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## Primary pure red cell aplasia association with Johnson-blizzard syndrome cured by homeopathy

**Ghulam Yaseen**

### Abstract

Primary Pure red cell aplasia is a syndrome due to failure of erythropoiesis, normocytic anemia with severe reticulocytopenia due to the marked reduction of erythroid precursors in the bone marrow. Johnson-Blizzard syndrome is rare, autosomal recessive or autosomal dominant, multisystem congenital disorder having abnormal development of pancreas, nose and scalp with mental retardation, hearing loss and growth failure. A case reported was a 3 months old male infant having prediagnosed primary pure red cell aplasia and Johnson-Blizzard syndrome with marked pancreatic insufficiency. He presented with profuse greenish-bulky stools and severe pallor complexion. Homeopathic medicines *Calcarea phos* and *Podophyllum* was given in 3x potencies according to the sign and symptoms. Frequency of stool reduced within a month and his haemoglobin level also increased with passage of time. Medicine continued for almost 10 months and follow-up visits showed no recurrence of pure red cell aplasia. Homeopathic remedy *Calcarea phos* 3x is highly effective to increase the haemoglobin level and *Podophyllum* 3x is helpful to manage profuse stools.

**Keywords:** Pure red cell aplasia, johanson-blizzard syndrome, pancreatic insufficiency, homeopathy

### 1. Introduction

Pure red cell aplasia is a syndrome due to failure of erythropoiesis, normocytic anemia with severe reticulocytopenia with marked reduction of erythroid precursors in the bone marrow [1]. Congenital PRCA and Diamond-Blackfan Anemia have resemblance in features as the maturation and production of RBCs arrests in both diseases [2]. The acquired form of PRCA can be primary or secondary to any other disease [3]. It is usually diagnosed in the first two years of life, but in severe conditions, it may be diagnosed in the first 2 to 3 months of life. Half of the children with pure red cell aplasia show sign and symptoms of mental retardation or malformation [4]. Johanson-Blizzard Syndrome is rare and fatal autosomal recessive or autosomal dominant, multisystem disorder distinguished by exocrine pancreatic insufficiency, hypothyroidism, growth failure, deafness, congenital aplasia of alae nasi, different level of mental retardation, and alopecia with wide open fontanelles [5].

Case reported narrates the cure of pure red cell aplasia in association to Johnson Blizzard Syndrome with dominance of pancreatic insufficiency. Treatment was purely based on homeopathic medicine.

### 2. Clinical presentation and investigation

There is no specific clinical presentation of congenital PRCA. Signs and symptoms are associated with anemia like fatigue, lethargy, and abnormal paleness of the skin. PRCA under-production anemia and continues decrease in production of haemoglobin lay to the adaptation of symptoms of anemia. Diagnosis of PRCA can be made by measuring the peripheral blood count and by bone marrow examination on the presence of hypoplastic and normoblastic erythroids. Further more eADA and fetal haemoglobin estimation is also important.

Johnson-Blizzard syndrome sometimes described as ectodermal dysplasia. It is profound multisystem development errors and exocrine pancreatic insufficiency, as an inherited disease of pancreas. Pancreatic insufficiency is an inability of the exocrine pancreas to secrete digestive enzymes for the breakdown of food particles. Signs and symptoms of pancreatic insufficiency include malabsorption, malnutrition, vitamin deficiencies, abdominal cramps, weight loss, and steatorrhea. Presence of elastase enzyme in the stool and fecal fat is the first sign of insufficiency.

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### 3. Role of homeopathy in cure

A number of homeopathic remedies are in use for the treatment of anemia, primary or secondary to any disease. Homeopathic preparations have the ability to revive health condition by treating the disease through the signs and symptoms of patients.

#### 3.1 Case history

A 3-months old boy, presented in the homeopathic opd with following complaints

- Five to eight numbers of greenish-bulky stools in a day from birth with continues weight loss. Initially, he was on breastfeeding but after few days of birth, he was shifted to formula milk.
- Progressive paler skin from a month with general laziness
- Frequent wash-ups with cold water after every motion caused discomfort and weeping tendency

#### 3.2 History of presenting complaints

He was under treatment in Children Hospital Lahore from 2.5 months, where doctors diagnosed that he has Johanson Blizzard syndrome due to typical clinical features as Syndromic appearance- aplastic alae nasi that gave him a beak-shaped nose, low set ears, triangular upper lip with flat philtrum, a number of Café au lait spots on his trunk, upper and lower limb.. Laboratory evaluation showed that his haemoglobin level was 5.4g/dl (Figure 1). Bone marrow aspiration revealed erythroid hypoplasia with normal myeloid and megakaryocytes series. A diagnosis of primary pure red cell aplasia was made. (Figure 2)

Serum folic acid, B<sub>12</sub>, erythrocyte adenosine deaminase (ADA) could not be performed due to non-availability of the facility at that time.

His echocardiography revealed dextrocardia with situs inversus and mild Av valve regurgitation (Figure 7). Audiometry showed that he has no residual hearing bilaterally. (Figure 8). His birth weight was 2.8 kg. During his admission in hospital and after investigation his weight reduced to 1.7 kg because doctors didn't allow mother feed or gave any supportive treatment. After diagnosis, corticosteroids administered but he didn't show any positive signs and symptoms of recovery, this case shifted to homeopathy as an alternative treatment.

#### 3.3 Family history

His parents were non-consanguineous and completely healthy persons.

#### 3.4 Birth history

He was born on term and during pregnancy his mother wasn't anaemic or had any signs and symptoms of any disease.

#### 3.5. Generals

He presented with 1.7 kg weight having very lean thin look with pale complexion. He was taking formula milk but

seemed hungry all the time as his mother told. He was very irritable and weepy want to be covered all the time. His head was very soft with poor growth. His weight reduced from 2.8 kg to 1.7 kg due to lack of nutrition.

#### 3.6. Local and systemic examination

His skull bones were very soft with open fontanelle that was another clinical feature of Johnson- blizzard syndrome. He had Syndromic appearance- aplastic alae nasi that gave him a beak-shaped nose, low set ears, triangular upper lip with flat philtrum, a number of Café au lait spots on his trunk, upper and lower limb. (Figure 3)

His eyes were white and skin color was pale. He had flabby moist tongue and abdomen was tender, seemed full with gases.

#### 6.7. Lab investigations

His initial laboratory evaluation showed hemoglobin (Hb) of 5.4 gm%, haematocrit (Hct) of 9.6%, reticulocytes of 1.8%, total red cell count of 1.89 million/mm<sup>3</sup>, mean corpuscular volume (MCV) of 89.9 fl, mean corpuscular haemoglobin (MCH) of 28.6 gm, mean corpuscular haemoglobin concentration (MCHC) of 31.8 g/dl. Total white cell count was 4.5 x 10<sup>9</sup>/l, with 34% polymorphs, 60% lymphocytes, 4% monocytes and 2% eosinophils. The platelets count was 371 x 10<sup>9</sup>/l, and blood peripheral morphology was unremarkable (Figure 2). Bone marrow aspiration revealed erythroid hypoplasia with normal myeloid and megakaryocytes series. A diagnosis of primary pure red cell aplasia was made. His serum T3 and T4 levels were 120.00 ng/dl (normal range 70- 200) and 5.700 ug/dl (normal range 0.3-6.0) respectively while TSH level was 6.300 u IU/ml (normal range 0.3-6.0). Serum ferritin was 1083 mg/dl and TIBC was 57 ug/dl. Blood sugar was normal. His Bilirubin level was 11.2mg/dl.

#### 7. Analysis of case

After taking a detailed history of the patient and analyzing the symptoms of case, characteristics symptoms, particular symptoms, and physical generals were considered for the totality of the case. Symptoms like profuse greenish diarrhea, anemia due to nutritional deficiencies, Softness of skull, aggravation by cold, amelioration by warm, flabby moist tongue were included for the totality of the case.

Miasmatic evaluation of presenting complaints was done which showed the predominance of Psora Miasm. Considering the complaints Boricke's Repertory was selected for Repertorization of the case using HOMPAT software. (Figure-5,6)

#### 8. Intervention

After reprioritisation, many medicines were competing with each other like Calcarea Phos, Mercurius, Sulphur, Nux vomica, Phosphorus, Podophyllum, while maximum numbers took by Calcarea Phos in all symptoms. Podophyllum covered all the symptoms of diarrhea. 3x potency prescribed in powder form of both medicines.

### 9. Follow up & outcomes

Follow-up & date	Indications of prescription	Medicines & doses
April 13, 2008	Low Red blood cells, pale skin, 5-8 stools a day, diarrhea greenish and slimy, Flatulence, continuous weight loss,	<i>Calc phos</i> 3x, QID <i>Podophyllum</i> 3x, QID
June 3, 2008	Frequency of stool reduced, skin still pale, Low RBCs	<i>Calc Phos</i> 3x, QID

		<i>Podophyllum</i> 3x, QID for 15 days
June 17, 2008	Frequency of stools reduced, the weight started to increase,	The same prescription repeated
July 25, 2008	Skin color started to change from extremely pale to pinkish, 2 kg weight increased.	The same prescription repeated
August 20, 2008	Blood tests performed. RBCs count improved. Bilirubin decreased from 11.2mg/dl to 0.1 mg/dl. Frequency of Stools normal	<i>Calc Phos</i> 3x, QID <i>Podophyllum</i> 3x, BID For 2 months.
October 23, 2008	Normal skin color, frequency of stools remained normal with medicine, vomiting from 2 days, overall active	<i>Calc phos</i> 3x, QID <i>Podophyllum</i> 3x, BID <i>Arsenic</i> 6, TID (For 3 days only)
January 9, 2009	RBCs count improved, stools ok but with continuous medicine, weight increased.	<i>Calc Phos</i> 3x, QID and <i>Podophyllum</i> 3x, BID repeated
March 21, 2009	Blood tests performed. Normal RBCs count. Frequency of stools normal	Medicine discontinued

Medicine continued even after recovery, to stop the chances of recurrence. Blood tests performed after 8 months of discontinuation of the medicine. His blood count was normal (Figure 4) and he was Pure Red cell Aplasia free. His parents strictly refused to go for bone marrow evaluation because at the time of first bone marrow procedure he couldn't bear the application of medicine and became cyanosed. During the period of growth his milestones were excellent. He started crawling at the age of 6 months and used to pass social smiles. Due to hearing impairment he couldn't communicate but at the age of 8 months he started to refuse or accept things by his gestures. He started walking at the age of 13 months. He is now 11 years old. He visits after 3-4 months to take medicine whenever he suffers from diarrhoea. Some doses of *podophyllum* act well. Recently pancreatic insufficiency investigated from UAE, because this test is not available in Pakistan. His pancreatic elastase is < 50 ug Elast./g. (Figure 9). But there are no symptoms of pancreatic insufficiency. Due to his syndromic condition some issues still persist as, hearing impairment and short height. Besides this he is active, healthy and very intelligent school going boy. He is surviving without any sort of medication for blood deficiency. (Figure 10)

## 10. Discussion

Congenital pure red cell aplasia sometimes assumed as lifelong syndrome in which maturation of erythroblasts arrests by mutation [6]. PRCA with pancreatic insufficiency is commonly known as Pearson syndrome. But in this case report patient has clinical features of JBS, that is markedly featured by pancreatic insufficiency. Homeopathic medicines can cure this effectively. In this case, pure red cell aplasia is cured by pure homeopathic remedies. *Calc Phos* affects the nutrition of bones and glands. It supplies new blood cells & has been proven a great remedy in anaemia & chlorosis in my experiences. Lower potency of *Calc Phos* works well for the regeneration of bone marrow, as it is proved tissue salts. It is absolutely essential to the proper growth & nutrition of the body as it is essential part of blood plasma, corpuscles, saliva, gastric juices, bones, connective tissue, teeth, and milk etc. have the greatest importance to the soft & growing tissues, promoting cell growth. It supplies the 1<sup>st</sup> basis of the new tissue & hence necessary to initial growth and Important for the life of blood, without it there is no coagulation. Particularly this drug proves itself a real tonic in many cases, also in chronic wasting diseases. Corresponding to poor nutrition, whether

childhood, puberty, or of old age.

Johansson Blizzard syndrome is a genetic disorder that affects the multi-system of body. The spectrum of clinical features in patients with JBS is varying from person to person.<sup>9</sup> However, the characteristics features include mal-absorption of fats and other nutrients due to pancreatic insufficiency, growth retardation, abnormalities of permanent teeth, beak-shaped nose with intellectual disability, abnormalities of the skull and facial features.<sup>10</sup> It is assumed that there is no curative treatment<sup>11</sup> of this syndrome but a number of health conditions can be managed effectively by the homeopathic system of medicine. *Podophyllum* is a proven remedy for greenish, putrid stools gushing out painlessly. Cholera infantum with involuntary stools during sleep- Clark cured many cases of the prolapsed anus in children with *podophyllum*.<sup>8</sup> In this case baby has profuse, greenish diarrhea that seemed painless (key symptom of *podophyllum*) Because his mother told he weeps only at the time of wash-ups. Homeopathic medicines have curative and supportive role for treatment of blood related disease and syndromic health conditions.

**THE CHILDREN'S HOSPITAL, LAHORE**  
**DEPARTMENT OF PATHOLOGY**

NAME: *Amir* MEDICAL RECORD #: *48282*  
AGE/SEX: *M/2* DATE: *21/4/08*  
WARD/BED: ROOM:  
Lab. No. *517* CENTRAL OPD EMERGENCY

**HAEMATOLOGY**

**COMPLETE BLOOD COUNT (CBC)**  
RBC *5.4* X 10<sup>12/l</sup>  
Haemoglobin *1.8* g/dl  
HCT *17.0* l/l  
MCV *89.9* fl  
MCH *31.8* pg  
MCHC *28.6* g/dl

**MORPHOLOGY**  
Anisocytosis *N/N* Poikilocytosis */*  
Microcytosis *N/N* Macrocytosis */*  
Hypochromia *N/N* Polychromasia */*  
Spherocytes *N/N* RBC Fragmentation */*  
Eliptocytes *N/N* Target Cells */*  
Reticulocytes *N/N* Nucleated RBC's /100 WBC's *1.8%*  
Others *N/N*

ESR in mm at the end of 1st hour (Westergren's) method

**DIFFERENTIAL**  
WBC *4.5* X 10<sup>9/l</sup> Metamyelocytes */* %  
Neutrophils *34* % Myelocytes */* %  
Lymphocytes *60* % Promyelocytes */* %  
Monocytes *04* % Blast Cells */* %  
Eosinophils *02* % Platelet Count (Normal 150-400 x 10<sup>9/l</sup>) %  
Basophils *371*

MALARIA PARASITE  
HAEMO PARASITE  
COOMBS TEST  
CONCLUSION:

CONSULTANT HAEMATOLOGIST  
*21/4/08*

Fig: 1



**HAEMATOLOGY & TRANSFUSION MEDICINE DIVISION**  
 THE CHILDREN'S HOSPITAL & THE INSTITUTE OF CHILD HEALTH, LAHORE.  
 Ferozpur road, Lahore - 54600 (Pakistan)  
 Tel (042) 9230901-23 Ext. 2206 Fax (042) 9230338

Name: Afnan Aftab Medical Record #: 2024820  
 Age and Sex: 2 months old male Date: 14/04/08  
 Ward / Bed: 4/000 B.M No: 331/08

**BONE MARROW REPORT**

Site(s): Right Tibia  
 Consistency: Firm  
 Cellularity: Slightly hypocellular fragments and cell trails.  
 Erythropoiesis: Hypoplastic and normoblastic.  
 Leucopoiesis: Adequate with orderly maturation.

Myeloid-Erythroid Ratio (M:E Ratio): 6:1  
 Megakaryocytes: Adequate  
 Lymphocytes/ Plasma cell: \_\_\_\_\_  
 Extra Medullary Cell: \_\_\_\_\_

Iron staining (Perls Staining): \_\_\_\_\_  
 Special Staining: Present (+++).  
 Iron Stain: \_\_\_\_\_  
 Bone Marrow Imprint: \_\_\_\_\_  
 Bone Marrow Clot: \_\_\_\_\_  
 Bone Marrow Trephine: \_\_\_\_\_

Conclusion: Erythroid hypoplasia on bone marrow aspiration favours the diagnosis of pure red cell aplasia.

DR. NADIA SAJJID MBBS, FCPS Consultant Haematologist  
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 DR. NISAR AHMED Head & Consultant Haematologist Haematology & Transfusion Medicine Division

Fig: 2



Fig: 3

**Indus LAB**  
 99, Main Boulevard, Shadman 1, Lahore.  
 Tel: 7550555, 7550666 Fax: 7551655  
 Website: www.induslabs.net Email: info@induslabs.net

Name: AFNAN Age/Sex: 2 Year(s) / Male  
 Lab ID: ICL-2255-112009 Date: 20 - Nov - 2009  
 Reference: Standard Consultant: \_\_\_\_\_

**HAEMATOLOGY**

Test	Value	Units	Normal Range
ESR	5	mm/1st Hour	M: 0-10 F: 0-15
Hb	11.0	g/dl	M: 13-16 F: 12-14
WBC	7.4	$\times 10^9/l$	4.0-11.0
Platelets	271	$\times 10^9/l$	150-450

**DLC**

Test	Value	Units	Normal Range
Neutrophils	30	%	50-70
Lymphocytes	59	%	20-45
Monocytes	04	%	02-08
Eosinophils	07	%	01-04

**RBC Indices**

Test	Value	Units	Normal Range
RBC	4.37	$\times 10^{12}/l$	M: 4.5-6.5 F: 4.0-6.0
HCT	32.9	%	M: 38-52 F: 36-46
MCV	75.3	fl	80-96
MCH	25.2	pg	27-32
MCHC	33.4	g/dl	30-35

**RBC Morphology**

Test	Value
Normocytic	
Normochromic	

Issued By: \_\_\_\_\_

Dr. Shahzad S. Qureshi MBBS(Pb) M Med Sci(Sheffield) FRCPATH(UK) Consultant Pathologist Histo/Cytopathologist  
 Dr. Muteen Izhar MBBS(Pb) MSc(London) PhD(Cambridge) FRCPATH(UK) Consultant Pathologist Clinical Microbiologist  
 Dr. Nisar Ahmed MBBS(Pb) FCPS Consultant Pathologist Clinical Haematologist  
 Dr. Ejaz Waris MBBS(Pb) FCPS Consultant Pathologist Histo/Cytopathologist

Fig: 4

**Fig: 5**

**Fig: 6**

**Fig: 7**



**THE CHILDREN'S HOSPITAL & THE INSTITUTE OF CHILD HEALTH**  
 LAHORE, PAKISTAN PH: (92-42) 9230901-23 EXT. 2105 FAX (92-42) 9230358  
**Department of Paediatric Audiology, Speech Therapy & Communication Aids.**



Name:	Afnan	Age/Sex	1 ½ Year	male
Hosp #		Date	12-11-09	

**DISTRACTION TEST**

	LEFT	RIGHT
HIGH FREQUENCY RATTLE	NR dBA	NR dBA
'SS'	NR dBA	NR dBA
G. CHIME BAR (1600 Hz)	NR dBA	NR dBA
VOICE	110	110
C. CHIME BAR (500 Hz)	NR dBA	NR dBA
WARBLE TONES 500 Hz	>80 dBA	>80 dBA
1000 Hz	>80 dBA	>80 dBA
2000 Hz	>80 dBA	>80 dBA
4000Hz	>80 dBA	>80 dBA
DRUM		
OTHERS		
APR	+/- ve	+/- ve

**TYMPANOMETRY**

	E.C.V	COMPLIANCE	M.E.P	REMARKS
RIGHT EAR	0.9 cc	Np cc	Np dapa	Compliance- Decreased Pressure- Decreased
LEFT EAR	1.0 cc	Np cc	Np dapa	Compliance- Decreased Pressure- Decreased

**COMMENTS:**

The findings of the above tests suggest that **he has profound degree hearing loss bilaterally.**

*Referred to ENT department (317)*

*Decision of hearing aids will be made after treatment from ENT.*


**Follow up after 4 weeks.**

14.12.09  
at 2.30  
at A


*Dr. Liaquat Ali*  
 (DR. LIAQAT ALI)  
 Principal Paediatric Audiologist &  
 Head of Paediatric Audiology Department

**Fig: 8**





**المختبر المرجعي الوطني**  
National Reference Laboratory  
Managed by LabCorp



cap  
ACCREDITED

Patient Report

SH-02915

**MARSAD, YASEEN**

**Patient Details**

DOB: 29/07/2008

Age: 11 years

Gender: Male

MRN: ELLSHA02486

**Account**

Excel Labs Pvt Ltd

110 Fazal e Haq Road, Blue Area

Islamabad

Accession: 25-19-210-00008

Requesting Physician:

Reference Number:

---

Date Coll: 29/Jul/2019 10:50

Date Rec: 06/Aug/2019 00:09

Date Verified: 09/Aug/2019 21:35


LabCorp Results

Test	Result	Units	Reference Range	Specimen Type
Pancreatic Elastase, Fecal	<50 <span style="color: red;">LOW R</span>	ug Elast./g	>200	Stool
R:	**Results verified by repeat testing**			
	Severe Pancreatic Insufficiency:	<100		
	Moderate Pancreatic Insufficiency:	100 - 200		
	Normal:	>200		

Performed At: BN LabCorp Burlington

1447 York Court Burlington, NC 272153361

Nagendra Sanjai MD Ph:8007624344



Shereen Hassan Atef, MD  
Consultant Clinical Pathologist  
License No. Haad-GD6320, DHA-P-0017830

Pending Tests

Procedure	Accession	Test Site
Pancreatic Elastase, Fecal	25-19-241-00001	NRLAD Lab Corp SO Be

This document contains private and confidential health information protected by local regulations

NRLAD : National Reference Laboratory (Headquarters) | Abu Dhabi Business Hub | Unit C25/26, ICAD 1 | P.O. Box 92323, Abu Dhabi, UAE

NRLDXB: National Reference Laboratory | Dubai Science Park | Laboratory Complex | Ground Floor Lab Number 0013 | P.O. Box 2087, Dubai, UAE

Page 1 of 1

Fig: 9



Fig: 10

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