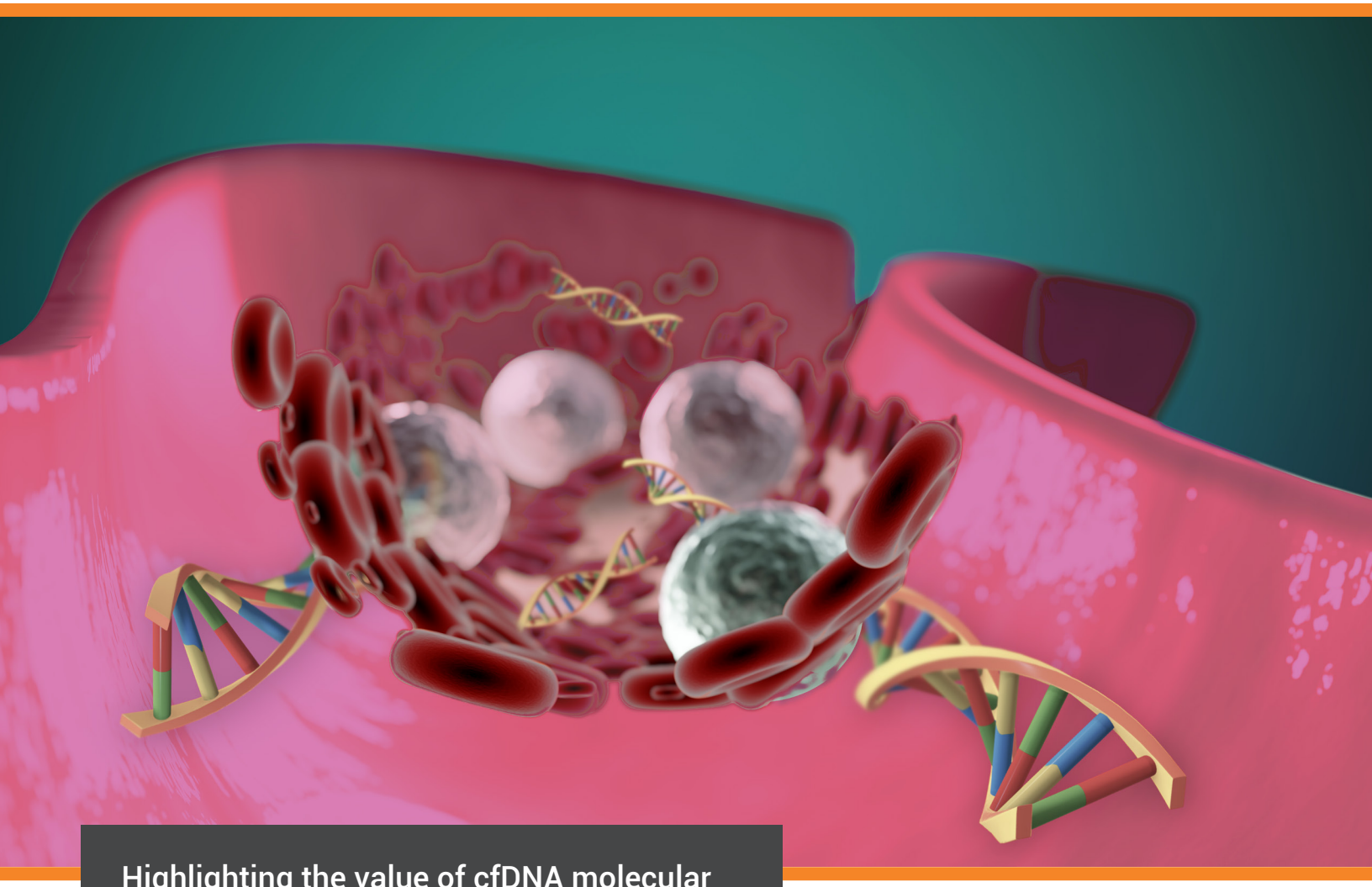
Case Study
Liquid Biopsy cfDNA

Utility of this technology in clinical oncology



Highlighting the value of cfDNA molecular profiling along with the incorporation of the patient's history for a complete landscape of the molecular features and interpretation of this information by experts.

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Background

Cell-free DNA (cfDNA) has become a reframing technology in clinical oncology. The ability to evaluate the presence, level, and constitution of tumor DNA from a routine, noninvasive blood draw has opened the door to a broad array of multiple clinical applications. Here we highlight the value of cfDNA molecular profiling along with the incorporation of the patient's history for a complete landscape of the molecular features and interpretation of this information by experts

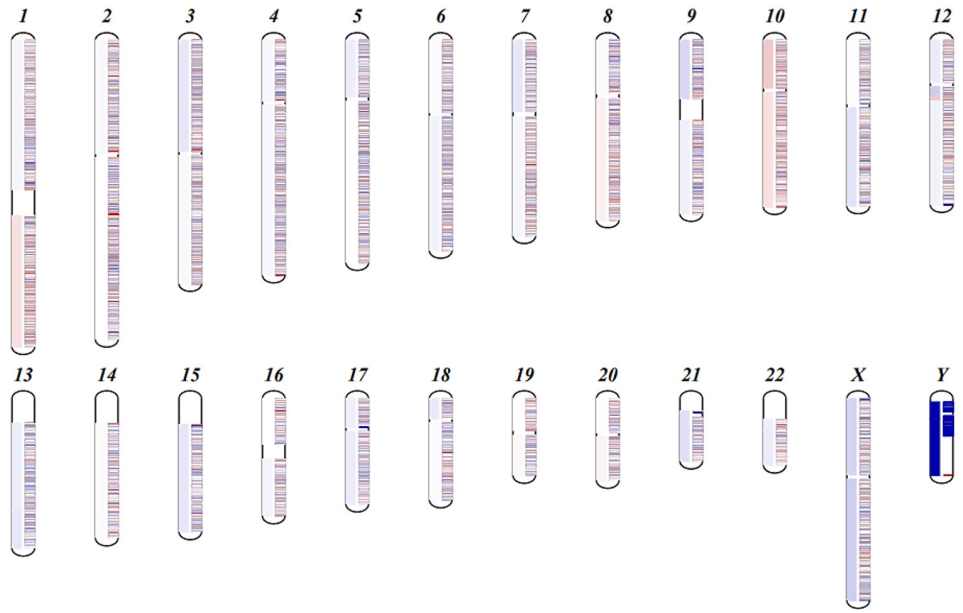

Figure 1
Chromosomal structural analysis showed1q+ and +10

Discussion

The presence of CHEK2, DNMT3A (2 mutations), JAK2 (V617F), and TET2 is consistent with chronic myeloproliferative neoplasm (MPN). The detection of the other mutations along with the chromosomal changes highlights the presence of circulating solid tumor. Taken together, the findings in this case support the co-presence of chronic myeloproliferative neoplasm along with circulating tumor DNA with high tumor burden. While MPNs typically present with elevated white and red blood cell counts in this case the patient presented with anemia and neutropenia. The patient's manifestations of MPN were masked by the fact that the patient was on chemotherapy and the routine CBC monitoring wouldn't be able to detect it in a timely manner. By monitoring the patient's cfDNA we were able to diagnose the patient and detect her tumor burden promptly and offer her the optimal supportive and prophylactic therapy.

cfDNA testing is also used in the clinic for mutational profiling. Integrating real-time cfDNA monitoring into clinical care provides invaluable opportunities to precisely track the course of a patient's response to therapy, and it could allow not just the early detection of imminent cancer progression but also the identification of the molecular mechanisms driving resistance or disease progression.

Therapeutic Recommendations*

- PIK3CA mutation suggests response to PI3K/mTOR inhibitors.
- PTEN, ARID1A and CHEK2 mutations suggest response to PARP inhibitors.
- AKT1 mutation suggests possible response to AKT inhibitors
- FGFR1 mutation suggests response to FGFR1 inhibitor

*See full list of therapeutics in the drug information section of the report

Clinical History

- 78-year-old female
- With anemia, neutropenia, and history of carcinosarcoma of the uterus

Molecular Profiling Findings

- Germline mutation in **CHEK2** gene, heterozygous
- Mutations in KDM5C, SETD2, DNMT3A (2 mutations), AKT1, **JAK2** (V617F), ARID1A, TET2, PTEN (2 mutations), PIK3R1, KMT2D (2 mutations), **FGFR1**, BCR, **PIK3CA**, TSC2, KMT2C, and DDR2 genes

References

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Genomic Testing Cooperative, LCA
175 Technology Dr, #100, Irvine, CA 92618
Tel: 1-949-540-9421 | Fax: 1-949-301-9719
Website: genomictestingcooperative.com
e-mail: gtc@genomictestingcooperative.com