform our understanding of response to immune-oncology therapies. We work in a highly multidisciplinary environment and blend rigorous statistical approaches and bioinformatics with detailed biological understanding.

Our studies are powered by the wealth of genomic and phenotypic data available in public initiatives such as The Cancer Genome Atlas (TCGA) as well as data from internal cancer cell line screens. Given that these datasets have millions of variants and tens of thousands of genes assessed in much smaller numbers of clinical samples, we are interested in developing and applying statistical approaches to high-dimensional data, including methods for feature space reduction (e.g., focusing on frequently mutated genes, immune-relevant pathways, or genes with validated phenotypic associations) to identify the most impactful variants and genes.

## Selected Publications

Association of MTOR mutations with developmental brain disorders, including megalencephaly, focal cortical dysplasia, and pigmentary mosaicism. [2]

Mirzaa GM\*, Campbell CD\*, Solovieff N, Gould CP, Jansen LA, Menon S, Timms AE, Conti V, Biag JD, Olds C, Boyle EA, Collins S, Ishak G, Poliachik SL, Girisha KM, Yeung KS, Chung BH, Rahikkala E, Gunter SA, McDaniel SS, Macmurdo CF, Bernstein JA, Martin B, Leary RJ, Mahan S, Liu S, Weaver M, Dorschner MO, Jhangiani S, Muzny DM, Boerwinkle E, Gibbs RA, Lupski JR, Shendure J, Saneto RP, Novotny EJ, Wilson CJ, Sellers WR, Morrissey MP, Hevner RF, Ojemann JG, Guerrini R, Murphy LO, Winckler W, Dobyns WB. \*Contributed equally.

*JAMA Neurol.* 2016 Jul 1;73(7):836-45

Estimating the human mutation rate using autozygosity in a founder population. [3]

Campbell CD, Chong JX, Malig M, Ko A, Dumont BL, Han L, Vives L, O'Roak BJ, Sudmant PH, Shendure J, Abney M, Ober C, Eichler EE.

*Nat Genet.* 2012 Nov;44(11):1277-81.

Profiling critical cancer gene mutations in clinical tumor samples. [4]

MacConaill LE, Campbell CD, Kehoe SM, Bass AJ, Hatton C, Niu L, Davis M, Yao K, Hanna M, Mondal C, Luongo L, Emery CM, Baker AC, Philips J, Goff DJ, Fiorentino M, Rubin MA, Polyak K, Chan J, Wang Y, Fletcher JA, Santagata S, Corso G, Roviello F, Shivdasani R, Kieran MW, Ligon KL, Stiles CD, Hahn WC, Meyerson ML, Garraway LA.

*PLoS One.* 2009 Nov 18;4(11):e7887.

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[2] <https://www.ncbi.nlm.nih.gov/pubmed/27159400>

[3] <https://www.ncbi.nlm.nih.gov/pubmed/23001126>

[4] <https://www.ncbi.nlm.nih.gov/pubmed/19924296>

[5] <https://www.ncbi.nlm.nih.gov/pubmed/?term=campbell+catarina>