

Supplementary Table 1. List of causative genes of candidate diseases and summary of sequencing data

Gene	Disease	Mean depth (X)	% Bases above 10	% Bases above 20	% Bases above 30
<i>PTPN11</i>	Noonan syndrome	150.81	100	100	100
<i>SOS1</i>	Noonan syndrome	138.28	100	100	100
<i>RAF1</i>	Noonan syndrome	155.25	100	100	100
<i>KRAS</i>	Noonan syndrome	134.56	100	100	100
<i>NRAS</i>	Noonan syndrome	138.45	100	100	99.13
<i>BRAF</i>	Noonan syndrome	124.23	100	99.11	96.06
<i>MAPK1</i>	Noonan syndrome	125.28	100	100	99.60
<i>MAP2K1</i>	Noonan syndrome	157.97	100	100	100
<i>SHOC2</i>	Noonan-like disorders	159.68	100	100	100
<i>CBL</i>	Noonan-like disorders	155.33	100	100	100
<i>HRAS</i>	Costello Syndrome	222.94	100	100	100
<i>RPS6KA3</i>	Coffin-Lowry syndrome	117.41	100	99.76	98.64
<i>ATRX</i>	Alpha-thalassemia-X-linked intellectual disability syndrome	141.05	100	100	99.83
<i>PHF6</i>	Börjeson-Forsman-Lehmann Syndrome	133.43	100	100	100
<i>ELN</i>	Williams syndrome	135.40	100	100	100
<i>RFC2</i>	Williams syndrome	139.51	100	100	100
<i>LIMK1</i>	Williams syndrome	156.50	100	99.80	99.33
<i>CLIP2</i>	Williams syndrome	227.64	100	100	100
<i>GTF2I</i>	Williams syndrome	98.47	100	100	100
<i>GTF2IRD1</i>	Williams syndrome	165.01	100	100	100