



Wiskott-Aldrich Registry Data Collection Form

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: _____



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Diagnostic Criteria

Definitive []

Male patient with congenital thrombocytopenia (less than 70,000 platelets/mm³), small platelets and **at least one of the following:**

- Mutation in WASP
- Absent WASP mRNA on northern blot analysis of lymphocytes
- Absent WASP protein in lymphocytes
- Maternal cousins, uncles or nephews with small platelets and thrombocytopenia

Probable []

Male patient with congenital thrombocytopenia (less than 70,000 platelets/mm³), small platelets and **at least one of the following:**

- Eczema
- Abnormal antibody response to polysaccharide antigens
- Recurrent bacterial or viral infections
- Autoimmune diseases
- Lymphoma, leukemia

Possible []

Male patient with thrombocytopenia (less than 70,000 platelets/mm³) and small platelets; **or**
A male patient splenectomized for thrombocytopenia who has **at least one of the following:**

- Eczema
- Abnormal antibody response to polysaccharide antigens
- Recurrent bacterial or viral infections
- Autoimmune diseases
- Lymphoma, leukemia

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Spectrum of disease

Congenital thrombocytopenia with small platelets is the diagnostic hallmark of Wiskott-Aldrich syndrome. Many patients present with bloody diarrhea in the first month of life. Eczema, which occurs in some but not all patients, may be the predominant clinical problem. Otitis and sinusitis, and infections due to herpes simplex and EBV are particularly troublesome. Many patients have increased IgE and IgA, with low IgM. There is an increased incidence of autoimmune manifestations (vasculitis, hemolytic anemia, glomerulonephritis) and tumors (leukemia, lymphoma). Occasional patients have moderate thrombocytopenia (50,000 to 100,000 platelets/mm³) and no other findings and these patients usually carry the diagnosis of X-linked thrombocytopenia (XLT).

Tests Performed (check all that apply)

Indicate those tests/data used to establish the diagnosis by checking the indicated box []

Typical platelet count

At diagnosis (pre-splenectomy) age or date	Typical post-splenx age or date	Typical post BMT age or date	Most recent age or date
<10,000	[]	[]	[]
10-20,000	[]	[]	[]
20-50,000	[]	[]	[]
50-100,000	[]	[]	[]
100-150,000	[]	[]	[]
>150,000	[]	[]	[]
or actual count	(_____)	(_____)	(_____)

Platelet Size

pre-splenectomy (age or date)
post-splenectomy (age or date)
most recent (age or date)

Platelet volume

_____ femtoliters
_____ femtoliters
_____ femtoliters

Platelet diameter

_____ microns
_____ microns
_____ microns

Normal for lab
_____ fL _____ microns

Genetic Information

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

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

Yes [], No [], Not tested [],

Gene test used? _____

Cells tested?

B Cells Yes / No
T Cells Yes / No
Platelets Yes / No

Other

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Pedigree Analysis

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

Relation	WAS – alive	WAS – deceased	Tested Normal	Carrier	Not Tested	Unknown	Undiagnosed male with suggestive symptoms
Mother							
Father							

Information on Other Affected Kindred Members listed above (e.g., sibling, cousin, &/or maternal uncle):

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

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WAS Gene Mutation

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

Mutation analysis performed by _____

Nucleotides affected (e.g., 361C>T) _____

Predicted Amino Acid Change (e.g., W140R) _____

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

Mutation tested but not found [] which gene(s) _____

Publications (please give citation - if published) _____

WASP protein expressed? Yes _____, No _____, Not tested _____ FACS [] Western []

Were WASP revertants observed? Not studied [], flow cytometry [], Western Blot [], other _____

Present in:

T cells? yes [], no [], frequency % [], date _____

B cells? yes [], no [], frequency % [], date _____

Myeloid cells? yes [], no [], frequency % [], date _____

NK cells? yes [], no [], frequency % [], date _____

What molecular/genomic change was observed? _____

Clinical Score (please circle one)

- 1 Thrombocytopenia and small platelets without any other symptoms, clinical findings, or laboratory abnormalities.
- 2 Patients with platelet abnormalities and mild, transient eczema with or without minor infections
- 3 Patients with persistent but manageable eczema or recurrent infections or both
- 4 Patients with persistent and difficult-to-control eczema and frequent life-threatening infections
- 5 Classic WAS or XLT plus malignancy &/or significant autoimmunity

Additional Clinical Features of Special Note

Types of Bleeding Episodes Experienced

	Not Seen	Observed	Prominent	Unknown
Upper GI hemorrhage				
Lower GI hemorrhage / Rectal bleeding				
Intracranial hemorrhage				
Subdural hematoma				
Hemarthrosis				
Epistaxis				
Oral				
Hematuria				
Subcutaneous bleeding				

Other (specify)	Observed	Prominent in this patient

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Autoimmune / Inflammatory Disorders

	Not Seen	Observed	Prominent	Unknown
Autoimmune hemolytic anemia []		_____	_____	_____
ITP []		_____	_____	_____
Neutropenia []		_____	_____	_____
Inflammatory Bowel Disease []		_____	_____	_____
Sclerosing Cholangitis []		_____	_____	_____
Arthritis, chronic []		_____	_____	_____
Arthralgia []		_____	_____	_____
Vasculitis				
Skin []		_____	_____	_____
Coronary []		_____	_____	_____
Renal []		_____	_____	_____
Cerebral []		_____	_____	_____
GI []		_____	_____	_____
Pulmonary []		_____	_____	_____
Other (specify) _____		_____	_____	_____
Juvenile Rheumatoid Arthritis []		_____	_____	_____
Nephritis []		_____	_____	_____
Other (specify) _____				

Other Treatments/Procedures Used:

Cytokine Treatments

	Yes	No	Unknown	Age	Date (mm/dd/yyyy)	Response
Gamma-interferon						
G-CSF						
GM-CSF						
IL-2						
Other (list):						

Did this treatment lead to significant complications? Yes [], no [], unknown []

*Complications:

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Other treatments used

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

	Yes	No	Unknown	Age or Date Begun	Result
Transfer Factor					

	Yes	No	Unknown	Age or Date	Cells:	Vector	Outcome
Gene Therapy							
Publication?							

Other Treatments?	Yes	No	Unknown	Age begun	Date begun (mm/dd/yyyy)	Duration	Still needed?	
							Yes	No

Did any of these treatments lead to significant complications? Yes [], no [], unknown [], explain:

*Complications:

Memo Field/Additional comments:
