 

**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 CriteriaO Gene mutation described in ‘‘classical’’ DBA O Positive family historyMinor Supporting CriteriaO Elevated erythrocyte adenosine deaminase activity O Congenital anomalies described in ‘‘classical’’ DBA O Elevated HbFO 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 CaucasianO 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 AfricaO Middle EastO French Canada O South AfricaO Ashkenazi Jewish O Sephardic Jewish O Jewish (Others) |
| O American Indian/Alaska Native O North AmericaO South America (Includes Central America) |
| O | Latino/HispanicO MexicanO Central American O South American O DominicanO Puerto Rican O CubanO West Indian |
| O Native Hawaiian/Pacific Islander O HawaiianO SamoanO Guamanian (Guamanian or Chamorro) O Pacific Islands |
| O | BlackO African American O West IndianO African Heritage |
| O | AsianO Chinese O Korean O Filipino O PakistaniO Vietnamese O Cambodian O Japanese O Malaysian O ThaiO 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 AnemiaO Blood transfusionsO Macrocytosis (large red cells) O Vitamin B12 deficiency | O Cleft palate/lipO Congenital heart disease O Thumb/hand anomalyO Other skeletal anomaly O Kidney anomaly | O Aplastic anemia (AA) O Cancer (specify type) O Leukemia Age at presentation ofcancer/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 AnemiaO Blood transfusionsO Macrocytosis (large red cells) O Vitamin B12 deficiency | O Cleft palate/lipO Congenital heart disease O Thumb/hand anomalyO Other skeletal anomaly O Kidney anomaly | O Aplastic anemia (AA) O Cancer (specify type) O Leukemia Age at presentation ofcancer/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/lipO Anemia O Congenital heart diseaseO Blood transfusions O Thumb/hand anomaly O Macrocytosis (large red O Other skeletal anomaly cells) O Kidney anomalyO Vitamin B12 deficiency O Cancer (specify type)O Aplastic anemia (AA) O Leukemia Age at presentation of(specify) 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 Cleft palate/lipO Anemia O Congenital heart diseaseO Blood transfusions O Thumb/hand anomaly O Macrocytosis (large red O Other skeletal anomaly cells) O Kidney anomalyO Vitamin B12 deficiency O Cancer (specify type)O Aplastic anemia (AA) O Leukemia Age at presentation of(specify) cancer/leukemia/AA  |
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|  |  |  |  |  |
| --- | --- | --- | --- | --- |
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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 O Depression O AnxietyO AutismO Asperger’s syndromeO Attention deficit disorderO Attention deficit disorder with hyperactivityO OtherAge at diagnosis  | EducationalO Educational level completed (specify)O Vocational training O Is employed | FinancialO Difficulty accessing medical care O Due to distance to travelO Due to financial constraintsO Public/state health insurance O Private insuranceO 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 ironquantitation by MRI ] |  |  |  |
| Liver Iron Concentration (LIC; mg/g, dry weight)O Ferriscan O SQUIDO 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 analysisO FISH resultsO 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 morphologyO White cell morphologyO Megakaryocyte morphology | O FISH resultsO 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 morphologyO Megakaryocyte morphology | O FISH resultsO 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/dlO No response noted |
| O Dexamethasone (Decadron) |  | O Stopped O Ongoing | O Response noted:O Hemoglobin 8-10 gm/dl O Hemoglobin >10 gm/dlO No response noted |
| O Cyclosporine A (CSA) |  | O Stopped O Ongoing | O Response noted:O Hemoglobin 8-10 gm/dl O Hemoglobin >10 gm/dlO 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/dlO 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/dlO No response noted |
| O Other (please specify) |  | O Stopped O Ongoing | O Response noted:O Hemoglobin 8-10 gm/dl O Hemoglobin >10 gm/dlO 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 | FrequencyO every 3 weeks O every 4 weeksO 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 pastStart 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**

**Tel: 516-562-1504**

**Fax: 516-562-1599**

**E-mail: eatsidaf@northwell.edu**