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CYTOGENETIC CHANGES IN MYELODYSPLASTIC SYNDROMES

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Introduction: Myelodysplastic syndromes are bone marrow disorders mainly occurring in old age. Their diagnostics mainly relies on cytogenetics. This study investigated the extent of cytogenetic changes in patients diagnosed with myelodysplastic syndromes. The most common abnormalities were identified and explained in relation to their cytomorphologic characteristics and to further the management and diagnostics of myelodysplastic syndromes. Aims: To investigate cytogenetic findings of patients with myelodysplastic syndromes in retrospective study and analyze datasets for frequency distribution. Moreover, to evaluate existing methods in order to properly combine them for myelodysplastic syndromes diagnostics and follow-up. Results: Out of 789 patients with myelodysplastic syndromes, abnormalities were found in 25.2% of patients using karyograms and in 23.4% of patients using fluorescent in situ hybridization. The 5q deletion was the most common abnormality and typically seen in combination with <3 abnormalities. Nearly a quarter of positive patients had trisomy 8, followed by monosomy 7, loss of Y chromosome, 7q deletion, and 20 q deletion. Numerical changes were more common than structural or combined abnormalities. Conclusion: In this retrospective study, we have identified the most common abnormalities associated with myelodysplastic syndromes, including 5q deletion, monosomy 7, and trisomy 8, which are consistent with the results of previous studies. While karyotyping and fluorescent in situ hybridization provided similar ratios of positive patients, karyotyping represents a general screening method for myelodysplastic syndromes, and should be done at start and every visit, followed by targeted fluorescence in situ hybridization analysis. Since the great ratio of newly diagnosed patients is negative on both methods, array comparative genomic hybridization is recommended for testing of chromosome imbalances. Thus hidden abnormalities could be revealed using just DNA sample, with no need for cell culture. Cytogenetic methods are very important in myelodysplastic syndromes management because current prognostic systems rely on chromosome abnormalities to define prognostic subytpes.

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PODUDARNOST CITOMORFOLOŠKE DIJAGNOZE I PROTOČNE CITOMETRIJE U MIJELODISPLASTIČNOM SINDROMU

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Uvod Dijagnoza mijelodisplastičnog sindroma (MDS) naslanja se na parametre krvne slike, citomorfološku analizu periferne krvi i koštane srži , a u slučaju odsutnosti značajnije displazije, analiza protočnom citometrijom pomaže u razlikovanju klonskih citopenija, identificiranja imunofenotipa u slučaju displazije barem jedne stanične linije, te otkrivanja aberantnog fenotipa nezrelih stanica mijeloidne loze. Cilj Analiza podudarnosti citomorfoloških karakteristika i protočne citometrije kod bolesnika sa novodijagnosticiranim MDS u KB Merkur u razdoblju od ožujka 2016. do travnja 2023. Metode Analizirali smo 69 bolesnika (38 muškaraca, 31 žena) kojima je postavljena dijagnoza MDS na osnovi citološke punkcije koštane srži. Rezultati Kod 57 bolesnika (82%) morfološka dijagnoza MDS potvrđena je i u nalazu protočne citometrije dok kod 12 bolesnika nalaz protočne citometrije nije ukazivao na MDS. Kod 11 (91%) od ukupno 12 bolesnika kod kojih analiza protočnom citometrijom nije dokazala MDS, nije uočeno ni blasta u citološkom nalazu. Potom smo analizirali izražaj CD34, CD117, CD13 i CD33 (cut off 20%) kod 57 bolesnika kod kojih je u mijelogramu i protočnoj citometriji postavljena sumnja na MDS. CD117 i CD33 se nisu razlikovali među skupinama, neovisno o podtipu MDS. Izražaj CD34 i CD13 statistički su značajno više izraženi kod MDS višeg rizika u odnosu na MDS nižeg rizika. Nema statistički značajne razlike u razini hemoglobina i izražaju CD antigena, dok razina trombocita negativno korelira s razinom CD117, CD13 i CD34. Izražaj CD34 pozitivno korelira s razinom CRP. Zaključak Imunofenotipizacija protočnom citometrijom u dijagnostici MDS-a može pružiti dodatne važne informacije koje nisu dobivene morfološkim nalazom, citogenetikom ili testovima molekularne dijagnostike te bi trebala biti integrirana u dijagnostičko izvješće, ali i praćenje pacijenata. Izražaj pojedinih CD antigena kod dijagnoze pokazao je ne samo dijagnostičku, nego i prognostičku vrijednost. Naši rezultati su u skladu s podacima iz literature sa oko 80% podudarnosti između nalaza protočne citometrije i citomorfološke dijagnostike, a u MDS visokog rizika podudarnost je i oko 90%.