

Supplement: Detailed dataset – all amino acid substitutions in ASXL1, EZH2, DNMT3A and TET2, with original literature

Gene	AAS	SNP/MUT	Disease	Reference
ASXL1	S25L	SNP		[1]
ASXL1	M29V	SNP		[1]
ASXL1	Q89R	SNP		[2]
ASXL1	A95V	SNP		[1]
ASXL1	C111Y	SNP		[1]
ASXL1	M168L	SNP		[2]
ASXL1	I268F	SNP		[2]
ASXL1	R394H	SNP		[3]
ASXL1	R402Q	MUT	MDS	[3]
ASXL1	S412F	SNP		[2]
ASXL1	E477Q	SNP		[2]
ASXL1	R489G	SNP		[2, 3]
ASXL1	I507N	SNP		[2]
ASXL1	R541W	SNP		[1]
ASXL1	I552V	SNP		[2]
ASXL1	A636V	SNP		[4]
ASXL1	A637T	SNP		[4]
ASXL1	G643V	SNP		[2]
ASXL1	G652S	SNP		[2, 4]
ASXL1	C687R	SNP		[2]
ASXL1	T688M	SNP		[4]
ASXL1	R693Q	SNP		[4]
ASXL1	G704R	SNP		[4]
ASXL1	P722R	SNP		[4]
ASXL1	D741V	SNP		[2]
ASXL1	G744R	MUT	MPN or MDS/MPN	[1]
ASXL1	V751I	SNP		[2]
ASXL1	P779L	SNP		[2]
ASXL1	E788G	MUT	MPN or MDS/MPN	[1]
ASXL1	D799Y	SNP		[2, 4]
ASXL1	T806A	SNP		[1]
ASXL1	P808H	MUT	MPN or MDS/MPN	[1]
ASXL1	T822I	MUT	MPN or MDS/MPN	[1]
ASXL1	L823S	SNP		[3]
ASXL1	K838R	SNP		[1, 2]
ASXL1	S846N	MUT	MDS, MDS/MPN	[5, 6]
ASXL1	E865K	SNP		[2, 4]
ASXL1	M875V	SNP		[4]
ASXL1	D979G	SNP		[1]
ASXL1	L983R	SNP		[2]
ASXL1	T1010M	SNP		[2]
ASXL1	G1026E	SNP		[2]

ASXL1	E1033V	SNP		[7]
ASXL1	P1035S	MUT	MPN or MDS/MPN	[1]
ASXL1	V1046M	SNP		[4]
ASXL1	R1051C	SNP		[4]
ASXL1	P1089S	MUT	MPN or MDS/MPN	[1]
ASXL1	V1092M	MUT	MPN or MDS/MPN	[1]
ASXL1	D1127N	SNP		[2]
ASXL1	M1161V	SNP		[1]
ASXL1	S1166R	SNP		[2, 3]
ASXL1	R1171S	SNP		[8]
ASXL1	G1187V	SNP		[7]
ASXL1	P1197S	SNP		[4]
ASXL1	R1247C	SNP		[2]
ASXL1	M1249V	SNP		[1, 2, 4]
ASXL1	D1252G	SNP		[2]
ASXL1	P1259L	SNP		[2]
ASXL1	V1297I	SNP		[2]
ASXL1	A1312V	SNP		[2-4]
ASXL1	R1316H	SNP		[4]
ASXL1	L1325F	SNP		[2, 9]
ASXL1	G1339E	MUT	MPN, Breast cancer	[10, 11]
ASXL1	A1380V	SNP		[1]
ASXL1	L1395V	SNP		[2, 12]
ASXL1	G1397S	SNP		[2-4]
ASXL1	M1402L	SNP		[1]
ASXL1	S1428P	SNP		[2, 4]
ASXL1	F1463L	SNP		[4]
ASXL1	A1486T	MUT	MDS	[3]
ASXL1	T1494I	SNP		[1]
ASXL1	S1496N	SNP		[1]
ASXL1	S1497G	SNP		[1]
ASXL1	T1498M	MUT	MPN or MDS/MPN	[1]
ASXL1	I1514L	SNP		[2]
ASXL1	C1519Y	MUT	MPN or MDS/MPN	[1]
EZH2	I55M	SNP		[2]
EZH2	M134K	MUT	MDS	[3]
EZH2	L149Q	MUT	MPN	[11, 13]
EZH2	D185H	SNP		[2]
EZH2	E249K	MUT	MDS	[11, 14, 15]
EZH2	Q250E	SNP		[2]
EZH2	N317S	SNP		[2]
EZH2	N361S	SNP		[3]
EZH2	A384T	MUT	MPN, Lung cancer	[11, 14]
EZH2	A482T	SNP		[2, 16]
EZH2	C534R	MUT	MDS	[3]
EZH2	T568I	MUT	MPN	[11, 13]

EZH2	L575P	MUT	MDS	[3]
EZH2	Y641C	MUT	AML, MDS, Lymphoid cancer	[11, 17-19]
EZH2	Q648E	MUT	MDS	[3]
EZH2	A651V	MUT	MDS	[11, 14]
EZH2	D652H	MUT	MDS	[3, 20]
EZH2	D659A	MUT	AML	[11]
EZH2	F655L	MUT	MDS	[3]
EZH2	L669V	MUT	MDS	[11, 15]
EZH2	D672H	MUT	MPN	[11, 13]
EZH2	V674M	MUT	MPN, AML	[11, 13]
EZH2	R679C	MUT	MPN, MDS/MPN	[11, 14]
EZH2	R685H	MUT	MDS, MPN, MDS/MPN, AML	[3, 5, 11, 13, 14, 21, 22]
EZH2	N688T	MUT	AML	[11, 22]
DNMT3A	E30A	SNP		[2]
DNMT3A	G278S	SNP		[2]
DNMT3A	S352N	MUT	AML	[11, 23]
DNMT3A	K468R	MUT	AML	[24]
DNMT3A	V480G	SNP		[2]
DNMT3A	N501S	SNP		[2]
DNMT3A	Y533C	MUT	AML, Lung cancer	[11]
DNMT3A	G543C	MUT	AML, MDS, Lymphoid cancer	[11, 25-28]
DNMT3A	W581C	MUT	AML	[11, 23]
DNMT3A	R635W	MUT	MDS, AML	[11, 27, 29, 30]
DNMT3A	V636M	MUT	AML	[11, 23, 30]
DNMT3A	I655A	MUT	AML	[11, 23]
DNMT3A	R676W	MUT	AML	[11]
DNMT3A	G699S	MUT	MDS	[11]
DNMT3A	S714C	MUT	MDS, AML, Breast cancer	[11, 26, 27, 29-31]
DNMT3A	P718L	MUT	AML	[11, 24]
DNMT3A	R729Q	MUT	AML	[11, 24]
DNMT3A	R729W	MUT	AML	[11, 24]
DNMT3A	F731C	MUT	AML	[11, 23]
DNMT3A	Y735C	MUT	AML	[11, 23]
DNMT3A	R736H	MUT	AML, Lymphoid cancer	[11, 24, 28]
DNMT3A	L737R	MUT	AML	[11, 29]
DNMT3A	A741V	MUT	AML	[11, 24]
DNMT3A	R749C	MUT	MPN, AML	[8, 11]
DNMT3A	R749P	SNP		[2]
DNMT3A	F752L	MUT	AML	[11, 23]
DNMT3A	W753G	MUT	AML	[11, 23]
DNMT3A	L754R	MUT	AML	[11, 23]
DNMT3A	S770W	MUT	AML	[11, 29]
DNMT3A	R771L	MUT	MDS	[11, 29]

DNMT3A	R771Q	MUT	MPN, AML, Lung cancer	[11]
DNMT3A	S775P	SNP		[2]
DNMT3A	R792H	MUT	AML, Lymphoid, Lung cancer	[11, 24, 28]
DNMT3A	W795C	MUT	AML	[11, 23]
DNMT3A	R803S	MUT	AML	[11, 24]
DNMT3A	K829R	MUT	AML	[11, 24]
DNMT3A	K841Q	MUT	AML	[11, 24]
DNMT3A	P849R	MUT	AML	[11, 23]
DNMT3A	M880V	MUT	MPN	[11, 32]
DNMT3A	R882C	MUT	MDS, MPN, MDS/MPN, AML, Lymphoid cancer	[5, 11, 23-28, 30, 32-41]
DNMT3A	R882H	MUT	MDS, MPN, MDS/MPN, AML, Lymphoid cancer	[5, 11, 23-30, 33-49]
DNMT3A	R882P	MUT	MDS/MPN, AML	[5, 11, 24, 26, 27, 36, 37]
DNMT3A	L889P	MUT	AML	[11, 23]
DNMT3A	V897D	MUT	AML	[11, 25, 26]
DNMT3A	P904L	MUT	AML, Lymphoid cancer	[11, 24, 28, 29]
DNMT3A	F909C	MUT	AML	[11, 24]
DNMT3A	A910V	MUT	AML, Endometrial, Intestinal cancer	[11, 23]
TET2	H8D	SNP		[2]
TET2	P22L	MUT	AML	[11, 43]
TET2	P29R	SNP		[2, 9, 50-53]
TET2	L34F	SNP		[2, 9, 50-55]
TET2	P101H	MUT	AML	[11, 51]
TET2	P174H	SNP		[2, 53, 54]
TET2	A215V	SNP		[56]
TET2	V218M	SNP		[2, 9, 50-53, 55]
TET2	A241V	SNP		[57]
TET2	S282F	MUT	AML	[11, 51]
TET2	A308T	MUT	AML	[58]
TET2	N312S	MUT	AML	[11, 54]
TET2	L346P	MUT	MDS/MPN	[11]
TET2	E350K	MUT	AML	[59]
TET2	G355D	SNP		[2, 9, 50-53]
TET2	P363L	SNP		[2, 9, 50-53, 55]
TET2	S393C	SNP		[52]
TET2	P399L	MUT	MDS/MPN	[58]
TET2	G429R	SNP		[2]
TET2	S460F	MUT	MDS/MPN, AML	[52, 54]
TET2	P472A	SNP		[2]
TET2	P474L	SNP		[2]
TET2	P480T	SNP		[56]
TET2	T492S	MUT	AML	[11, 51]
TET2	Q548L	MUT	AML	[11, 51]

TET2	D666G	MUT	MDS/MPN	[54]
TET2	S716L	SNP		[56]
TET2	P761L	SNP		[2]
TET2	N767D	SNP		[2, 60]
TET2	D768V	SNP		[56]
TET2	G773V	MUT	AML	[11, 51]
TET2	E788L	MUT	AML	[11, 61]
TET2	Q810R	SNP		[2, 9]
TET2	R814C	SNP		[62]
TET2	S817T	MUT	MDS/MPN	[58]
TET2	S826I	MUT	AML	[11, 61]
TET2	F868L	SNP		[62]
TET2	A912G	SNP		[2]
TET2	H924R	SNP		[2, 50, 51]
TET2	P941S	MUT	MDS/MPN	[54]
TET2	P968R	SNP		[52]
TET2	E1010D	SNP		[60]
TET2	S1015N	SNP		[56]
TET2	I1025T	SNP		[56]
TET2	S1039L	SNP		[62]
TET2	E1073V	SNP		[54]
TET2	Q1084P	SNP		[2, 50, 51, 53, 54, 56, 58]
TET2	L1120M	SNP		[2]
TET2	C1135Y	MUT	MDS, MPN, MDS/MPN	[54, 63, 64]
TET2	E1144A	MUT	MPN	[11]
TET2	R1167T	MUT	AML	[58]
TET2	C1193W	MUT	MDS, MDS/MPN	[54, 56, 65]
TET2	K1197R	MUT	MDS	[11, 66]
TET2	V1199I	MUT	AML	[11, 51]
TET2	S1204C	MUT	MPN	[54]
TET2	L1210P	MUT	MPN	[67]
TET2	C1211Y	MUT	MDS/MPN, AML	[11, 60, 68, 69]
TET2	R1214W	MUT	MDS, MDS/MPN, AML	[50, 53, 54, 65, 70]
TET2	H1219Q	MUT	AML	[11, 60]
TET2	H1219R	MUT	AML	[11, 60]
TET2	E1222G	MUT	AML	[11, 71]
TET2	L1229R	MUT	AML	[11, 53]
TET2	E1234G	MUT	AML	[11, 71]
TET2	D1242V	MUT	MPN, AML	[11, 54, 72-74]
TET2	Y1245S	MUT	MPN	[54]
TET2	L1248P	MUT	AML	[11, 53]
TET2	G1256A	MUT	AML	[11]
TET2	G1256D	MUT	AML	[11]
TET2	G1256R	MUT	AML	[71]
TET2	R1261C	MUT	MPN, MDS/MPN, AML	[11, 53, 54, 60, 68, 75]
TET2	R1261H	MUT	MDS/MPN, AML	[54, 55, 76]

TET2	C1263R	MUT	AML	[11, 60]
TET2	N1266Y	MUT	AML	[11]
TET2	T1270A	MUT	AML	[11, 61]
TET2	C1271S	MUT	AML	[11, 53]
TET2	C1271W	MUT	MDS, MDS/MPN, AML	[11, 51, 77-79]
TET2	C1271Y	MUT	AML	[11, 53]
TET2	G1275E	MUT	MDS, MPN, AML	[11, 56, 62, 67]
TET2	G1275R	MUT	MPN	[11, 57]
TET2	G1282D	MUT	MDS, AML	[11, 51, 56]
TET2	S1286P	MUT	MDS	[66]
TET2	F1287L	MUT	MDS/MPN	[58]
TET2	G1288S	MUT	AML	[11, 62]
TET2	G1288V	MUT	MPN, MDS/MPN	[54, 56]
TET2	C1289Y	MUT	AML	[11, 53, 60]
TET2	W1291R	MUT	MDS, AML	[11, 50, 62]
TET2	S1292R	MUT	AML	[11, 51]
TET2	C1298W	MUT	AML	[5, 59]
TET2	C1298Y	MUT	MDS/MPN, AML	[11, 60]
TET2	K1299N	MUT	MDS/MPN	[58]
TET2	R1302G	MUT	MPN, AML	[58, 73, 80]
TET2	E1318G	MUT	MDS/MPN	[58]
TET2	L1340R	MUT	AML	[11, 53]
TET2	A1355P	MUT	MDS	[56]
TET2	R1359C	MUT	MPN, MDS/MPN, AML	[11, 54, 62, 71, 72, 81, 82]
TET2	R1359H	MUT	MDS/MPN, AML	[11, 62, 65]
TET2	L1360Q	MUT	MDS	[66]
TET2	G1361D	MUT	MDS/MPN, AML	[11, 60, 68]
TET2	G1361R	MUT	MPN	[54]
TET2	R1366H	MUT	AML	[60, 62, 66, 75, 83]
TET2	P1367S	MUT	MDS/MPN	[58]
TET2	F1368L	MUT	AML	[11, 60]
TET2	F1368Y	MUT	AML	[11, 51]
TET2	G1370V	MUT	AML	[11, 51]
TET2	T1372I	MUT	MPN	[11, 67]
TET2	D1376E	MUT	AML	[11, 60]
TET2	D1376G	MUT	AML	[66]
TET2	D1376N	MUT	MPN	[11, 57]
TET2	H1380Y	MUT	MDS, MDS/MPN, AML	[11, 53, 56, 60-62, 76]
TET2	H1382Y	MUT	AML	[11, 60]
TET2	D1384H	MUT	MDS, AML	[66, 69]
TET2	L1398R	MUT	MDS	[11, 50]
TET2	V1417F	MUT	MDS, MDS/MPN, AML, Lymphoid cancer	[6, 11, 41, 54, 60, 84]
TET2	P1419R	MUT	MDS, AML, Lymphoid cancer	[11, 53, 66, 84]
TET2	D1427Y	MUT	AML	[11, 62]

TET2	R1440Q	SNP		[57]
TET2	A1505T	SNP		[60]
TET2	A1505V	MUT	AML	[11]
TET2	E1513G	SNP		[62]
TET2	V1521L	SNP		[43]
TET2	S1556Y	MUT	AML	[59]
TET2	Y1579S	MUT	AML	[11, 62]
TET2	N1641I	MUT	AML	[11, 71]
TET2	G1697R	SNP		[60]
TET2	M1701I	SNP		[2, 9, 50, 53, 55, 56]
TET2	G1719R	MUT	MPN	[59]
TET2	L1721W	SNP		[2, 9, 50-53, 55]
TET2	P1723S	SNP		[9, 50, 52-55, 57]
TET2	H1757D	MUT	MDS/MPN	[54]
TET2	I1762V	SNP		[2, 9, 50-53, 55]
TET2	H1778R	SNP		[2, 9, 50-53, 55]
TET2	L1801F	MUT	AML	[11, 62]
TET2	C1811R	MUT	MDS/MPN	[54]
TET2	L1816F	SNP		[2]
TET2	H1817N	MUT	MPN, AML	[75, 85]
TET2	Q1828L	MUT	MPN	[54]
TET2	A1831S	SNP		[9]
TET2	V1833I	SNP		[60]
TET2	A1863D	MUT	AML	[11]
TET2	A1865G	MUT	AML	[62]
TET2	H1868Y	MUT	MPN, AML	[11]
TET2	S1870P	MUT	AML	[11, 53]
TET2	I1871S	MUT	AML	[11, 62]
TET2	I1873N	MUT	MDS	[56]
TET2	I1873T	MUT	MDS, MPN, MDS/MPN, AML	[11, 50, 53, 54, 58, 60, 62, 65, 68, 72, 80, 83, 85-88]
TET2	C1875R	MUT	MDS	[11, 50]
TET2	A1876V	MUT	AML	[11, 60]
TET2	H1881R	MUT	MPN, MDS/MPN	[54, 68, 81]
TET2	A1882P	MUT	AML	[11, 60]
TET2	T1884A	MUT	MDS/MPN, AML	[53, 65, 68, 76]
TET2	P1889L	MUT	AML	[11]
TET2	R1896M	MUT	AML	[11, 75, 80]
TET2	R1896S	MUT	MDS	[11, 50]
TET2	T1898F	MUT	MPN, MDS/MPN, AML, Lymphoid cancer	[11, 69, 80, 84, 89]
TET2	L1899P	MUT	AML	[11, 60]
TET2	V1900A	MUT	AML	[54, 76]
TET2	H1904R	MUT	MDS, MPN, AML	[53, 56, 85, 86]
TET2	G1913D	MUT	MDS	[11, 50, 58]
TET2	A1919V	MUT	MPN	[54]

TET2	R1926H	MUT	MDS/MPN	[54]
TET2	E1929K	SNP		[2]
TET2	P1941S	MUT	MDS/MPN, AML	[54, 56]
TET2	R1966H	MUT	MDS/MPN	[54]
TET2	R1974M	MUT	MDS/MPN	[54]
TET2	T1979P	MUT	MDS	[6]
TET2	R2000K	MUT	AML	[11, 54, 71]

Supplemental references

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