

Clinical Genetics

AB040. Preliminary study of chimerism detection in allogeneic hematopoietic stem cell transplantation using massively parallel sequencing

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Background: Allogeneic hematopoietic stem cell transplantation (HSCT) is a common procedure for several hematologic disorders. The DNA chimerism detection of patient and donor of post-HSCT specimens is a crucial step for physician's precision. Detection of patient and donor DNA chimerism has been relied on short tandem repeats (STRs) analysis using capillary electrophoresis (CE) platform as a gold standard. Recently, a massively parallel sequencing technology (MPS) has shown to improve sequencing performance with high throughput outcomes and with a large set of DNA markers. The objective of this study was to demonstrate the efficiency and performance of MPS towards chimerism analysis.

Methods: A paired-specimens from donors and recipients that underwent HSCT were collected at 18 days, 2 and

3 months post HSCT. These samples were tested on both CE and MPS platforms. Forenseq™ DNA Signature Prep kit (Illumina) was prepared following the manufacture's protocol for Miseq FGx (Illumina®) instrument.

Results: For the sample of the 18 days post-HSCT, MPS demonstrated a mixed recipient chimerism while CE resulted in complete chimerism. Three STR loci (D3S1358, TH01 and D19S433) showed mixed genotypes on MPS while CE revealed only a donor genotype (complete chimerism). Moreover, since Forenseq™ DNA signature Prep kit comprised of 28 autosomal STRs and 94 identity SNPs, two additional STR loci (D6S1043, D4S2408) and 4 additional SNPs indicated a mixed recipient chimerism in 18 days post-HSCT sample. For the specimen at 2 and 3 months post-HSCT, MPS showed a mixed chimerism which were concordant with results from CE platform

Conclusions: The MPS was proofed to be a diagnostic tool for early detection of recipient chimerism in HSCT patients.

Keywords: Chimerism analysis; massively parallel sequencing (MPS); hematopoietic stem cell transplantation (HSCT); short tandem repeats (STRs)

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