Caring for the Adult Patient

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Changing Paradigm in DBA?

Childhood Disorder → Disorder of Young Adults

Supportive Care
Transfusion Therapy
Steroid Therapy
Stem Cell Transplantation

J. Liu, 2005

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Evolution of Care in Diamond Blackfan Anemia

bjh review

Diagnosing and treating Diamond Blackfan anaemia: results of an international clinical consensus conference

Adrianna Vlachos,[1,2] Sarah Ball,[3] Niklas Dahl,[4] Blanche P. Alter,[5] Sujit Sheth,[6] Ugo Ramenghi,[7] Joerg Meerpohl,[8] Stefan Karlsson,[9] Johnson M. Liu,[1,2] Thierry Leblanc,[10] Carole Paley,[11] Elizabeth M. Kang,[12] Eva Judmann Leder,[1] Eva Atsidaftos,[2] Akiko Shimamura,[13] Monica Bessler,[14] Bertil Glader[15] and Jeffrey M. Lipton,[1,2] on behalf of the participants of the Sixth Annual Daniella Maria Arturi International Consensus Conference

Vlachos A et al, Brit J Haematol, 2008

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Agenda

1. How DBA is inherited and how to get genetic testing done
2. How to screen for iron overload and treat it
3. What is involved in SCT for DBA
4. How and when to screen for cancer and MDS
5. What is needed if you are a DBA patient and are pregnant
6. What is needed to do IVF/PGD
7. What endocrine complications are seen and how to screen for them
8. What cardiac problems are possible and how to screen for them
9. How to care for the adult DBA patient in general

What Are the Impediments?

Background: A major impediment to effective life-long care of patients with the inherited bone marrow failure syndrome, Diamond Blackfan anemia (DBA), is the transition from pediatric to adult care.

There are many historical reasons for this, including structural deficiencies in health care delivery for a rare and complex disorder and lack of resources and institutional commitment.

Major Impediment in Transition

Pediatric Hematologist ⟶
Adult Hematologist (Benign Specialist)

"….finding an appropriate adult provider with expertise in SCD was frequently identified as the most significant transition issue…."

Transition from pediatric to adult care for sickle cell disease: Results of a survey of pediatric providers. Sobota, Neufeld, Sprinz, and Heeney, Am J Hematol, 2011

What Do We Hope to Achieve?

Objectives: We have attempted to create a new model of transition from pediatric to adult care of DBA patients, emphasizing a team-based approach with active participation and communication from multiple subspecialty physicians from the Departments of Pediatrics and Medicine.

We hypothesize that the integration of an adult provider into the pediatric paradigm is essential for successful transition.

What is Our Ongoing Plan of Action?

Methods: As part of the ongoing surveillance program to follow the natural history of DBA patients enrolled in the North American Diamond Blackfan Anemia Registry (DBAR), we have attempted to formalize the transition of patients over the age of 18 by having them evaluated by an adult hematologist (embedded in the pediatric program) with expertise in bone marrow failure syndromes.

DBA Clinical Care Team Physicians

Johnson Liu, Professor, Departments of Pediatrics and Medicine
Adrianna Vlachos, Head of Bone Marrow Failure Program, Division of Pediatric Hematology/Oncology
Jeffrey Lipton, Chief, Division of Pediatric Hematology/Oncology
Sandeep Jauhar, Director Heart Failure Center, Division of Cardiology, Department of Medicine
Yael Harris, Division of Endocrinology, Department of Medicine

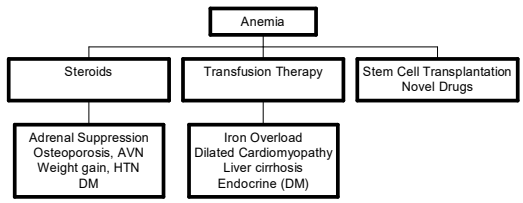
Adult-Intrinsic Management Issues

Post-adolescence and hormonal changes
Maturation and aging
Concomitant chronic diseases (atherosclerosis, chronic obstructive lung disease, etc.)
Sexually transmitted diseases
Lifestyle diseases (alcohol abuse, smoking, illicit drugs)
Fertility and pregnancy
Psychosocial and financial concerns

Adult DBA Patient: Typical Scenarios

General considerations
Iron overload complications and management
Steroid therapy complications and osteoporosis
Cancer surveillance
Complications of pregnancy in DBA
Transition from pediatric to adult care (internal medicine)

Current Therapies: Scylla and Charybdis



What Have We Found?

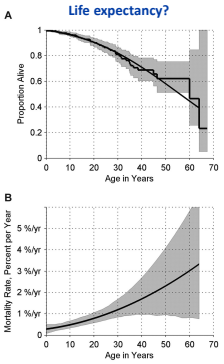
Patients with identified medical problems were referred as needed to adult cardiologists, endocrinologists, and orthopedic surgeons. Several patients required hospital admission (adult service) for catheter-associated infections or intensive iron chelation.

DBA guidelines working meeting, Atlanta March 10, 2018

Very little knowledge

Very few papers dedicated to DBA adult patients

Life expectancy?



(Vlachos et al, Blood)

DBA guidelines working meeting, Atlanta March 10, 2018

Anemia & blood support	Immunodeficiency
Iron overload management - Hemochromatosis - Side effects of iron chelators	Cancer risk
Other tranfusions side-effects	Reproductive choices Pregnancies
Anemia and steroids	New phenotypes
Hematological follow-up	Psychosocial issues

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DBA guidelines working meeting Atlanta, March 10, 2018

Guidelines to be commented & expanded

Agreement of « poor level » of any evidence-based approach…
- *we have to use available consensus made for patients with a different disease but common problems (as B-cell ID in pts with CVD, or osteoporosis in pts on steroids)*

As for other topics, guidelines in adult patients must lead to cooperative studies in order to be able to improve them

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Thank You

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