Other Covered Immune, Autoimmune, and Rheumatoid Disorders

The following is a list of conditions that have a known genetic association. Due to their relative rareness, it may be appropriate to cover these genetic tests to establish or confirm a diagnosis.

I. Genetic testing to establish or confirm one of the following immune, autoimmune, or rheumatoid disorders to guide management is considered **medically necessary** when the member demonstrates clinical features consistent with the disorder (the list is not meant to be comprehensive, see II below):

A. Agammaglobulinemia: X-Linked and Autosomal Recessive (*BTK*)

B. Autoimmune Lymphoproliferative Syndrome (ALPS) (*FAS*)

C. Chronic Granulomatous Disease (CGD) *(CYBA, CYBC1, NCF1, NCF2,* and *NCF4, CYBB)*

D. Complement Deficiencies

E. Congenital Neutropenia Syndromes (e.g., *ELANE*-Related Neutropenia) (*ELANE, HAX1)*

F. Familial Hemophagocytic Lymphohistiocytosis (HLH) (*PRF1*, *STX11*, *STXBP2*, or *UNC13D*)

G. Hyper IgE Syndrome (HIES) (*STAT3*)

H. Hyper IgM Syndromes (*CD40LG*)

I. Leukocyte Adhesion Deficiency (LAD) (*CD18, Kindlin-3, ITGB2)*

J. NEMO Deficiency Syndrome (*NEMO,* aka *IKK gamma* or *IKKG*)

K. Severe Combined Immune Deficiency (SCID) and Combined Immune Deficiency (*IL2RG*)

L. WHIM Syndrome (Warts, Hypogammaglobulinemia, Infections, and Myelokathexis) (*CXCR4)*

M. Wiskott-Aldrich Syndrome (*WAS*).

II. Genetic testing to establish or confirm the diagnosis of all other immune, autoimmune, or rheumatoid disorders not specifically discussed within this or another medical policy will be evaluated by the criteria outlined in the *General Approach to Laboratory Testing* (see policy for coverage criteria).

NOTE: Clinical features for a specific disorder may be outlined in resources such as GeneReviews, OMIM, National Library of Medicine, Genetics Home Reference, or other scholarly source.

REFERENCES

1. Immune Deficiency Foundation. “Specific PI Diagnoses”. 2020. https://primaryimmune.org/specific-pi-diagnoses. Accessed April 19th, 2024.
2. Adam MP, Ardinger HH, Pagon RA, et al., editors. GeneReviews [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2024. Available from: https://www.ncbi.nlm.nih.gov/books/NBK1116/
3. Online Mendelian Inheritance in Man, OMIM. McKusick-Nathans Institute of Genetic Medicine, Johns Hopkins University (Baltimore, MD). World Wide Web URL: https://omim.org/
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