

Agammaglobulinemia Registry Data Collection Form

Patient Initials: ___/___/___

Patient Identification:

Patient Name (first, middle, last) _____

Patient's USIDNET Registry Number assigned after online enrollment _____

Date of Birth ___/___/___ (mm/dd/yyyy) or Year of Birth _____

Gender: male [], female []

Home Address:

Address: _____

State: _____

Zip Code: _____

Phone: _____

Email: _____

State or Province of birth: _____

Country of birth: _____

Date of this Record Completion (mm/dd/yyyy): ___/___/___

Date of Visit (mm/dd/yyyy): ___/___/___

Is this the initial registration of this patient [] or follow-up? []

Submitting Physician Information:

Name: _____

Address: _____

State: _____

Zip Code: _____

Phone: _____

Email: _____

Fax: _____

Diagnostic Criteria for X-Linked Agammaglobulinemia

Skip this section for all agammaglobulinemias except XLA.

Definitive []

Male patient with less than 2% CD19+ B cells and **at least one of the following.**

- [] 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

Probable []

Male patient with less than 2% CD19+ B cells in whom **all of the following are positive.**

- [] 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
- [] Other causes of hypogammaglobulinemia have been excluded

Possible []

Male patient with less than 2% CD19+ B cells in whom other causes of hypogammaglobulinemia have been excluded **and at least one of the following is positive.**

- [] Onset of recurrent bacterial infections in the first 5 years of life
- [] Serum IgG, IgM and IgA more than 2 SD below normal for age
- [] Absent isohemagglutinins

Diagnostic Criteria for Autosomal Recessive Agammaglobulinemia

Skip this section for all agammaglobulinemias except autosomal recessive

Definitive []

Male or female patient with less than 2% CD19+ B cells in whom X-linked agammaglobulinemia has been excluded **and:**

- [] **Mutation in Lambda V**
- [] **Mutation in Ig Alpha**
- [] **Mutation in Ig Beta**
- [] **Mutation in Mu heavy chain**
- [] **Mutation in BLN**

Possible []

Male or female patient with less than 2% CD19+ B cells in whom X-linked agammaglobulinemia has been excluded **and at least one of the following is positive.**

- [] Onset of recurrent bacterial infections in the first 5 years of life
- [] Serum IgG, IgM and IgA more than 2 SD below normal for age
- [] Absent isohemagglutinins

Tests Performed:

Indicate those tests/data used to establish/confirm the diagnosis.

Additional Serum Immunoglobulin

IgG1 _____ mg/dl

IgG2 _____ mg/dl

IgG3 _____ mg/dl

IgG4 _____ mg/dl

SIG (+) B cells

Blood _____ %

Flow cytometry for Btk Negative Reduced Indeterminant Normal

Flow cytometry for B cell markers

CD20 _____% _____/ul

CD19 _____% _____/ul

Genetic Information

Sporadic [] (no family history) or X-linked [] pattern of inheritance

Skewed X-chromosome Inactivation of B cells in mother / related female carrier (who? _____)

Yes [], No [], Not tested [], gene tested? _____

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Gene Mutation

(Number nucleotides using Human Mutation 11:1-3, 1998)

Btk Lambda V Ig alpha Ig beta
Mu heavy chain BLNK

Mutation analysis performed by _____

Nucleotides affected (e.g., 229A>C) _____

Predicted Amino Acid Change (e.g., T33P) _____

Insertion / Deletion / Frameshift / Splice Site (please explain) : _____

Mutation tested for but not found _____

Publications (please give citation - if published) _____

Is the protein or mutant protein expressed? Yes / No / Unknown

Cells tested

Chose from the following: **Tcells (T), B/pre-B (B), monocytes (M), platelets (P), whole mononuclear (MNL), bone marrow (BM), other cells?** _____

protein	Yes	No	Not tested	By Western	By FACS	Other?	Cells
BTK	[]	[]	[]	[]	[]	_____	_____
Lambda V	[]	[]	[]	[]	[]	_____	_____
Ig alpha	[]	[]	[]	[]	[]	_____	_____
Ig Beta	[]	[]	[]	[]	[]	_____	_____
Mu H chain	[]	[]	[]	[]	[]	_____	_____
BLNK	[]	[]	[]	[]	[]	_____	_____
Other	[]	[]	[]	[]	[]	_____	_____

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Pedigree Analysis

Family history unknown

Please list additional relationships. If more space is needed, please use the Memo section at the end of this form.

Relation	XLA or AR Agamma – alive	XLA or AR Agamma – deceased	Tested Normal	Carrier	Not Tested	Unknown	Undiagnosed male or female with suggestive symptoms
Mother							
Father							

Information on other affected kindred members listed above (e.g., sibling, cousin, &/or maternal uncle):

Relation	Initials	Year of Birth	Gender	Listed in Registry? Yes/No/Unknown

Clinical Features of Special Note List all complications at the end of this section *.

	Observed	Prominent	Unknown	Organisms
Enterovirus Encephalitis				
Unexplained CNS Deterioration				
Neutropenia				
Malabsorption				
Dermatomyositis not infectious				
Dermatomyositis- enteroviral				
Arthralgia not infectious				
Arthritis not infectious				
Arthritis - enteroviral				
Arthralgia-chronic viral				
Nephritis				
Colon Cancer				
Pseudomonas skin infection				
Other – Specify:				
Comments:				

*Complications:

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Other treatments used List all complications at the end of this section *.

	Yes	No	Unknown	Age begun		OR Date begun (mm/dd/yyyy)	Duration	Still needed?	
				Months	Years			Yes	No
Chronic oxygen									
Parenteral nutrition									
Other _____									

Other? Please explain

Additional comments:
