

Other genes mutated in MPN

Recent studies have found many genes mutated in MPN, often with low to medium frequency. Their importance in the diagnosis and prognostic evaluation is yet to be precised. Those found with significant frequency on MPN can be sequenced to affirm the clonality of a disorder. The most common are presented below, with a list of labs offering mutation screening.

CBL

CBL encodes for an ubiquitin ligase able to target cytokine receptors for proteasome degradation. Its mutation has been described in MPN and mixed MDS-MPN.

TET2

TET2 has been found mutated in various myeloid disorders with frequencies varying from 5 to 20% depending on the MPN subtype.

EZH2 – ASXL1

These genes play a role in the epigenetic regulation.

Country	Name	TET2	CBL	EZH2	ASXL1	Others
France	Dr Lippert		ex 7-9			DNMT3A ex 15-to-23
Germany	Dr Schnittger	yes	ex 8-9	yes	exon 12	
Italy	Prof. Vannucchi ; Dr. Guglielmelli	ex 3 to 11	ex 7-8	Ex 2 to 20	exon 12	IDH1/IDH2 ex 4; DNMT3A ex 15-to 23
Portugal	Dr Bento ; Dr Coucelo					LNK (ex-2)

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