How to order Genetic Testing – Molecular

Intro – Genetic testing is complex, expensive and takes a long time for results to return; but, despite these cons, it can be very beneficial to the patient and their family when a genetic diagnosis is obtained. Genetic testing is most appropriate when a Clinical Geneticist or Genetic Counselor is involved in the process. However, other specialist at CNMC will sometimes recommend genetic testing when asked to consult on an inpatient. When possible, it is best to defer inpatient testing to the outpatient setting to ensure the family undergoes consent for the testing and that the appropriate insurance authorization has been obtained. Unfortunately, there are critically ill children in our hospital who could benefit during that same hospitalization from genetic testing. The following is a primer on some of the commonly ordered genetic tests recommended in the inpatient setting to help ensure that it is ordered correctly in the EMR. Please do not hesitate to contact the genetics clinical service or the genetics lab with any questions.

1) Chromosomal Microarray (CMA) – First tier test for individuals with developmental delay and/or intellectual disability, congenital anomalies, and/or dysmorphic features
    a. Offered in house by Molecular Genetics Lab
    b. Draw 2 ml blood in EDTA Lavender top tube
    c. Turnaround time (TAT) is 2-4 weeks

2) Karyotype – Not typically ordered unless there is a high suspicion for Trisomy 13/18/21
    a. Not done in house, send out to Quest
    b. Draw 3 ml blood in Sodium Heparin Green top tube
    c. TAT is 2 weeks (STAT can be requested by calling Quest)

3) Cystic Fibrosis Genetic Testing
    a. Offered in house by Molecular Genetics Lab
    b. Always order "Cystic Fibrosis PCR 60 Mutations" first (search with 'cystic' and not 'cf')
        i. Draw 4 ml blood in EDTA Lavender top tube
        ii. TAT is 1 week
    c. Pending results and/or ethnic background, may also need to order "CFTR Sequencing" (ok to search with 'cf')
        i. Draw 1 ml blood in EDTA Lavender top tube but may already have DNA in lab
        ii. TAT is 2-4 weeks

4) Next-Gen Sequencing (NGS) – May be ordered as inpatient under extenuating circumstances but in general should be done as outpatient with appropriate insurance authorization. Generally done after CMA but occasionally done as first tier for very specific symptoms (phenotypes)
    a. Offered in house by Molecular Genetics Lab
    b. Currently able to sequence ~5,000 genes and will be offering ~7,000 by 2017
    c. Draw 1 ml blood in EDTA Lavender top tube
    d. TAT is 6-12 weeks
        i. Configured Panels (these have their own test codes within the system, searchable by the name of the panel)
            1. Brain
                a. STAT Epilepsy Panel
                b. Comprehensive Epilepsy Panel
                c. Several other pre-built gene lists for neurology indications available as PSPs by Gene List (see below)
            2. Heart
                a. ARVD Panel
                b. CPVT Panel
                c. Long QT Syndome
                d. Brugada Syndrome Panel
                e. Hypertrophic Cardiomyopathy Panel
                f. Dilated Cardiomyopathy Panel
                g. Left Ventricular NonCompaction Panel
                h. Comprehensive Cardiomyopathy Panel
                i. Comprehensive Arrhythmia Panel
                j. Arrhythmia & Cardiomyopathy Panel

3. Bones
    a. NAT Panel (COL1A1/COL1A2 sequencing only)
    b. Dominant Osteogenesis imperfecta Panel
4. Connective Tissue
    a. FBN1 Sequencing (Marfan syndrome)
    b. Ectopia Lentis Panel
    c. Cutis Laxa Panel
    d. Aortic Aneurysm Panel
    e. Loeys-Dietz Panel
    f. Ehlers Danlos Syndrome Panel
    g. Stickler Syndrome Panel
    h. Comprehensive Connective Tissue Panel
5. Hearing Loss
    a. Connexin Panel
    b. Comprehensive Hearing Loss Panel
6. Other
    a. Noonan Spectrum Disorders Panel
    b. PTEN Sequencing

ii. Personalized Sequencing Panels (aka PSPs) – total genes ordered can range from 1 to 500
1. Personalized Sequencing Panel Gene List
    a. Order when you or the consultant has a specific list of genes in a consult note
    b. Order when there is a pre-built list in the lab for indications not listed above as a configured panel (neuromuscular disorders, autism/intellectual disability, mitochondrial disorders, congenital disorders of glycosylation, congenital brain malformations)
    c. Consider ordering when consultant asks for test to be sent out to Lab ABC, odds are they are unaware it can actually be done in house
    d. OK to contact the laboratory directly to find out more information
2. Personalized Sequencing Panel Phenotype
    a. Order when you or the consultant wants the lab to create the gene list based on the patient's clinical features. Please ensure that there is excellent documentation in the EMR!
    b. Gene list is created by Lab Med Genetic Counselors. It would be beneficial to notify the Genetic counselors by email to alert them, as this list needs to be compiled and approved within 3 days of ordering date. See email contact below.
    c. OK to contact the laboratory directly to find out more information

5) None of the above / Totally lost / Still confused / Ready to give up
a. Consider ordering 'DNA Extraction' if there is concern patient will expire before contacting the Geneticist on call
    i. Draw 5 ml blood in EDTA Lavender top tube
    ii. Contact the Lab directly to notify that DNA extraction was ordered and that a genetic testing will be added on in near future. If this is not done, testing will not be initiated. Once a specific gene test is decided upon, the lab med genetic counselors need to be informed so that testing is initiated. Without additional communication, the DNA will remain in the lab without any testing performed.
    iii. There is a limited amount of time to add on testing to a child who expires. Therefore, the communication with the lab needs to be timely.
b. Still need to contact the Molecular Genetics Lab for assistance?
    i. Call us: 202-476-2631
    ii. Email us: LabMed_Genetic_Counselors@childrensnational.org; kcozog@cnmc.org; shofherr@cnmc.org
    iii. Hours of operation: M-F 9am-5pm