Genetic Testing Laboratory Informed Consent for DNA testing for Noonan Syndrome

I, ____________________________, hereby request DNA based testing for Noonan syndrome. I have received verbal and/or written information from my physician or from a genetic counselor that described, in words that I understood, the nature of the genetic testing that I (or my minor child) am about to undergo.

I understand that blood samples will be drawn from me and/or members of my family or if prenatal testing is to be performed, fetal cells obtained by amniocentesis, chorionic villus sampling or by fetal blood sampling will be used. I understand that the samples will be used for determining if I and/or members of my family are carriers of a mutation in the PTPN11 gene which is the most common cause of Noonan syndrome and that mutations in the PTPN11 gene account for ~50% of Noonan cases (higher in familial than sporadic cases). Mutations in other genes responsible for Noonan syndrome have been identified, therefore, if a mutation is not found, I understand that an abnormality in another gene may exist. In addition, the overall ability of this test to find a PTPN11, if there is one, is ~98%.

The nature of DNA testing has been explained to me and the accuracy of the test and its limitations have been detailed. I understand that while results obtained from DNA testing are usually highly accurate, infrequent errors may occur. The likelihood of this occurring has been estimated to be less than 1%. An error in the diagnosis may occur if the true biological relationships of the family members involved in this study are not as I have stated and this test may detect non-paternity.

The results of my test will be explained to me by a genetic counselor or by my physician who will have the opportunity to discuss my results with a Clinical Geneticist. For prenatal tests, any question of contamination with maternal cells may result in no diagnosis being made and a repeat procedure may be necessary.

I understand that if I checked “No” on the statement below, no test will be performed and reported on my sample other than the one authorized by my doctor; and any unused portion of my sample will be discarded within 60 days of the receipt of the specimen.

I give consent to the Laboratory Director to use my sample for research purposes that might lead to further knowledge about Noonan syndrome or other related diseases. The sample used for research will be stripped of all identifiers and made anonymous. In addition, the sample will be destroyed within 20 years.

Yes ☐  No ☐

I have had the opportunity to have all of my questions answered. If I am signing this form on behalf of a minor for whom I am the legal guardian, I am satisfied that I have received enough information to sign on his or her behalf. I understand that this consent if being obtained in order to protect my right to have all of my questions answered before testing. I also understand that the results of this testing will become part of my medical record and may only be disclosed to individuals who have legal access to this record or to individuals who I designate to receive this information.

______________________________________ __________________
Signature of Person Being Tested (or guardian)  Date

______________________________________ __________________
Witness       Date