

'Omics Medicine

Medicine uses many tools to measure the human condition, in wellness and illness. These tools include medical, family, and social history; physical examination, imaging, and laboratory evaluation.

?Omics medicine is a term we use for the next generation of laboratory tools, which open new windows into the molecular makeup of an individual. They are powerful contributors to our knowledge network and our ability to prevent and treat disease across the lifespan:

- Testing can diagnose individuals that are carriers for inherited diseases and guide counseling and intervention before pregnancy.
- Prenatal genomics can detect genetic disease in the womb and pinpoint genome editing targets for future therapies.
- Genomics and metabolomics are used to diagnose newborns with serious medical conditions and can change the course of the disease in many babies and children.
- In healthy individuals, genomics can assess the risk for cancer and other diseases and reveal how individuals will interact with certain medications.
- Genomics in tumor testing are providing insights into the molecular underpinnings of cancer and find new targets for therapies.

More Than Just Genomics

There are many 'omics that science is looking into:

- The epigenome is the expression of your genome via **transcriptomics**.
- **Proteomics** studies the products of RNA.
- **Metabolomics** is the study of metabolites.
- DNA-based tools can also detect bacteria, parasites and viruses that coexist in our bodies through **metagenomics**.
- **Inflamomics, lipidomics, glycomics**, and other large molecular data sets can be used to diagnose and predict disease.

?Omics at UCSF

UCSF's Genomic Medicine Initiative [2] bridges genomic technologies and medical practice to provide more precise patient diagnostics and care. GMI researchers apply genomics to better understand the clinical uses of prenatal, neonatal and pediatric genomes. They also created a tumor diagnostic tool, UCSF500, to classify tumors and guide therapeutic intervention.

The clinical services at the UCSF Health Center for Clinical Genetics and Genomics [3] are using ?omics tools to care for patients. For instance, patients with rare, undiagnosed conditions often struggle with symptoms for which the cause is unknown, despite a battery of standard tests. With no clear answers, patients may experience a ?diagnostic Odyssey?, taking one test or procedure after another in an attempt to understand and find treatment for

their symptoms. With 'omic technologies and careful phenotyping, researchers and clinicians look to decipher an individual's disease and provide an effective treatment plan.

Big Data and Collaborations Across UCSF

To predict who gets disease and how we preempt illness, 'omics need to be integrated with patient histories, environmental exposures, lifestyles, and other data. This requires a 'big data' analytic approach and increased collaborations across UCSF and beyond. Our measures of success will include quicker and more precise diagnosis and reduced hospital and clinic time per patient.

Current Projects

- **Clinical Cancer Genomics Laboratory (CCGL)** [4]
As part of the GMI, researchers have developed a working process to test patient samples using the 'UCSF500,' a panel of more than 500 gene mutations implicated in cancer to provide more precise diagnosis and treatment plans for patients.
- **Screening Pediatric Diseases** [5]:
Sequencing the exome (the coding regions of the genome) in infants to identify genetic conditions and undiagnosed diseases, and to assess whether this approach is a beneficial and feasible replacement for standard newborn testing.
- **Diagnosing infectious disease** [6]:
Validating the use of next-generation sequencing for patients with encephalitis, meningitis, sepsis and pneumonia.
- **Epilepsy Phenome/Genome Project:** [7]
UCSF has been a leader in an international effort to make sense of the multiple genetic mutations and clinical symptoms behind epilepsy. The project, which involves 4,000 epilepsy patients and family members from 40 medical institutions, has uncovered new genes never before associated with this debilitating disease. Among them was KCNT1, a gene also involved in heart disease, which has led to the successful use of a common, FDA-approved cardiovascular drug to treat children with a severe form of epilepsy that has resisted current treatments. Through these types of connections, 'omics can not only expand our understanding of the pathways of disease, but also bring us closer to precise and effective therapies.

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Links

- [1] <https://precisionmedicine.ucsf.edu/content/building-knowledge-network>
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