Targeted molecular characterization shows differences between primary and secondary myelofibrosis

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Résumé en anglais	INTRODUCTION: In BCR-ABL1-negative myeloproliferative neoplasms, myelofibrosis (MF) is either primary (PMF) or secondary (SMF) to polycythemia vera or essential thrombocythemia. MF is characterized by an increased risk of transformation to acute myeloid leukemia (AML) and a shortened life expectancy. METHODS: Because natural histories of PMF and SMF are different, we studied by targeted next generation sequencing the differences in the molecular landscape of 86 PMF and 59 SMF and compared their prognosis impact. RESULTS: PMF had more ASXL1 (47.7%) and SRSF2 (14%) gene mutations than SMF (respectively 27.1% and 3.4%, P = .04). Poorer survival was associated with RNA splicing mutations (especially SRSF2) and TP53 in PMF (P = .0003), and with ASXL1 and TP53 mutations in SMF (P < .0001). These mutations of poor prognosis were associated with biological features of scoring systems (DIPSS and MYSEC-PM score). Mutations in TP53/SRSF2 in PMF or TP53/ASXL1 in SMF were more frequent as the risk of these scores increased. This allowed for a better stratification of MF patients, especially within the DIPSS intermediate-1 risk group (DIPSS) or the MYSEC-PM high risk group. AML transformation occurred faster in SMF than in PMF and patients who transformed to AML were more SRSF2-mutated and less CALR-mutated at MF sampling. CONCLUSIONS: PMF and SMF have different but not specific molecular profiles and different prognosis depending on the molecular profile. This may be due to differences in disease history. Combining mutations and existing scores should improve prognosis assessment.
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