

Web Table I Genotype Phenotype Correlation in Cardio-facio-cutaneous Syndrome (CFC) and Noonan Syndrome – Like Disorder With Loose Anagen Hair (NSLAH)

<i>Disease</i>	<i>Mutation,</i>	<i>No.</i>	<i>Age, sex</i>	<i>Face, skin and hair</i>	<i>Cardiac</i>	<i>Cognition and development</i>
CFC(n=5)	<i>BRAF</i> , exon 6, c.770A>G, commonest, CFC-1	2	1 y, F	Coarse face, Curly hair bulbous nose tip	Pulmonary stenosis	Severe developmental delay, failure to thrive, feeding difficulty developmental delay, failure to thrive
			5 y, M	Coarse face, curly hair, ichthyotic skin hypopigmented facial macules	Pulmonary, stenosis Septal hypertrophy	
	<i>BRAF</i> , exon 15, c.1802A>T, CFC-1	1	2 y, M	Coarse face, curly hair,		Developmental delay, failure to thrive
	<i>MAP2K1</i> , Exon 3 . 389A>G, Pathogenic reported CFC-3	1	6 y, M	Coarse face, curly hair , hyperkeratosis Papilloma	Atrial septal defect	Developmental delay
	<i>MAP2K2</i> Exon 6, c.619G>A, likely pathogenic CFC-4	1	13 y, M	Coarse face Hyperkeratosis Curly hair Squint operated		Developmental delay, behavioral abnormalities
NSLAH (n=3)	<i>SHOC2</i> , c.4A>G	3	3 y, 4 y and 5 y,	Coarse facies Ptosis, sparse slow growing hair, hyper- keratosis, Café au lait spots, Thick gums, Thrombotic stroke Chin hemangioma,	Atrial septal defect (n=2)	Mild developmental delay

Web Table II Mutation Spectrum of Noonan Syndrome (N=19)

<i>Gene</i>	<i>No. of cases</i>	<i>Mutation</i>	<i>ClinVarId, Accession date – (23.5.2020)</i>	<i>OMIM</i>
<i>PTPN11</i> (n=11; 56%)	3	Exon 3, c.218C>T	VCV000013334.6	NS-1(163950)
ENST00000351677	1	Exon 3, c.179G>T	VCV000055797.2	
	1	Exon 2, c.124A>G	VCV000040482.3	
	1	Exon 8, c.923A>G	VCV000013327.8	
	1	Exon 8, c.922A>G	VCV000013326.12	
	2	Exon 12, c.1403C>T	VCV000013331.11	
	1	Exon 13, c.1510A>G	VCV000040562.15	
	1	Exon 13, c.1528C>G	VCV000040566.7	
<i>SOS1</i> (n=2; 11%)	2 (one Turner mosaic)	Exon 16, c.2536G>A	VCV000040706.6	NS-4(610733)
ENST00000426016		Exon 11, c.1654A>G	VCV000012871.7	
<i>RIT1</i> (n=2; 11%)	2	Exon 4, c.221C>G	VCV000060506.8	NS-8 (615355)
ENST00000368323		Exon 5, c.321G>A	VCV000190305.2	
<i>SOS2</i> (n=2; 11%)	2	Exon 6, c.800T>A	VCV000209092.3	NS-9(616559)
ENST00000216373		Exon 6, c.800T>G	VCV000577079.2	
<i>KRAS</i> (n=1; 6%)	1	Exon 2, c.13A>G	VCV000012596.3	NS-3(609942)
ENST00000311936				
<i>RAF1</i> (n=1; 6%)	1	Exon 17, c.1837C>G	VCV000013960.5	NS-5(611553)
ENST00000251849				