

Direct to Consumer Genetic Testing

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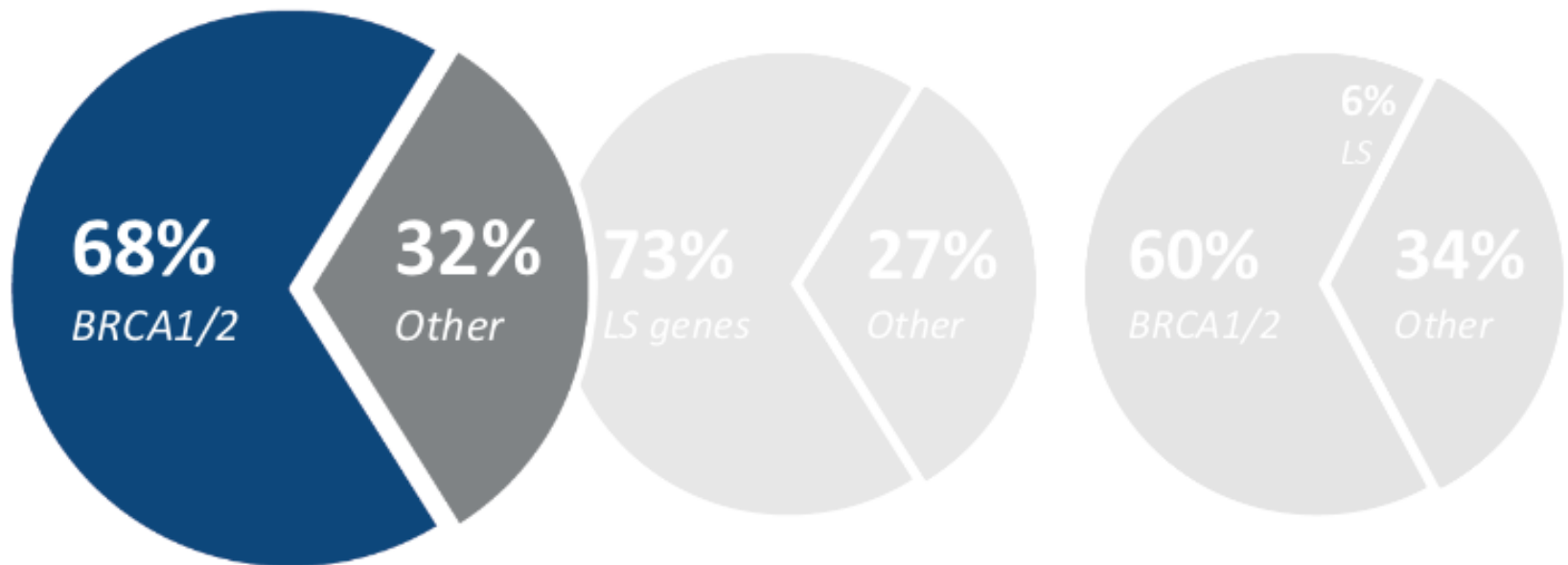
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Genetics of Cancer

- ~5-10% of cases have a strong hereditary component
- ~15-20% are “familial”/multifactorial
- ~70-75% are thought to be sporadic
- Not always BRCA 1 and 2, multiple other genes

Approximately One-Third of Mutations Would be Missed with Traditional Testing Approaches

In 1,781 patients with Breast Cancer



Prevalence of Gene Mutations Among Hereditary Breast and Ovarian Cancer Patients Using a 25-gene Panel, Nadine Tung et. al. Presented at ACMG in March 2014

Multi-gene panel testing in patients suspected to have Lynch syndrome, Matthew B. Yurgelun et. al. Presented at ASCO June 2014

A Study of Ovarian Cancer Patients Tested With a 25-gene Panel of Hereditary Cancer Genes, Lucy R. Langer et. al. Presented at

Direct to Consumer Testing?

- Color Genomics
- At home saliva sample, no physician needed, no counseling
- Costs \$249
- Test 19 genes in a panel including BRCA 1 and 2



Genetic Counseling



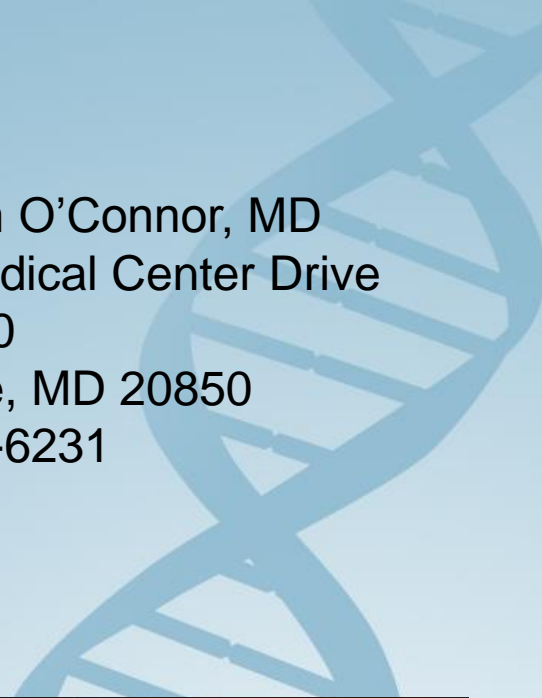
- Full pedigree analysis and risk assessment
- Assessment of
 - Personal risks of cancer based on family history
 - Appropriate genetic testing (panel or specific gene)
 - Full informed consent prior to genetic testing
 - Personalized, risk-based screening and prevention options (risk models)
 - Laws (GINA) about discrimination, implications
 - Who else in the family should be tested?

Unanswered Questions without Counseling

- Even if genetic testing is negative, still may be at high risk and might need different screening recommendations
- Annual MRI? Chemoprevention?
Colonoscopies
- True negative? Gene panel?
- Insurance denials-then maybe Color Genomics



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