NORTH CENTRAL REGIONAL HEALTH AUTHORITY

HEAD OFFICE: Building 39, Eric Williams Medical Sciences Complex
Uriah Butler Highway, Champs Fleurs
PABX: 868-645-2640-9 or 645-HOPE (4673)

March 26th 2019

MEDICAL REPORT

Joanna Brathwaite
DOB: 19/07/2012
REG: 757468
CONTACT: 791-7837/ 292-4501
DIAGNOSIS: Beta Thalassaemia Major
On regular red blood cell transfusions

Joanna is a 6 year old girl with Bata Thalassaemia Major. Her diagnosis was confirmed via molecular genetics in November 2013. She initially presented on 14/08/2013, to the Paediatric Emergency at Mt. Hope, Trinidad, with a cold. She was noted as lethargic, pale and had abdominal distension secondary to splenomegaly. Her Hb was 5.6g/dl and she was subsequently transfused with leucocyte poor packed red blood cells.

HbEP results were obtained as below:

Joanna	Hb A 3.3%	Hb F 94.6%	Hb A_2 2.1%
Mother	Hb A 92.7%	Hb F 1.8%	Hb A_2 5.5%
Father	Hb A 93.6%	Hb F 1.1%	Hb A_2 5.3%

Molecular genetics on the family were reported as:

Joanna Brathwaite:
Identification of the **G>A mutation at nucleotide 5 of IVS-I** (or nt 147 G>A):
[HBB:c.92+5G>A]
Identification of the **A>G mutation at nucleotide 849 of IVS-II** (or nt 1344 A>G):
[HBB:c.316-2A>G]
Conclusion: Joanna Brathwaite is a compound heterozygote for β^{+}(severe)/ β^{0}thalassaemia

Mother: Coletta Brathwaite
Identification of the **A>G mutation at nucleotide 849 of IVS-II** (or nt 1344 A>G):
[HBB:c.316-2A>G]

Father: Jason Brathwaite
Identification of the **G>A mutation at nucleotide 5 of IVS-I** (or nt 147 G>A):
[HBB:c.92+5G>A]

HLA typing for Joanna, her sibling Cayla, her mother and father are attached as hard copies.

TFT's (9/11/18)

TSH	1.74mIU/L	(0.465 - 4.68)
Free T_3	5.34Pmol/L	(4.26 - 10.0)
Free T4	16.5Pmol/L	(10.0 - 28.2)

HbA_1C 6.6% (19/12/18)
PTH pending
FSH 1.07
LH <0.216

Echocardiogram (21/2/18): Normal study

Vaccinations:
DTP/Polio x5
MMR x2
Hepatitis B x3
Yellow fever x1
PCV 10 x4
HIB x3

Growth/Nutrition and Development
There are no developmental concerns. She has a healthy appetite and her growth has been satisfactory.

Family History
The family consists of a 8 year old sister and her parents. Her family has HLA typing done under the guidance of Dr. Weaveney Charles (Adult Haematologist). None of the results was a complete match but many foreign centres are capable of doing Haploidentical Bone Marrow Transplants with less than perfect matches.

Physical Features:
Her weight was 22.5kg and height 115.7cm on 01/02/2019 corresponding to 75^{th} – 90th and 50^{th} – 75^{th} centile respectively. She has no maxillary or frontal bossing. There was no splenomegaly nor hepatomegaly. The cardiovascular, respiratory and neurological systems were unremarkable. She has a 2/6 ejection systolic murmur on the left sternal border consistent with a flow murmur.

In summary, Joanna has βThalassaemia Major and relies on monthly blood transfusions along with daily iron chelation therapy. These are mandatory for her health and survival. A bone marrow transplant (BMT) is the only curative option available for her survival without disease.

Her family has expressed the desire to pursue a BMT as it is the only curative treatment for Beta Thalassaemia Major. They have identified The Bambino Genu Paediatric hospital in Rome, as a Centre for a Haploidendical Bone Marrow Transplant. This centre has favourable results and it is important to choose a centre with positive results for this type of BMT.

Joanna has no organ disease and as a result of iron overload and no evidence of infection therefore now is the perfect time to pursue the BMT before complications set in. It has always been a challenge in Trinidad to ensure a regular supply of blood and Exjade (Deferasirox) for chelation.

As she gets older, complications such as: cardiac failure, liver disease, endocrine organ disease, osteoporosis, infections and dental malocclusions can develop as a consequence of iron overload and the disease itself. Therefore BMT centres prefer to perform the transplants on younger, healthy patients.

Kindly consider this child for Haploidentical Bone Marrow Transplant.

Sincerely,

Dr. Camille Greene MBBS, MRCPCH
Paediatric Consultant
Haematology Clinic
E.W.M.S.C

Joanna was started on regular monthly leucocyte depleted red blood cell transfusions around May 2014 and has had no transfusion reactions. She is O Rhesus Positive. Her pre-transfusion Hb ranges from 9-10gldl. Transfusion volume ≅ 15mg/kg each month. Her last transfusion was 27th March 2019.

Medication:
Deferasirox 500mg po od
Folic acid 2.5mg po od

Blood Investigations:
Blood Group: O Positive
Ferritin: 1180ng/ml (9/11/2018)

U+E's (9/11/2018)

BUN	16
Cr	0.41
Na	142
K	4.6
CI	104
Ca	9.1
Phos	5.3

LFT's (09/11/2018)

Total bili	1.6
AST	29
ALT	10
Total protein	6.6
Albumin	4.6

PTT	33.9
Control	30.1
PT	13.1
Control	11.6

Hepatitis C – Negative (15/12/2017)
Hepatitis B surface Ag – non-reactive (15/12/2017)
HIV (non-reactive) (15/12/2017)

CBC (06/03/2019) pre transfusion
Wbc $9.27x10^3$/uL
Hb 8.4g/dl
Hct 24.8%
Plt $627x10^3$/uL
Neut $4.29x10^3$/uL
MCV 74.3fL
MCHC 34.1g/dl
Retic 1.46%