Molecular Diagnostics Lab
1600 Rockland Road, Wilmington, DE 19803
p: 302.651.6775 email: MDL@nemours.org

Test Requisition – page 1 of 2

Patient Information	
First Name	Last Name
Medical Record Number	Date of Birth (MM/DD/YYYY)
Sex assigned at birth O Female O Male	Gender (optional)
Sample Type O Blood O DNA O Buccal swab O Other_____ Collection date (MM/DD/YYYY)	

Ordering Physician (required)		Additional report recipient (genetic counselor, send-out lab)	
Name		Name	
Address		Address	
Phone	Fax	Phone	Fax
Email		Email:	
selection indicates your choice to send report via secure: O Fax O Email		**selection indicates your choice to send report via secure:** O Fax O Email	

Billing Information O Institutional Billing O Patient Billing

Please note: We do not bill third party payers (insurance companies). The person or organization sending the sample is responsible for full payment of the invoice. Please include a name of the billing contact at your organization and an email to send the invoice. If patient is paying by credit card – a completed credit card form must be included (or faxed to the lab) before testing can begin.

Organization	Email
Attention to	Phone Fax
Street	**send invoice via secure** O Email O standard mail
City/State/Zip Code	

Clinical Information

Brief description

O Symptomatic O Asymptomatic O Carrier testing O Variant Confirmation **(documentation required)**

O Family history (if mutation is known, please specify here and attach report)

Name and/or patient ID of family members previously tested in our lab

Pedigree attached O Yes O No O Adopted Consanguinity O Yes O No

For SMN sequencing: Number of copies of SMN1_____.
Attach SMA deletion testing or copy number report (**required**).

NEMOURS CHILDREN'S HEALTH

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○ **Acrodysostosis 1 –** *PRKAR1A* seq. ○ **Allan-Herndon-Dudley Syndrome (MCT8)** *SLC16A2* seq. ○ ***APOL1* Genotyping-** *APOL1* seq.	○ ***LIG4*- Related Disorders** - *LIG4* seq.
Autosomal Dominant Leukodystrophy (ADLD) ○ *LMNB1* gene dup. ○ *LMNB1* upstream deletion	**Megalencephalic Leukoencephalopathy w/ Subcortical Cysts** ○ *MLC1 gene* seq. ○ *HEPACAM* seq.
Autosomal Dominant Torsion Dystonia 4 (DYT4) ○ *TUBB4A* seq. ○ *DYT4* targeted mutation seq.	**Meier- Gorlin Syndrome** ○ *ORC1* seq ○ *ORC 6* seq. ○ *CDC6* seq ○ *ORC4* seq ○ *CDT1* seq
○ **Barth Syndrome-** *TAZ* seq.	**Microcephalic Osteodysplastic Primordial Dwarfism** **MOPD type I** **MOPD type II** ○ *RNU4ATAC* seq. ○ *PCNT2* sequencing
Benign Hereditary Chorea/Brain-Lung-Thyroid/CAHTP ○ *NKX2.1 / TTF1* seq. & del/dup ○ **Calcium Homeostasis Disorders-** *CASR* seq. ○ **Congenital Nongoitrous Hypothyroidism 2-** *PAX8* seq.	**Noonan Syndrome & CFC Syndrome** sequencing by tiered approach ○ Noonan ○ CFC ○ *PTPN11* seq. ○ RAF1 seq. ○ *SOS1* seq. ○ *SHOC2* seq. ○ *KRAS* seq. ○ *BRAF* seq. ○ *MEK1(MAP2K1) MEK2(MAP2K2)* seq.
Costello Syndrome ○ *HRAS* exons 2 & 3 seq. ○ *HRAS* full gene seq.	○ **Pelizaeus-Merzbacher Disease-** *PLP1* gene del/dup
○ **Deafness and Myopia Syndrome** – *SLITRK6* seq. ○ **DFNB59 Nonsyndromic Deafness** – *DFNB59* seq.	○ **Pelizaeus-Merzbacher Disease & Spastic Paraplegia 2-** *PLP1* seq.
○ **Duchenne muscular dystrophy** – DMD gene deletions	○ **Pelizaeus-Merzbacher Like Disease** *GJC2* seq.
○ **X-linked Emery-Dreifuss Muscular Dystrophy-** *EMD* seq. ○ **Escobar/Multiple Pterygium Syndrome-** *CHRNG* seq.	○ **Pendred Syndrome** -*SLC26A4* seq.
Familial Hypercholesterolemia (*LDLR, APOB, PCSK9*) ○ Tier I gene seq. w/ reflex ○ Tier II gene seq./ del/dup ○ Both tiers seq. simultaneously	**Pol III- Related Leukodystrophies, HLD7** and **HLD8** ○ *POLR3A* seq. ○ *POLR3B* seq.
○ **Familial Isolated Hypoparathyroidism** - *GCM2* seq.	**Renal hypouricemia Type 1 and Type 2** ○ Tier 1 *SLC22A12* seq. ○ Tier 2 *SLC2A9* seq. ○ Tier 3 *SLC2A9* del/dup
○ **Fatal Infantile Cardioencephalomyopathy**- *SCO2* seq. **Feingold Syndromes 1 & 2** analysis by tiered approach ○ *MYCN* seq. and del/dup ○ *miR 17-92* del/dup analysis	**Rett Syndrome** ○ *MECP2* seq. ○ *MECP2* del/dup analysis
***GJB2* and *GJB6* Related Disorders** ○ *GJB2* seq. ○ *GJB6 deletion &* seq.	**Sickle cell disease/ Sickle cell trait** - *HBB* ○ Targeted sequencing ○ Carrier Variant Testing
Glycogen Storage Disease Type V ○ *PYGM* select exons seq. (exons 1 & 5 only) ○ Exons 1 & 5 with reflex to remaining exons	○ **Smith-McCort Dysplasia -** *RAB33B* seq.
○ **Hypomyelinating Leukodystrophy 5-** *FAM126A seq.*	**Spinal Muscular Atrophy (SMA)** ○ *SMN* gene del/dup ○ *SMN* gene deletion <u>only</u> ○ *SMN* gene seq.
Hypomyelinating Leukodystrophy 6 (HABC) ○ *TUBB4A* targeted mutation seq. ○ *TUBB4A* full gene seq. ○ *TUBB4A* seq. by tiered approach	○ **SMA w/ Respiratory Distress (SMARD) -** *IGHMBP2* seq.
○ **Hypomyelinating Leukodystrophy 9**- *RARS* seq.	○ **Spondyloocular Syndrome (SOS) -** *XYLT2* sequencing
○ **Hypomyelinating Leukodystrophy 11 –** *POLR1C seq.*	○ **TARP Syndrome-** *RBM10* sequencing
Idiopathic Infantile Hypercalcemia ○ *CYP24A1* full gene seq. and copy number ○ *CYP24A1* Tier I seq. ○ *CYP24A1* Tier II seq.	○ **Timothy Syndrome-** *CACNA1C* sequencing- *exons 8 & 8a*
Leukoencephalopathy w/ Brainstem & Spinal Cord Involvement ○ *DARS2* full gene seq. ○ *DARS2* Tier I seq. ○ *DARS2* tier 2: seq. remaining exons	***TRPV4*-Related Disorders** ○ Full gene sequencing ○ Exons 5 & 6 sequencing only