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Analysis of a Series of Cases of Myeloid Disorders Using a Panel Of 62 Genes by Next Generation Sequence. Diagnostic Performance

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ABSTRACT

Introduction: Various types of myeloid disorders require genetic panels for identification of variants to complement Prognosis, Diagnosis or Treatment along with cytogenomic and molecular studies. We present results of 32 adults (62.5%) and pediatric (37.5%) patients studied at our institution between 2020 - 2022.

Methods: With prior informed consent, bone marrow DNA was obtained for our analysis. NGS was performed on a MiSeq in libraries with a 62-gene panel covering Acute Myeloid Leukemia (AML), Myelodysplastic Syndrome (MDS), Juvenile Myelomonocytic Leukemia (JMML), Chronic Myelomonocytic Leukemia (CMML), and chronic myeloproliferative disorders (MPC). This panel does not efficiently cover FLT3-ITD alterations that should be studied by other methods.

Results: A total of 32 patients were analyzed, of which 5 had a diagnosis of MPC, 10 AML, 9 MDS/AML, 1MMLC, 6 JMML and 1 AML associated with Trisomy 21.

The overall diagnostic efficiency was 84.4%. for MPC and LMA, the diagnostic efficiency was 80% (4/5) and (8/10) respectively; For SMD/LMA the efficiency was 77.8% (7/9), and 100% for LMMC (1/1). 6 patients with JMML and PTPN11 variants. MPC variants in JAK2 (Val617Phe), KRAS (Gly13Asp), CALR (c.1097+21097+53del) and CBL (Arg420Gly). In AML, MDS/AML, 3 patients with variants in NPM1, 2 NRAS, 2 IDH2, 2 TET2, 2 DNMT3A, 2 RUNX1, 1PTPN11 and FLT3, 1 TP53 were identified.

Conclusions: NGS studies are mandatory for risk classification, diagnosis and therapeutic options. We present results of 32 patients analyzed with a diagnostic efficiency of 84.4% for myeloid disorders using NGS sequence.

REFERENCES

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