



## Wiskott-Aldrich Registry Data Collection Form

### Patient Identification:

Patient Name (first, middle, last) \_\_\_\_\_

Date of this Record (mm/dd/yyyy): \_\_\_\_/\_\_\_\_/\_\_\_\_

Patient's Number Assigned by Registry \_\_\_\_\_

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

Patient Initials \_\_\_\_/\_\_\_\_/\_\_\_\_      Date of Birth \_\_\_\_/\_\_\_\_/\_\_\_\_      Gender: male [  ], female [  ]  
(mm/dd/yyyy)

Home Address: \_\_\_\_\_

State: \_\_\_\_\_

Zip Code: \_\_\_\_\_

Phone: \_\_\_\_\_

Email: \_\_\_\_\_

State or Province of birth: \_\_\_\_\_

Country of birth: \_\_\_\_\_

Data transferred from earlier IDF Registry? Yes [  ] no [  ]

### Submitting Physician Information:

Name: \_\_\_\_\_

Address: \_\_\_\_\_

State: \_\_\_\_\_

Zip Code: \_\_\_\_\_

Phone: \_\_\_\_\_

Email: \_\_\_\_\_

Fax: \_\_\_\_\_

### Diagnostic Criteria

Definitive [  ]

Male patient with congenital thrombocytopenia (less than 70,000 platelets/mm<sup>3</sup>), 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/mm<sup>3</sup>), 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/mm<sup>3</sup>) 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/mm<sup>3</sup>) 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 <b>post-splenx</b> age or date	Typical <b>post BMT</b> 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? \_\_\_\_\_

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**Pedigree Analysis [ ]**

<b>Relation</b>	<b>Tested Normal</b>	<b>Carrier</b>	<b>Not Tested</b>	<b>WAS – alive</b>	<b>WAS – deceased</b>	<b>Unknown</b>	<b>Undiagnosed male with suggestive symptoms</b>
Mother							
Father							
Brothers (fill in # in each category)							
Sisters (fill in # in each category)							
Maternal Uncles (fill in # in each category)							
Maternal Cousins (fill in # in each category)							
Maternal Grandfather (mother's father)							
Other affected family members							

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

<b>Relation</b>	<b>Initials</b>	<b>DOB (mm/dd/yyyy)</b>	<b>Gender</b>



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**Autoimmune / Inflammatory Disorders**

	<b>Not Seen</b>	<b>Observed</b>	<b>Prominent in this patient</b>
Autoimmune hemolytic anemia	[ ]	_____	_____
ITP	[ ]	_____	_____
Neutropenia	[ ]	_____	_____
Inflammatory Bowel Disease	[ ]	_____	_____
Sclerosing Cholangitis	[ ]	_____	_____
Arthritis, chronic	[ ]	_____	_____
Arthralgia	[ ]	_____	_____
Vasculitis			
Skin	[ ]	_____	_____
Coronary	[ ]	_____	_____
Renal	[ ]	_____	_____
Cerebral	[ ]	_____	_____
Other (specify) _____		_____	_____
Juvenile Rheumatoid Arthritis	[ ]	_____	_____
Nephritis	[ ]	_____	_____
Other (specify) _____		_____	_____

**Other Treatments/Procedures Used:**

**Cytokines**

	Yes	No	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:

