Ambry Genetics®

RESEARCH SUMMARY

Clearly Identify Hereditary Breast Cancer

SEPTEMBER 2017

Emphasizes the importance of using quality, comprehensive genetic tests, like BreastNext, when assessing patients for hereditary breast cancer risk.

Mulitgene panel tests (MGPT), like BreastNext, help find more patients with hereditary breast cancer.[1]

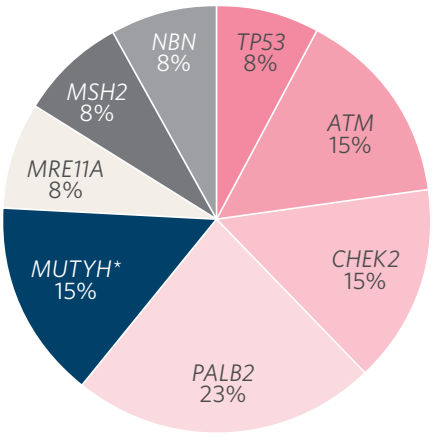
KEY STUDY FINDINGS

- Approximately half (52%) of all identified mutations were found in genes other than *BRCA1/2*

POINTS FOR YOUR PRACTICE

- Providers who utilize MGPT can increase the detection of clinically significant mutations in families that appear to have hereditary breast and ovarian cancer
- Many of the genes identified beyond *BRCA1/2* have associated NCCN® management guidelines for screening and/or prevention, allowing you to make personalized recommendations for your patients[2]
- Using MGPT, like BreastNext, can give you more precise data to accurately manage your patients' health

Non-*BRCA1/2* Mutations



*Monoallelic carriers

Latest findings: *NF1* should be included on hereditary breast cancer panels[3]

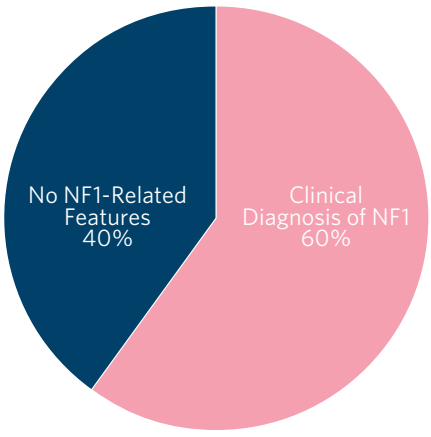
KEY STUDY FINDINGS

- 40% of *NF1* mutation carriers identified on MGPT did not have any Neurofibromatosis Type 1 (NF1)-related clinical features

POINTS FOR YOUR PRACTICE

- NCCN® guidelines indicate that women with a mutation in *NF1* should undergo annual mammograms starting at 30y and consider breast MRI from 30y-50y due to increased breast cancer risk.
- It is important to use a MGPT, like BreastNext or CancerNext, that includes *NF1* to avoid missing this important genetic diagnosis

Mutation Carriers Clinical Histories of NF1



The importance of looking beyond *BRCA1* for patients with triple-negative breast cancer (TNBC)[4]

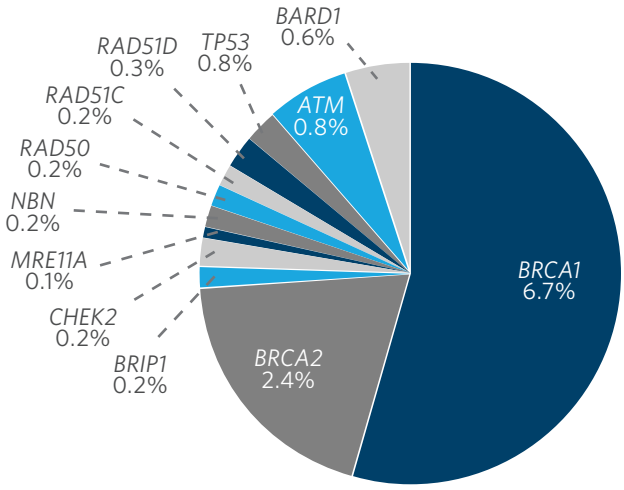
KEY STUDY FINDINGS

- In a large cohort of >6,000 unselected patients with TNBC, 14% carried a mutation in a cancer predisposition gene
- In addition to *BRCA1*, *BARD1*, *BRCA2*, *PALB2*, *RAD51D*, and *RAD51C* were all associated with moderate to high risks of TNBC

POINTS FOR YOUR PRACTICE

- Providers should consider panel testing, such as BreastNext, for their patients with TNBC to avoid missing clinically significant mutations
- *BRCA2*, *PALB2*, *RAD51C*, and *RAD51D* all have NCCN® guidelines[2] for medical management including recommendations for increased breast cancer screening and/or the need to consider preventive surgeries to reduce cancer risk

Mutation Distribution for Patients with TNBC



REFERENCES

1. Kapoor N, *et al.* "Multi-Gene Panel Testing Detects Equal Rates of Pathogenic BRCA1/2 Mutations and has a Higher Diagnostic Yield Compared to Limited BRCA1/2 Analysis Alone in Patients at Risk for Hereditary Breast Cancer". Annals of Surgical Oncology 2015
2. NCCN Clinical Practice Guidelines in Oncology®. Genetic/Familial High-Risk Assessment: Breast and Ovarian. V1.2017
3. Summerour P, *et al. NF1 Mutations Detected on Multi-gene Cancer Panel Testing in Probands with Atypical Phenotypes.* NSGC, New Orleans, LA, September 17-20, 2014.
4. Couch F, *et al. Risks of triple negative breast cancer associated with cancer predisposition gene mutations.* ASCO 2016, Chicago, IL, June 3-7, 2016