

## Diagnostic Criteria

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|--|--|
| Category   | <input type="radio"/> Mutation in IKBKG (NEMO, IKK-gamma)  |
|  | <input type="radio"/> Mutation in NFKBIA (I-kappa-B-alpha) |
|  | <input type="radio"/> None of these apply                  |
| Patient has experienced ectodermal dysplasia and/or abnormal tooth shape | <input type="radio"/> Yes                                  |
|  | <input type="radio"/> No                                   |

### MUTATION IN IKBKG

- Select all that apply
- Atypical mycobacterial infection
  - Disseminated cytomegalovirus
  - Ectodermal dysplasia or abnormal teeth
  - Family history of IKBKG or NFKBIA mutation
  - Maternal cousins, uncles, or nephews with confirmed diagnosis of NEMO deficiency
  - Pneumocystis disease
  - Reduced or Absent toll-like receptor-induced TNF
  - Serum IgG concentration at least 2 SD below the normal for age

### MUTATION IN NFKBIA

- Select all that apply
- Atypical mycobacterial infection
  - Defective polysaccharide-specific antibody production
  - Deficient antigen-induced lymphocyte proliferation
  - Disseminated cytomegalovirus
  - Functional NK cell deficiency
  - Inflammatory bowel disease
  - Lymphedema
  - Osteoporosis
  - Pneumocystis disease
  - Reduced or Absent toll-like receptor-induced TNF
  - Septicemia
  - Serum IgG greater than 2 SD above normal for age
  - Serum IgA at least 2 SD below normal for age
  - Serum IgM at least 2 SD above normal for age

## Testing

Toll-Like Receptor Function

- All receptors tested abnormal
- Some receptors tested abnormal
- Normal function
- Unknown

Response to TNF

- Absent
- Decreased
- Normal
- Unknown

CD40 Function

- Abnormal CD40-induced class switching
- Abnormal CD40-induced proliferation
- Abnormal CD40-induced B cell activation
- Normal CD40 function
- Unknown

I $\kappa$ B Degradation

- Absent
- Decreased
- Normal
- Unknown