

Laboratory for Molecular Medicine
65 Landsdowne Street,
Cambridge, MA 02139
Tel: 617-768-8500 / Fax: 617-768-8513
Email: LMM@PARTNERS.ORG

RASopathy and Noonan Spectrum Disorders (15 Genes)				
Gene	Expanded RASopathy Panel	Inheritance Pattern	Association	Syndromes/Clinical Features
<i>BRAF</i>	X	AD	Strong	Cardio-facio-cutaneous syndrome/Noonan
<i>CBL</i>	X	AD	Strong	Noonan syndrome/Juvenile Myelomonocytic Leukemia
<i>HRAS</i>	X	AD	Strong	Costello syndrome
<i>KRAS</i>	X	AD	Strong	Cardio-facio-cutaneous syndrome/Noonan syndrome
<i>MAP2K1</i>	X	AD	Strong	Cardio-facio-cutaneous syndrome
<i>MAP2K2</i>	X	AD	Strong	Cardio-facio-cutaneous syndrome
<i>NF1</i>	X	AD	Strong	Neurofibromatosis 1
<i>NRAS</i>	X	AD	Strong	Noonan syndrome/Juvenile Myelomonocytic Leukemia
<i>PPP1CB</i>	X	AD	Strong	Noonan syndrome-like disorder with loose anagen hair
<i>PTPN11</i>	X	AD	Strong	LEOPARD syndrome/Noonan syndrome/Juvenile Myelomonocytic Leukemia/Metachondromatosis
<i>RAF1</i>	X	AD	Strong	Noonan syndrome/Hypertrophic cardiomyopathy
<i>RIT1</i>	X	AD	Moderate	Noonan syndrome/Hypertrophic cardiomyopathy
<i>SHOC2</i>	X	AD	Strong	Noonan syndrome
<i>SOS1</i>	X	AD	Strong	Noonan syndrome
<i>SPRED1</i>	X	AD	Strong	Legius syndrome

www.partners.org/personalizedmedicine/Laboratory-For-Molecular-Medicine