Sample:	56G-HD701		

Sample 56G-HD701

Primary finding

Mutation	NM_002524.5:c.181C>A(p.Gln61Lys) **HGVS Protein:** Q61K(p.Gln61Lys) **HGVS Coding:** c.181C>A	**Classification**	Likely pathogenic
Mutation	NM_033360.4:c.38G>A(p.Gly13Asp) **HGVS Protein:** G13D(p.Gly13Asp) **HGVS Coding:** c.38G>A	**Classification**	Likely pathogenic
Mutation	NM_004333.6:c.1799T>A(p.Val600Glu) **HGVS Protein:** V600E(p.Val600Glu) **HGVS Coding:** c.1799T>A	**Classification**	Likely pathogenic
Mutation	NM_033360.4:c.35G>A(p.Gly12Asp) **HGVS Protein:** G12D(p.Gly12Asp) **HGVS Coding:** c.35G>A	**Classification**	Likely pathogenic
Mutation	NM_006218.4:c.3140A>G(p.His1047Arg) **HGVS Protein:** H1047R(p.His1047Arg) **HGVS Coding:** c.3140A>G	**Classification**	Likely pathogenic
Mutation	NM_006218.4:c.1633G>A(p.Glu545Lys) **HGVS Protein:** E545K(p.Glu545Lys) **HGVS Coding:** c.1633G>A	**Classification**	Likely pathogenic
Mutation	NM_000222.2:c.2447A>T(p.Asp816Val) **HGVS Protein:** D816V(p.Asp816Val) **HGVS Coding:** c.2447A>T	**Classification**	Likely pathogenic
Mutation	NM_005228.5:c.2155G>A(p.Gly719Ser) **HGVS Protein:** G719S(p.Gly719Ser) **HGVS Coding:** c.2155G>A	**Classification**	Likely pathogenic
Mutation	NM_005228.5:c.2573T>G(p.Leu858Arg) **HGVS Protein:** L858R(p.Leu858Arg) **HGVS Coding:** c.2573T>G	**Classification**	Likely pathogenic
Mutation	NM_005228.5:c.2235_2249del(p.Glu746_Ala750del) **HGVS Protein:** E746_A750del(p.Glu746_Ala750del) **HGVS Coding:** c.2235_2249del	**Classification**	Likely pathogenic

Mutation	NM_005228.5:c.2369C>T(p.Thr790Met) **HGVS Protein:** T790M(p.Thr790Met) **HGVS Coding:** c.2369C>T	**Classification**	Uncertain significance

KRAS information

Gene	KRAS (Synonyms: KRAS1)

Clinical Genomic Database (09_sep_2019)

Condition	Noonan syndrome; Cardiofaciocutaneous syndrome	**Inheritance**	AD
Comments		**Intervention categories**	Cardiovascular, Hematologic, Oncologic

NRAS information

Gene	NRAS (Synonyms: N-ras)

Clinical Genomic Database (09_sep_2019)

Condition	Autoimmune lymphoproliferative syndrome type IV; Noonan syndrome 6	**Inheritance**	AD
Comments	Noonan syndrome may include multiple congenital anomalies	**Intervention categories**	Cardiovascular, Oncologic

BRAF information

Gene	BRAF (Synonyms: BRAF1)

Clinical Genomic Database (09_sep_2019)

Condition	Noonan syndrome; Cardiofaciocutaneous syndrome 1; LEOPARD syndrome 3	**Inheritance**	AD
Comments	Conditions may be frequently clinically recognized due to characteristic facial features as well as other manifestations	**Intervention categories**	Cardiovascular, Hematologic, Oncologic

KIT information

Gene	KIT (Synonyms: C-Kit, CD117, SCFR)

Clinical Genomic Database (09_sep_2019)

Condition	Gastrointestinal stromal tumor; Mastocytosis, cutaneous	**Inheritance**	AD
Comments		**Intervention categories**	Oncologic

EGFR information

Gene	EGFR (Synonyms: ERBB1)

Clinical Genomic Database (09_sep_2019)

Condition	Acute myeloid leukemia, familial; Lung cancer, familial, susceptibilty to; Inflammatory skin and bowel disease, neonatal, 2	**Inheritance**	AD/AR
Comments	In familial AML, one family has been reported, in which there was an inherited genomic ERBB rearrangement	**Intervention categories**	Allergy/Immunology/Infectious, Cardiovascular, Oncologic

PIK3CA information

Gene	PIK3CA (Synonyms: PI3K)

Clinical Genomic Database (09_sep_2019)

Condition	Cowden syndrome 5	**Inheritance**	AD
Comments		**Intervention categories**	Oncologic