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“Demonstrating the clinical utility of whole exome sequencing for cancer patients”

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Abstract:
Breast cancer in young women often has a genetic cause. The BRCA genes explain only 20 to 25 percent of hereditary breast cancers, but there are still many families where a genetic cause is suspected, but the root is never found through routine genetic testing. Researchers from UC San Diego Moores Cancer Center and The Salk Institute for Biological Studies propose to sequence the entire exome (the coding part of the genome) of a family with a strong suspicion of a hereditary breast cancer gene. Two sisters, whose genetic testing was normal, both had breast cancer in their 30’s. Researchers hope to uncover a new gene that increases the risk of early-onset breast cancer in this family. Once a potential high-risk gene (or genes) has been identified, researchers will examine how that gene works within cancer cells in an effort to understand how this disease can develop in women at a young age. The resulting genetic information may be of immediate benefit to this family and to other families with unexplained early breast cancer. Families with this gene will be able to identify who has inherited the gene and should be monitored carefully or undertake preventive measures.