**LETTER OF MEDICAL NECESSITY FOR NOONAN SYNDROME RELATED DISORDERS/RASOPATHIES GENETIC TESTING**

Date: Date of service/claim

To: Utilization Review Department

Insurance Company Name, Address, City, State

Re: Patient Name, DOB, ID #

ICD-10 Codes:

The ICD-10 codes listed below are commonly received by Ambry from ordering providers for the testing described in this letter. Ambry provides this information as a customer service but makes no recommendations regarding the use of any diagnosis codes. As a reminder, it is the ordering provider’s responsibility to always determine, for the specific date of service, the appropriate diagnostic codes based on the patient’s signs and symptoms.

Code Description

I42.2 OTHER HYPERTROPHIC CARDIOMYOPATHY

I28.8 OTHER DISEASES OF PULMONARY VESSELS

Q24.4 CONGENITAL SUBAORTIC STENOSIS

F84.9 PERVASIVE DEVELOPMENTAL DISORDER, UNSPECIFIED

R93.1 ABNORMAL FINDINGS ON DIAGNOSTIC IMAGING OF HEART AND CORONARY CIRCULATION

Z80.7 FAMILY HISTORY OF OTHER MALIGNANT NEOPLASMS OF LYMPHOID, HEMATOPOIETIC AND RELATED TISSUES

This letter is regarding my patient and your subscriber, referenced above, to request full coverage of medically indicated genetic testing for Noonan syndrome related disorders to be performed by Ambry Genetics Corporation.

Noonan syndrome related disorders (NSRDs), also known as RASopathies, are a group of developmental conditions that are caused by genes in the Ras/MAPK pathway. Dysregulation of this pathway, caused by pathogenic germline variants in the NSRD genes, can cause disruption in normal growth and development.1,2 There is considerable variability and overlap in clinical features between the NSRDs, which can make diagnosis challenging without genetic testing.

**Significant aspects of my patient’s medical and/or family history that suggest an NSRD are as follows: [check all that apply]**

* Developmental delay and/or intellectual disability
* Dysmorphic facial features
* Heart defects such as pulmonary valve stenosis and others
* Hypertrophic cardiomyopathy
* Lymphatic dysplasia
* Pectus carinatum or excavatum
* Postnatal growth deficiency/short stature
* Skin/ectodermal lesions and/or pigmentation anomalies
* Undescended testes/cryptorchidism
* Webbed and/or short neck
* Other: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

**The American Academy of Pediatrics has recognized the clinical utility of genetic testing for NSRDs and support it as standard of care.**3-5

Identification of a mutation in one of these genes through genetic testing can confirm a diagnosis of an NSRD.Genetic testing also informs prognosis, physician referrals, screening and treatment options, and genetic counseling, which can vary depending on the specific gene implicated in the disease. 2-9 **Published management guidelines are available for many NSRDs.**3-5,9 Specifically for this patient, the impact of testing may include: [check all that apply]

* Confirmation of a suspected diagnosis
* Aid in diagnosis for patients with an atypical presentation of disease
* Allow tailoring of medical treatment based on specific gene results
* Allow immediate management and treatment to anticipate and control common clinical findings associated with NSRDs
* Assist in long-term management and monitoring of suspected disease progression
* Guide informed decision making for other family members with similar conditions, or who may be at risk for similar conditions
* Other: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

**Based on the treatment modifications indicated above, this test has clear clinical utility for my patient.** I am requesting coverage for this testing as medically necessary care and affirm that my patient has provided informed consent for genetic testing. I recommend that you support this request for coverage of diagnostic genetic testing for NoonanNext for my patient.

Thank you for your time, and please don’t hesitate to contact me with any questions.

Sincerely,

Ordering Clinician Name (Signature Provided on Test Requisition Form)

(MD/DO, Clinical Nurse Specialist, Nurse-Midwives, Nurse Practitioner, Physician Assistant, Genetic Counselor\*)

\*Authorized clinician requirements vary by state

Test Name: NoonanNext

CPT codes: 81442

Laboratory: Ambry Genetics Corporation (TIN 33-0892453 / NPI 1861568784), a CAP-accredited and CLIA-certified laboratory located at 7 Argonaut, Aliso Viejo, CA 92656

**References**

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7. Aoki Y, *et al.* Gain-of-function mutations in RIT1 cause Noonan syndrome, a RAS/MAPK pathway syndrome. [Am J Hum Genet.](https://www.ncbi.nlm.nih.gov/pubmed/23791108) 2013 Jul 11;93(1):173-80.
8. Gripp KW, *et al.* Five additional Costello syndrome patients with rhabdomyosarcoma: proposal for a tumor screening protocol. [Am J Med Genet.](https://www.ncbi.nlm.nih.gov/pubmed/11857556) 2002 Feb 15;108(1):80-7.
9. Gripp KW, *et al.* Costello syndrome: Clinical phenotype, genotype, and management guidelines. Am J Med Genet. 2019 Sept; 179(9):1725-1744.