

Appendix 4. Variants excluded from criteria BA1, BS1, and PM3 that require PM2_Supporting to be met

HGVS name	ithaID	Common name	Regions with high prevalence	PMIDs
NM_000518.5(HBB):c.92+5G>C	107	IVS I-5 (G>C)	Middle East, South-East Asia	9576331, 18294253, 12752111, 26865931
NM_000518.5(HBB):c.-79A>G	25	-29 (A>G)	Jamaica	10691857
NM_000518.5(HBB):c.79G>A	88	CD 26 GAG>AAG [Glu>Lys] (Hb E)	South-East Asia	26402558, 24488783
NM_000518.5(HBB):c.92G>C	100	CD 30 (G>C) or IVS I (-1) AGG>ACG (Arg>Thr)	India, Pakistan, Maldives	23162295
NM_000518.5(HBB):c.118C>T	142	CD 39 (CAG>TAG)	Mediterranean	1734721, 21353607, 18096416
NM_000518.5(HBB):c.126_129delC TTT	147	CD 41/42 (-TTCT or -CTTT or -TCTT)	South-East Asia	24534675, 26079343
NM_000518.5(HBB):c.20A>T	824	CD 6 GAG>GTG [Glu>Val] (Hb S)	Africa, South-East Asia, Mediterranean	28361595, 21353607, 26984585
NM_000518.5(HBB):c.19G>A	810	CD 6 GAG>AAG [Glu>Lys] (Hb C)	Africa, South-East Asia	2606477
NM_000518.5(HBB):c.364G>C	1217	CD 121 GAA>CA [Glu>Gln] (Hb D-Punjab)	Middle-East, South Asia	
NM_000518.5(HBB):c.27dupG	62	CD 8/9 +G	Middle East	22460247, 16533735
NM_000518.5(HBB):c.92+1G>A	101	IVS I-1 G>A	Mediterranean, Middle-East	21353607, 27199182, 18096416, 29637841
NM_000518.5(HBB):c.93-21G>A	113	IVS I-110 G>A	Mediterranean, Middle-East	27199182, 21353607
NM_000518.5(HBB):c.92+6T>C	111	IVS I-6 (T>C)	Mediterranean, Middle-East	25408857, 21353607

*ithaID: ID assigned by the IthaGenes database; access using <https://www.ithanet.eu/db/ithagenes?ithaID=<ithaID>>