Early Comprehensive Genomic Profiling of 82 High Risk Breast Cancers Results in Early Treatment Changes

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Methods:
• 82 patients with high risk breast tumors underwent genomic profiling early in treatment with:
  • XT Comprehensive Genomic assay of 595 genes with a tumor normal match:
    • somatic + germline alterations
    • Whole Transcriptome mRNA sequencing capturing all novel fusions, copy gains/losses, etc.

Results:
• 3 BRCA Somatic Mutations were identified
  • Qualifying these patients for PARP Inhibitors
• 2 Germline mutations were discovered
  • MSH6 (Lynch syndrome) was found; patient did not originally meet germline NCCN family history criteria for genetic testing
  • Shows the tremendous value in a ‘Tumor Normal Match’ assay
• 54% (n=45) of luminal B pts were discovered to have ESR1 alterations in mRNA
  • Tracking response to Tamoxifen currently
• 40% (n=33) of patients carried a PIK3CA somatic mutation; 7 also carrying TP53
  • These tend to be very aggressive cancers
• 12 pts showed HER2 RNA over expression
  • 4 of which showed discordant to FISH / IHC results
  • Do we give these women anti hormonal therapy??
• 33/82 patients had PRG (progesterone) abnormality
• Multiple clinical trial matches were available and accessible to patients with early stage, as well as recurrent or metastatic disease

Background
• Comprehensive Genomic Profiling (DNA, RNA, Germline, etc.) can help identify targets for systemic therapy, as well as advanced her-2 analysis; and facilitate earlier entry into clinical trials
• This information can change patient outcomes earlier in the treatment plan

Conclusion:
• Comprehensive Genomic Testing can impact and personalize management of High Risk Cancers
• Luminal B patients with a very low CR to pre op chemo may see the highest benefit
• Early entry into clinical trials on genetic profiling has the potential to increase CR for all high risk tumors