

# DIAMOND BLACKFAN ANEMIA AFRICA

## ***AN AFRICAN REGISTRY MAKING STRIDES FOR DBA***

*Dr Colin Noel    RareX 2018*



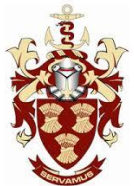
# DIAMOND BLACKFAN ANEMIA (DBA)

- ▶ Rare congenital red blood cell aplasia
  - ▶ Failed erythropoiesis
  - ▶ Congenital abnormalities
  - ▶ Cancer predisposition
- ▶ Incidence (1:100 000-200 000 live births)
- ▶ 90% Dx <1yo
- ▶ >50% Haploinsufficiency of ribosomal proteins
  - ▶ GATA-1, TSR2
- ▶ 45% Familial - Autosomal dominant
- ▶ Complex MDT approach to management



# SOUTH AFRICA / AFRICA

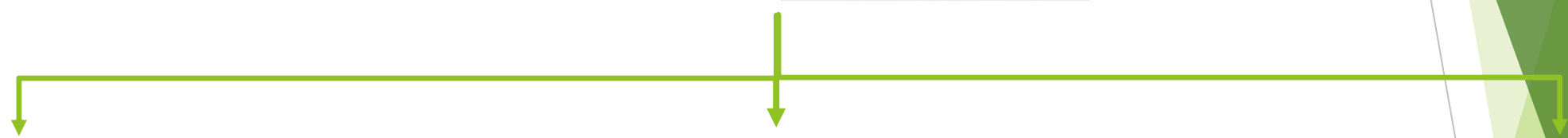
UNKNOWN



OCF TRANSFUSION CLINIC

OCF PATIENT SUPPORT





**DOCTORS PORTAL**

**DIAMOND BLACKFAN ANEMIA  
AFRICA REGISTRY (DBAAR)**

**OCF RESEARCH ARM**



# DOCTORS PORTAL

- ▶ Online Up-To-Date Platform
  - ▶ DIAGNOSIS
  - ▶ TREATMENT
  - ▶ ONGOING MONITORING GUIDELINES





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[WHAT IS DBA?](#)

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[GENETICS OF DBA](#)

[DBA AFRICA REGISTRY](#)



DIAMOND BLACKFAN ANEMIA  
AFRICA

# DOCTORS PORTAL

- ▶ Online Up-To-Date Platform

- ▶ DIAGNOSIS
- ▶ TREATMENT

- ▶ **Contact portal for advice from a MDT with invested interest**

- ▶ Support

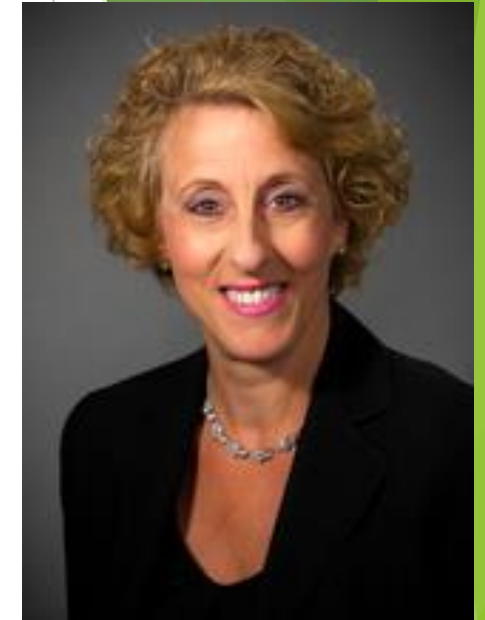
- ▶ Dr Hedi Van Der Watt
- ▶ Dr Almero Du Pisani
- ▶ Prof Vernon Louw
- ▶ Dr Jan Du Plessis





# DIAMOND BLACKFAN ANEMIA AFRICA REGISTRY (DBAAR)

- ▶ University of Cape Town, Groote Schuur Hospital, Department of Haematology
- ▶ Investigators:
  - ▶ PI - Dr C.B. Noel
  - ▶ CI - Prof V. Louw, Dr K. Noel, Dr H van der Watt
- ▶ Collaborating group - DBAR
- ▶ RedCap Database



# DIAMOND BLACKFAN ANEMIA AFRICA REGISTRY (DBAAR)

- ▶ Identify families with DBA → Support
- ▶ Identify current care → Advocate standard of care
- ▶ Describe DBA in Africa → Resource allocation
- ▶ Impact of DBA in Africa
- ▶ Contribute and collaborate in research
- ▶ Advocate for funding, research grants, treatment facilities for DBA patients



The Online Designer will allow you to make project modifications to fields and data collection instruments very easily using only your web browser. NOTE: While in development status, all field changes will take effect immediately in real time.

**Data Collection Instruments**

[Survey Queue](#) [Survey Login](#)

[Survey Notifications](#)

**Add new instrument:**

[+ Create](#) a new instrument from scratch

[↓ Import](#) a new instrument from the official [REDCap Shared Library](#)

[↑ Upload](#) instrument ZIP file from another project/user or [external libraries](#)

Instrument name	Fields	View PDF	Enabled as survey	Instrument actions	Survey-related options
Basic Demography	21		<a href="#">Enable</a>	<a href="#">Choose action</a>	
Birth History	18		<a href="#">Enable</a>	<a href="#">Choose action</a>	
Medical History at Diagnosis	65		<a href="#">Enable</a>	<a href="#">Choose action</a>	
Family History	20		<a href="#">Enable</a>	<a href="#">Choose action</a>	
Blood Results	22		<a href="#">Enable</a>	<a href="#">Choose action</a>	
Peripheral Blood Chromosomal Analysis	7		<a href="#">Enable</a>	<a href="#">Choose action</a>	
Bone Marrow Chromosomal analysis	7		<a href="#">Enable</a>	<a href="#">Choose action</a>	
Bone Marrow Aspirate/Biopsy	5		<a href="#">Enable</a>	<a href="#">Choose action</a>	
Medication History	15		<a href="#">Enable</a>	<a href="#">Choose action</a>	
Blood Transfusions	12		<a href="#">Enable</a>	<a href="#">Choose action</a>	
Chelation therapy	9		<a href="#">Enable</a>	<a href="#">Choose action</a>	
Bone Marrow Transplant	18		<a href="#">Enable</a>	<a href="#">Choose action</a>	
Spontaneous remission	3		<a href="#">Enable</a>	<a href="#">Choose action</a>	
Current Status	2		<a href="#">Enable</a>	<a href="#">Choose action</a>	

**Data Collection** [Edit instruments](#)

- Manage Survey Participants**  
- Get a public survey link or build a participant list for inviting respondents
  - Record Status Dashboard**  
- View data collection status of all records
  - Add / Edit Records**  
- Create new records or edit/view existing ones
- Show data collection instruments ▾

**Applications**

- Calendar
- Data Exports, Reports, and Stats
- Data Import Tool
- Data Comparison Tool
- Logging
- Field Comment Log
- File Repository
- User Rights and DAGs
- Data Quality
- REDCap Mobile App
- External Modules

**Help & Information**

- Help & FAQ
- Video Tutorials
- Suggest a New Feature

[Contact REDCap administrator](#)

# O<sub>2</sub>CF RESEARCH ARM

## ▶ Gene Therapy

▶ Collaboration with GSH, Department of Virology, UCT.

▶ Dr Walter Nevondo

▶ Lentiviral Vectors in RPL35A gene therapy

## ▶ Chelation Therapy

▶ Collaboration with GSH, Department of Hematology, UCT.

▶ Prof Vernon Louw



# THANK YOU

## HOW TO CONTACT US

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# THANK YOU

