

Diagnostic Criteria

- Category
- X-Linked Agammaglobulinemia
 - Autosomal Recessive Agammaglobulinemia

X-LINKED AGAMMAGLOBULINEMIA

- Select all that apply
- Patient is Male
 - Patient has less than 2% CD19+ B Cells
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- Select all that apply
- Mutation in BTK
 - Absent Btk mRNA on northern blot analysis of neutrophils or monocytes
 - Absent or reduced amount of Btk protein in monocytes or platelets
 - Maternal cousins, uncles or nephews with less than 2% CD19+ B cells
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- Select all that apply
- Other causes of hypogammaglobulinemia have been excluded
 - Onset of recurrent bacterial infection in the first 5 years of life
 - Serum IgG, IgM and IgA more than 2SD below normal for age
 - Absent isohemagglutinins and/or poor response to vaccines

AUTOSOMAL-RECESSIVE AGAMMAGLOBULINEMIA

- Select all that apply
- X-Linked Agammaglobulinemia has been excluded
 - Patient has less than 2% CD19+ B cells
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- Select all that apply
- Mutation in BLNK
 - Mutation in Ig Alpha
 - Mutation in Ig Beta
 - Mutation in Lambda V
 - Mutation in Mu heavy chain
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- Select all that apply
- Onset of recurrent bacterial infection in the first 5 years of life
 - Serum IgG, IgM and IgA more than 2SD below normal for age
 - Absent isohemagglutinins and/or poor response to vaccines