 

**DIAMOND BLACKFAN ANEMIA REGISTRY (DBAR)**

**ENROLLMENT QUESTIONNAIRE**

This information is confidential and for research purposes only.

|  |  |  |  |
| --- | --- | --- | --- |
| Patient’s Last Name | | First Name | Middle Name |
| Parents’ Full Names | | | |
| Address | | | |
| City | State | Zip Code | Country |
| Date of Birth | Sex | Primary Phone | Secondary Phone |
| E-mail Address | | Alternate E-mail Address | |
| Name of Primary Physician | | Address of Primary Physician | |
| Phone Number/ Fax of Physician | |
| Name of Hematologist | | Address of Hematologist | |
| Phone Number/ Fax of Hematologist | |
| Date Questionnaire Completed | |
| Form Completed by:  O Self O Parent O Physician/Nurse O Other | |

# 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: | |

**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 |  |  |  |  |

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
|  | 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:

|  |
| --- |
|  |
|  |
|  |
|  |
|  |
|  |

**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) | |

**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 Cleft palate/lip  O Anemia O Congenital heart disease  O Blood transfusions O Thumb/hand anomaly O Macrocytosis (large red O Other skeletal anomaly cells) O Kidney anomaly  O Vitamin B12 deficiency O Cancer (specify type)  O Aplastic anemia (AA) O Leukemia Age at presentation of  (specify) cancer/leukemia/AA |
|  |  |  | O Diamond Blackfan anemia O Cleft palate/lip  O Anemia O Congenital heart disease  O Blood transfusions O Thumb/hand anomaly O Macrocytosis (large red O Other skeletal anomaly cells) O Kidney anomaly  O Vitamin B12 deficiency O Cancer (specify type)  O Aplastic anemia (AA) O Leukemia Age at presentation of  (specify) cancer/leukemia/AA |

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
|  |  |  | 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 |
|  |  |  | 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 |
|  |  |  | 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 |

**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 Cleft palate/lip  O Anemia O Congenital heart disease  O Blood transfusions O Thumb/hand anomaly O Macrocytosis (large red O Other skeletal anomaly cells) O Kidney anomaly  O Vitamin B12 deficiency O Cancer (specify type)  O Aplastic anemia (AA) O Leukemia Age at presentation of  (specify) cancer/leukemia/AA |
|  |  |  | O Diamond Blackfan anemia O Cleft palate/lip  O Anemia O Congenital heart disease  O Blood transfusions O Thumb/hand anomaly O Macrocytosis (large red O Other skeletal anomaly cells) O Kidney anomaly  O Vitamin B12 deficiency O Cancer (specify type)  O Aplastic anemia (AA) O Leukemia Age at presentation of  (specify) cancer/leukemia/AA |

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
|  |  |  | 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 |
|  |  |  | 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 |
|  |  |  | 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 |

Please attach more complete information on siblings and family members where applicable and available.

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

|  |  |  |
| --- | --- | --- |
| Psychological 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 | Educational  O Educational level completed  (specify)  O Vocational training O Is employed | Financial  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 |

**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

**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 Chromosome analysis  O FISH results  O Chr 5 O Chr 7 O Chr 8 O Chr 9  O Other |
| O Red cell morphology |
| O White cell morphology |
| O Megakaryocyte morphology |
| \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ | O Cellularity | O Chromosome analysis |
| O Red cell morphology  O White cell morphology  O Megakaryocyte morphology | O FISH results  O Chr 5 O Chr 7 O Chr 8 O Chr 9  O Other |
| \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ | O Cellularity | O Chromosome analysis |
| O Red cell morphology  O White cell morphology  O Megakaryocyte morphology | O FISH results  O Chr 5 O Chr 7 O Chr 8 O Chr 9  O Other |

**Genetic Mutation Analysis** (Please send copy of mutation analysis report.)

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

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

**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 | Source | |
| O DBA diagnosis | O Bone marrow | |
| O Transfusion dependence | O Cord blood | |
| O Pancytopenia/Aplastic anemia | O Peripheral blood | |
| (low white cells and low platelets as well) |  | |
| O Leukemia/Lymphoma | Degree of match | |
|  | O 10/10 | O 6/6 |
| Donor | O 9/10 | O 5/6 |
| O Sibling donor | O 8/10 | O 4/6 |
| O Other related donor |  | |
| O Unrelated donor | 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 O In the past  Start date Date  Age at remission Duration O Never been in remission |

**Current Status of Patient 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

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)

**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**

**DBA Clinical Research Coordinator**

**The Feinstein Institute for Medical Research 350 Community Dr.**

**Manhasset, NY 11030**

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