Medical genetics team pinpoints causes of inherited diseases

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(Medical Xpress) -- A child's diagnosis with a congenital deformity or developmental delay raises challenging questions: Could the problem be inherited? What's the prognosis? If other children are born to the same parents, what is the recurrence risk?

The medical genetics team at Lucile Packard Children's Hospital helps answer these questions. The group combines a human touch with diagnostic, clinical and research expertise to help families and referring physicians navigate genetic disease. They handle an array of conditions, including craniofacial anomalies, such as cleft palate; autism-like symptoms; chromosomal defects, such as Down syndrome and Fragile X syndrome; and rare biochemical defects, such as phenylketonuria and mitochondrial disorders.

"When it comes to our capacity for handling the complex cases, I cannot think of a better place for patients," said Gregory Enns, MD, director of the Biochemical Genetics Program and associate professor medical genetics in pediatrics. His colleagues offer diagnosis, genetic counseling, family planning, prenatal diagnostics, neonatal newborn screening and management of chronic genetic and metabolic disease, he noted.

The team receives referrals from practitioners across the United States and around the world. For each referral, a genetic counselor contacts the family by telephone to learn what diagnostic testing the child has received and obtain medical records. Clinic visits focus on the next steps in diagnosis and treatment, often including testing from the hospital's

biochemical genetics lab, which is unique in Northern California.

“Partly because of the support of our lab, we pursue rare diagnoses much further than many institutions,” said Jonathan Bernstein, MD, PhD, clinical associate professor of pediatric genetics. “Reaching a diagnosis gives families the peace of mind of knowing what’s going on at the level of cause as opposed to symptoms.”

Regardless of whether a diagnosis is established, the medical geneticists work closely with other providers at Packard Children’s to ensure patients receive all the treatment they need. In some cases, referrals are directed within the team itself, such as to its Down syndrome clinic. Many patients are seen by other Packard departments. When relevant, patients go to the cancer genetics group at Stanford Hospital & Clinics. And the team works to help families obtain support from others facing similar diagnoses.

“We’re like quarterbacks, arranging coordination of services to meet our patients’ needs,” Enns said.

For rare metabolic diseases, Packard Children’s offers a standard of care unmatched elsewhere in the world. In 2010, a multidisciplinary team at Packard used a first-of-its-kind combination of drugs to ward off brain damage in a newborn who had a rare urea cycle disorder until he received a curative liver transplant. “It was gene therapy with a scalpel,” Enns said. In many other cases, patients with rare disorders can benefit from research conducted at Stanford or in collaboration with other institutions.

Referrals to medical genetics should go through Packard Children’s referrals center, Enns said.

“We are always happy to hear from referring physicians before or after a

patient's visit," Bernstein added. "If they want to discuss whether a referral is appropriate for a specific patient, we're glad to work on that, too."

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