Noonan Spectrum Disorders/RASopathies Multigene Panel

I. The use of a multigene panel to confirm or establish a diagnosis of a Noonan spectrum disorder/RASopathy (e.g., Noonan syndrome, Legius syndrome, Costello syndrome, Cardio-facial-cutaneous syndrome, NF1, Noonan-like syndrome) is considered **medically necessary** when:

   A. The member has at least one of the following:

      1. Characteristic facies (low-set, posteriorly rotated ears with fleshy helices, vivid blue or blue-green irises, widely spaced, down slanted eyes, epicanthal folds, ptosis), **OR**
      2. Short stature, **OR**
      3. Congenital heart defect (most commonly pulmonary valve stenosis, atrial septal defect, and/or hypertrophic cardiomyopathy), **OR**
      4. Developmental delay, **OR**
      5. Broad or webbed neck, **OR**
      6. Unusual chest shape with superior pectus carinatum, inferior pectus excavatum, **OR**
      7. Widely spaced nipples, **OR**
      8. Cryptorchidism in males, **OR**
      9. Lentigines, **OR**
      10. Café au lait macules.

II. The use of a multigene panel to confirm or establish a diagnosis of a Noonan spectrum disorder/RASopathy (e.g., Noonan syndrome, Legius syndrome, Costello syndrome, Cardio-facial-cutaneous syndrome, NF1, Noonan-like syndrome) is considered **investigational** for all other indications.

DEFINITIONS

1. **Developmental delay** (DD) is defined as slow-to-meet or not reaching milestones in one or more of the areas of development (communication, motor, cognition, social-emotional, or adaptive skills) in the expected way for a child's age.

REFERENCES

1. Roberts AE. Noonan Syndrome. 2001 Nov 15 [Updated 2022 Feb 17]. In: Adam MP, Mirzaa MP, Pagon RA, et al., editors. GeneReviews [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2024. Available from: https://www.ncbi.nlm.nih.gov/books/NBK1124/
2. Rauen KA. The RASopathies. *Annu Rev Genomics Hum Genet*. 2013;14:355-369. doi:10.1146/annurev-genom-091212-153523.