

***PTPN11* Known Mutation Single Exon Sequencing**

Noonan syndrome (NS) is a dysmorphic syndrome with a number of distinctive characteristics. Noonan syndrome patients can present with short stature; congenital cardiac anomalies; broad or webbed neck; epicanthic folds, deafness, and characteristic facies. Also seen are an unusual chest shape with superior pectus carinatum, inferior pectus excavatum, apparently low-set nipples; a variable developmental delay, and cryptorchidism. Varied coagulation defects and lymphatic dysplasias are frequently observed. Congenital heart disease occurs in greater than 50% of NS individuals. Final adult height approaches the lower limit of normal. Mild mental retardation is seen in up to one third of NS patients however most do well in a normal educational setting, with perhaps 10-15% requiring special education. Clinical diagnosis of NS is made by observation of the key features outlined above.

NS is inherited in an autosomal dominant manner. Children with a Noonan syndrome parent have a 50% chance of inheriting the syndrome. Mutations in the gene *PTPN11* have been identified in ~50% of patients. Sequencing for all 15 *PTPN11* exons is available and is done at PreventionGenetics in a cost effective, sequential manner. This testing is described [elsewhere](#). In addition, we offer exon specific screening to confirm known familial mutations in additional family members.

A positive report listing a documented causative *PTPN11* mutation indicates only the potential for the development of NS. In addition, a negative report does not rule out other causative mutations in unscreened / non-coding regions of the *PTPN11* gene, in other known NS susceptibility genes, or in other as yet undefined genes that may contribute to NS Susceptibility.

PreventionGenetics is working closely with the Noonan Syndrome Support Group, Inc. on making this test more accessible to the NS patient population. There is a 10% price discount for Support group members. People interested in this test are encouraged to contact the Noonan Syndrome Support Group for more information (www.noonansyndrome.org) phone: 888-686-2224.

Specimen Requirements

- Collect 2-5 ml of whole blood in EDTA (purple top tube) or ACD (yellow top tube). 5 ml is the preferred volume.
- Only one blood tube is required for multiple tests.
- Ship whole blood specimens at room temperature.
- Do not freeze blood.
- During hot weather, include a frozen ice pack in the shipping container. Do not allow the ice pack to come in direct contact with the specimen tube.
- In cold weather, include an unfrozen ice pack to help moderate extremes in temperature. The DNA in whole blood is stable for at least 48 hours at 21°C, 5-7 days at 4°C.

Single exon Sequence analysis screening for the presence previously identified mutations in the *PTPN11* gene **\$230.00.**

Molec Diag, Ascertainment	83890	
Molec Diag, Isolation	83891	
Molecular Diag, Amplif	83898	Turnaround time is 1~2 weeks
Mutat Id By Seq, Single Seg	83904	
Molecular Diag, Separation	83894	
Interpretation and Report	83912	

Accreditation Info. CLIA ID #: 52D1027685 (expires 1/18/07) (CAP#: 7185561, AU ID: 1407125 expires 12/20/06)

Ship to:

Attn: Diagnostics Lab
PreventionGenetics LLC
3700 Downwind Drive
Marshfield, WI 54449 USA
Phone: 715-387-0484

Contact for info:

Dr. Eric W. Johnson
Director, Molecular Diagnostics and BioBanking
715-387-0484
clinicaltesting@preventiongenetics.com
FAX: 715-384-3661