Genetic testing on newborns in the NICU

Quick reference guide

Up to 30%[1] of babies in the Neonatal Intensive Care Unit (NICU) setting have an underlying genetic condition. Receiving a genetic diagnosis while in the NICU may aid the medical team with additional information to further create a personalized care plan, which may include targeted therapies, procedures or redirection of care. The following list is meant to be a guide to help aid clinicians in identifying babies that may benefit from genetic testing.

Inclusion criteria: Baby in the NICU born 32+ weeks gestation and without a planned discharge date.

Clinical indications where a NICU panel/exome may be beneficial:

Anomalies

- ☐ One or more congenital anomalies/birth defects that are not known to be caused by maternal alcohol, drug or teratogen exposure
- ☐ Abnormalities of skeleton, joint or muscle mass
- ☐ Neurological abnormalities (including seizures) not resulting from birth asphyxia
- ☐ Unexplained pulmonary disease, such as pulmonary hypoplasia, alveolar dysplasia, surfactant deficiency, recurrent pneumonia and bronchiectasis, but excluding transient tachypnea and other pulmonary complications of prematurity
- ☐ Deafness, sensorineural hearing impairment or inner ear malformation without previous history of congenital infection or meningitis
- ☐ Vision impairment including glaucoma, cataracts, coloboma, abnormalities of the retina and optic nerve
- ☐ Skin disease including abnormal pigmentation, neurocutaneous syndromes, congenital vesicular eruptions, ichthyosis and epidermolysis bullosa
- ☐ Cardiovascular disorder including cardiomyopathy, left ventricular noncompaction, atrial or ventricular arrhythmia, aortic dilation; arterial tortuosity and vasculopathy including diffuse vascular calcification or Moyamoya
- ☐ Functional gastrointestinal disease including malabsorption, brush border enzyme deficiencies and pseudo-obstruction
- ☐ Renal disease such as cystic renal disease, prune belly and obstructive uropathy, nephrotic syndrome, nephronophthisis and unexplained renal failure

Laboratory/endocrine

- ☐ Rare metabolic abnormalities including anion gap acidosis, lactic acidemia, hyperammonemia, persistent hypoglycemia, direct hyperbilirubinemia, abnormal plasma or urine amino acids, abnormal plasma or urine organic acids, abnormal carnitine metabolites and other metabolic disorders as designated by the treating physician or consultants
- ☐ Rare clinical or laboratory abnormalities (e.g., laboratory test results far outside of expected ranges, rare anatomical variants and abnormalities in newborn screening)
- ☐ Endocrine disorder, such as dyshormonogenesis, panhypopituitarism or abnormal development of an endocrine organ, like thyroid or adrenal glands; congenital diabetes and hyperinsulinism; persistent hypocalcemia
- ☐ Primary immune deficiency including thymic hypoplasia, lymphopenia, immunoglobulin deficiencies, T-cell deficiencies, NK cell deficiencies, complement deficiencies, recurrent severe infections or autoinflammatory disorder
- ☐ Disorders of blood and coagulation including congenital anemia, persistent thrombocytopenia, neutropenia and pancytopenia; recurrent thrombosis and bleeding disorders

General

- ☐ A clinical feature that may implicate a genetic cause, but the features do not correspond with a disorder for which a genetic test is available
- ☐ Multiple clinical features that are insufficient by themselves to make a clinical diagnosis, but that are known to be associated with multiple genetic disorders
- ☐ Family history of occurrence of similar disorder in a close relative
- ☐ The patient has an atypical course for the clinical diagnosis (e.g., unexpected severity, duration, failure of response to therapy)
- ☐ Congenital leukemia, tumors or other neoplastic disease; bilateral and second site tumors; multiple neoplasms of different tissue origin

Provider judgment

- ☐ The clinical presentation of the member in the NICU does not fall into a defined category above and results would impact treatment decisions.

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[1] Wojcik, M.H., Schwartz, T.S., Yamin, I., et al. Genetic disorders and mortality in infancy and early childhood: Delayed diagnoses and missed opportunities. *Genet Med.* 2018;20(11):1396-1404. doi: 10.1038/gim.2018.17