Feinstein Institute for Medical Research
Northwell Health

DBAR
DIAMOND BLACKFAN ANEMIA REGISTRY

DIAMOND BLACKFAN ANEMIA REGISTRY (DBAR)
ENROLLMENT QUESTIONNAIRE

This information is confidential and for research purposes only.

<table>
<tr><td colspan="2">Patient's Last Name</td><td>First Name</td><td>Middle Name</td></tr>
<tr><td colspan="4">Parents' Full Names</td></tr>
<tr><td colspan="4">Address</td></tr>
<tr><td>City</td><td>State</td><td>Zip Code</td><td>Country</td></tr>
<tr><td>Date of Birth</td><td>Sex</td><td>Primary Phone</td><td>Secondary Phone</td></tr>
<tr><td colspan="2">E-mail Address</td><td colspan="2">Alternate E-mail Address</td></tr>
<tr><td colspan="2">Name of Primary Physician</td><td colspan="2" rowspan="2">Address of Primary Physician</td></tr>
<tr><td colspan="2">Phone Number/ Fax of Physician</td></tr>
<tr><td colspan="2">Name of Hematologist</td><td colspan="2" rowspan="4">Address of Hematologist</td></tr>
<tr><td colspan="2">Phone Number/ Fax of Hematologist</td></tr>
<tr><td colspan="2">Date Questionnaire Completed</td></tr>
<tr><td colspan="2">Form Completed by:
O Self O Parent O Physician/Nurse O Other</td></tr>
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Diagnosis of DBA

Age at presentation of anemia: ________ weeks /months /years	Age at diagnosis of DBA: ________ weeks /months /years

"Classic" DBA Diagnostic Criteria (check all that apply)
- O Age less than 1 year at presentation
- O Macrocytic anemia with no other significant cytopenias
- O Reticulocytopenia
- O Normal marrow cellularity with a paucity of erythroid precursors

Major Supporting Criteria
- O Gene mutation described in "classical" DBA
- O Positive family history

Minor Supporting Criteria
- O Elevated erythrocyte adenosine deaminase activity
- O Congenital anomalies described in "classical" DBA
- O Elevated HbF
- O No evidence of another inherited bone marrow failure syndrome

Birth History

Term: (check one) O Full Term O Premature O Postmature	Type of delivery: O Vaginal O C-Section	Gestational age: _______ weeks
Birth measurements: Weight______lbs______oz or_______kg Length_______in/cm Head Circumference_______in/cm		
Complications of this pregnancy: O Bleeding O Maternal anemia O Preeclampsia/Eclampsia O Intrauterine growth retardation		

Development

Overall physical development: O Normal O Delayed	**Puberty:** O Normal O Delayed
Overall speech development: O Normal O Delayed	**Age at Menstruation:** ______ yrs
Learning disabilities: O Absent O Present If present, please describe:	

Subject Initials ____ ____ ____ DBAR#___________

Physical Abnormalities (Please check None or all positives; also mark right, left, or both where applicable. Please elaborate any descriptions in the Comments section below.)

	Right	Left	Both	Description
O None				
O Low birthweight				
O Short stature				
Head and Face:				
O Head size (large, small)				
O Small jaw				
O Cleft palate				
O Cleft lip				
O Cleft lip and palate				
O Macroglossia (large tongue)				
O Flat nasal bridge				
O Abnormal ears				
O Decreased hearing				
O Abnormal eyes:				
O Hypertelorism (wide-spaced)				
O Epicanthal folds				
O Ptosis (droopy eyelids)				
O Strabismus				
O Congenital cataracts				
O Microphthalmia (small eyes)				
O Glaucoma				
Neck:				
O Short				
O Webbed				
O Sprengel deformity				
O Klippel-Feil anomaly				
Thumbs:				
O Triphalangeal				
O Duplicated or bifid				
O Subluxed				
O Hypoplastic				
Hands:				
O Flat thenar muscle				
O Absent radial pulse				
Feet:				
O Webbed toes				
Other Skeletal:				
O Hip dysplasia				

Subject Initials ____ ____ ____ DBAR#___________

	Right	Left	Both	Description
Kidneys:				
O Absent				
O Single				
O Dysplastic				
O Horseshoe				
O Duplicated ureter				
O Ectopic				
O Abnormal posterior valves with reflux				
Heart:				
O Ventricular septal defect (VSD)				
O Atrial septal defect (ASD)				
O Coarctation of the aorta				
O Patent foramen ovale (PFO)				
O Patent ductus arteriosus (PDA)				
O Other (fill in)				
O Asplenia				
Gonads:				
O Cryptorchidism				
O Microphallus (Small penis)				
O Small testes				

Comments:

Subject Initials ____ ____ ____ DBAR#___________

Ancestry (Please indicate the ancestral groups to which the parents belong.)

O Caucasian
- O Northern European (England, Scotland, Wales, Ireland, N.France, Holland, Belgium, Switzerland)
- O Scandinavian (Denmark, Norway, Sweden, Finland)
- O Southern European (Spain, Portugal, Italy, S.France)
- O Central European (Germany, Austria, Hungary)
- O Eastern European (Russia, Poland, Romania, Ukraine, Lithuania, Latvia, Estonia, Czech Republic)
- O East Mediterranean (Greece, Turkey, Croatia, Bosnia, Yugoslavia, Albania)
- O Northern Africa
- O Middle East
- O French Canada
- O South Africa
- O Ashkenazi Jewish
- O Sephardic Jewish
- O Jewish (Others)

O American Indian/Alaska Native
- O North America
- O South America (Includes Central America)

O Latino/Hispanic
- O Mexican
- O Central American
- O South American
- O Dominican
- O Puerto Rican
- O Cuban
- O West Indian

O Native Hawaiian/Pacific Islander
- O Hawaiian
- O Samoan
- O Guamanian (Guamanian or Chamorro)
- O Pacific Islands

O Black
- O African American
- O West Indian
- O African Heritage

O Asian
- O Chinese
- O Korean
- O Filipino
- O Pakistani
- O Vietnamese
- O Cambodian
- O Japanese
- O Malaysian
- O Thai
- O Indian

O Not Available (No Ethnicity data available)

Subject Initials ____ ____ ____ DBAR#___________

Family History (Please check all that apply. If pedigree is available, please enclose copy.)

Mother's Name	Maiden Name	Date of Birth
O Diamond Blackfan anemia O Anemia O Blood transfusions O Macrocytosis (large red cells) O Vitamin B12 deficiency	O Cleft palate/lip O Congenital heart disease O Thumb/hand anomaly O Other skeletal anomaly O Kidney anomaly	O Aplastic anemia (AA) O Cancer (specify type) ___________________________ O Leukemia ________________ Age at presentation of cancer/leukemia/AA__________
Mother's Height ______ft______in or__________cm	Number of: (please indicate 0 if none) Pregnancies_______Miscarriages________Stillbirths _______	
Complications with previous pregnancies: O Bleeding O Maternal anemia O Preeclampsia/Eclampsia O Intrauterine growth retardation		
Mother's Laboratory Date: (if available) Hemoglobin________gm/dl Hematocrit_______% MCV __________		
Father's Name	Date of Birth	Are parents related? O Yes O No
O Diamond Blackfan anemia O Anemia O Blood transfusions O Macrocytosis (large red cells) O Vitamin B12 deficiency	O Cleft palate/lip O Congenital heart disease O Thumb/hand anomaly O Other skeletal anomaly O Kidney anomaly	O Aplastic anemia (AA) O Cancer (specify type) ___________________________ O Leukemia ________________ Age at presentation of cancer/leukemia/AA__________
Father's Height ______ft______in or__________cm		
Father's Laboratory Date: (if available) Hemoglobin________gm/dl Hematocrit_______% MCV __________		

Siblings (Please list all full and half siblings of the patient. Indicate whether half siblings are maternal or paternal. Also include deceased siblings, stillbirths and abortions. Specify history of DBA-related illnesses.)

Relationship to patient	Sex	Date of Birth	History of: (check all that apply)	
			O Diamond Blackfan anemia O Anemia O Blood transfusions O Macrocytosis (large red cells) O Vitamin B12 deficiency O Aplastic anemia (AA) O Leukemia______________ (specify)	O Cleft palate/lip O Congenital heart disease O Thumb/hand anomaly O Other skeletal anomaly O Kidney anomaly O Cancer (specify type) ________________________ Age at presentation of cancer/leukemia/AA_______
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Other Family Members (Please list any family members with anemia, leukemia, cancer, and/or congenital anomalies. Indicate relationship to patient.)

Relationship to patient	Sex	Date of Birth	History of: (check all that apply)
			O Diamond Blackfan anemia O Anemia O Blood transfusions O Macrocytosis (large red cells) O Vitamin B12 deficiency O Aplastic anemia (AA) O Leukemia______________ (specify) O Cleft palate/lip O Congenital heart disease O Thumb/hand anomaly O Other skeletal anomaly O Kidney anomaly O Cancer (specify type) ______________________ Age at presentation of cancer/leukemia/AA_______
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Please attach more complete information on siblings and family members where applicable and available.

Psychosocial/Financial Issues (Please check all that apply.)

Psychological	Educational	Financial
O Depression O Anxiety O Autism O Asperger's syndrome O Attention deficit disorder O Attention deficit disorder with hyperactivity O Other Age at diagnosis ______________	O Educational level completed ______________ (specify) O Vocational training O Is employed	O Difficulty accessing medical care O Due to distance to travel O Due to financial constraints O Public/state health insurance O Private insurance O No Insurance

Subject Initials ____ ____ ____ DBAR#___________

Laboratory Findings of Patient (Please define units if different than those listed.)

	Birth or Initial*	At Diagnosis	Most Recent
Date of Lab Test			
Hemoglobin (g/dl)			
Hematocrit (%)			
WBC (x 1000)			
Diff: N/B/L/M/E/Bas**			
Platelet count			
Retic count (%)			
MCV (fl)			
MCH (pg)			
MCHC (g/dl)			
Red cell dist width			
Haptoglobin (mg/dl)			
Hb A2 (%)			
Hb F (%)			
Red cell Adenosine Deaminase Activity (eADA)			
Vitamin B12 (pg/ml)			
RBC Folate (ng/ml)			
Iron (mcg/dl)			
Total iron binding capacity (mcg/dl)			
Ferritin (ng/ml)			
Erythropoietin (iu/ml)			
IgG (mg/dl)			
IgA (mg/dl)			
IgM (mg/dl)			
Parvovirus Antibody			
Bone Marrow DNA for Parvovirus			
T2* (msec) [heart iron quantitation by MRI]			
Liver Iron Concentration (LIC; mg/g, dry weight) O Ferriscan O SQUID O Liver MRI			

*Please specify if results are from birth or, if not available, list first known blood results and indicate date done.

**Diff=differential: N/B/L/M/E/Bas = neutrophils/bands/lymphocytes/monocytes/eosinophils /basophils

Subject Initials ____ ____ ____ DBAR#___________

Bone Marrow Examination Results (Please send copies of bone marrow aspirate, bone marrow biopsy and chromosome analysis reports.)

Date Done	Aspirate/Biopsy Results	Chromosome Results
____________	O Cellularity ____________ O Red cell morphology O White cell morphology ____________ O Megakaryocyte morphology	O Chromosome analysis ____________ O FISH results O Chr 5 ____________ O Chr 7 ____________ O Chr 8 ____________ O Chr 9 ____________ O Other ____________
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Genetic Mutation Analysis (Please send copy of mutation analysis report.)

O *RPS7*
O *RPS10*
O *RPS17*
O *RPS19*
O *RPS24*
O *RPS26*
O *RPS15A*
O *RPS29*
O *RPS28*
O *RPS27*
O *RPS20*
O *RPL15*
O *RPL31*

O *RPL5*
O *RPL11*
O *RPL19*
O *RPL26*
O *RPL35a*
O *RPL17*
O *RPL18*
O *RPL27*
O *RPL35*
O *GATA1*

Specific mutation if known: ____________________

O Don't know
O Not tested
O Would like to be tested

Mutation analysis done at which lab?
(check all that apply)

O Boston Children's
O Johns Hopkins
O GeneDx
O Ambry Genetics
O Other (specify) ________________
O Don't know where done

Medications for Anemia (Please check drugs given and indicate start and stop dates, or if ongoing. Also indicate response of hemoglobin to treatment.)

	Date Started	Date Completed	Response
O Corticosteroids (Prednisone, Prednisolone)		O Stopped ________________ O Ongoing	O Response noted: O Hemoglobin 8-10 gm/dl O Hemoglobin >10 gm/dl O No response noted
O Dexamethasone (Decadron)		O Stopped ________________ O Ongoing	O Response noted: O Hemoglobin 8-10 gm/dl O Hemoglobin >10 gm/dl O No response noted
O Cyclosporine A (CSA)		O Stopped ________________ O Ongoing	O Response noted: O Hemoglobin 8-10 gm/dl O Hemoglobin >10 gm/dl O No response noted
O Erythropoietin (EPO, Epogen, or Procrit or Aranesp)		O Stopped ________________ O Ongoing	O Response noted: O Hemoglobin 8-10 gm/dl O Hemoglobin >10 gm/dl O No response noted
O Anti-thymocyte Globulin (ATG)		O Stopped ________________ O Ongoing	O Response noted: O Hemoglobin 8-10 gm/dl O Hemoglobin >10 gm/dl O No response noted
O Other (please specify)		O Stopped ________________ O Ongoing	O Response noted: O Hemoglobin 8-10 gm/dl O Hemoglobin >10 gm/dl O No response noted

Red Cell Transfusions and Medications for Iron Overload (Chelation Therapy)

	Date Started	Date Completed	Comments
O Red Bloood Cell Transfusions		O Ongoing	O Frequency O every 3 weeks O every 4 weeks O other (specify) _____________
O Desferioxamine (Desferal)		O Ongoing	
O Deferasirox (Exjade)		O Ongoing	
O Deferiprone		O Ongoing	

Other Medications

	Date Started	Date Completed	Comments
O Co-trimoxazole (Bactrim, Septra, or TMP-SMZ)		O Ongoing	
O Insulin		O Ongoing	
O Growth hormone		O Ongoing	
O Thyroid hormone		O Ongoing	

O Birth Control medication (specify)		O Ongoing	
O Testosterone		O Ongoing	
O IV/SC Immunoglobulin (IgG)		O Ongoing	
O Vitamins/ amino acids and/or herbal supplements (specify)__________________		O Ongoing	
O Other (specify)		O Ongoing	
O Other (specify)		O Ongoing	

Other Treatments

	Date of Surgery or Procedure	Comments
O Splenectomy		
O Stem cell/Bone marrow transplant		

Reason
O DBA diagnosis
O Transfusion dependence
O Pancytopenia/Aplastic anemia
(low white cells and low platelets as well)
O Leukemia/Lymphoma

Donor
O Sibling donor
O Other related donor
O Unrelated donor

Source
O Bone marrow
O Cord blood
O Peripheral blood

Degree of match
O 10/10 O 6/6
O 9/10 O 5/6
O 8/10 O 4/6

Complications
O Graft vs host disease
O Veno-occlusive disease of the liver

Remission (defined as 6 months or longer without medications or transfusions for anemia)

O At present
Start date_______________
Age at remission__________

O In the past
Date _________________
Duration______________
O Never been in remission

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Current Status of Patient

O Steroid dependent
O Chronic transfusion dependent
O Remission
O Status post Stem cell/Bone marrow transplant
O Aplastic anemia
O Leukemia (specify)
____
O Cancer (specify)
____________ __________________
O Myelodysplastic syndrome
O Other ___________________________
(specify)

Death of Patient

Date of Death ___________
Cause of Death
O Iron overload
O Stem cell/Bone marrow transplant complication
Aplastic Anemia
O Leukemia (specify) _____
O Cancer (specify) _
O Myelodysplastic syndrome
O Infection (specify) ___________________
O Other (specify) _____________________
O Unknown

Comments

____
____
____
____
____
____
____
____

Please return completed form to:

Adrianna Vlachos, MD
The Feinstein Institute for Medical Research
350 Community Dr.
Manhasset, NY 11030

Tel: 516-562-1504
Fax: 516-562-1599
E-mail: avlachos@northwell.edu

For questions or assistance in completing this questionnaire, please contact:

Eva Atsidaftos, MA
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Manhasset, NY 11030

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