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The Characteristics of Thalassemia Patients in Northern China

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Abstract

Thalassemia was not uncommon in north of China.

Patient Concerns: The clinical data of 21 northern birthplace thalassemia patients were collected. **Diagnosis:** They were diagnosed as carrier thalassemia.

Interventions: The thalassemia patients in north of China were analyzed.

Outcomes: For a thalassemia patients, there were 4 parameters, included MCV (p=0.034), MCHC (p=0.03), RDW-CV (p<0.001) and RDW-SD (p<0.001), had significant differences and 2 parameters, included Hb (p=0.304) and MCH (p=0.08), had no differences with the normal population. For b thalassemia patients, there were 4 parameters, included MCV (p=0.031), MCH (p=0.044), RDW-CV (p<0.001), RDW-SD (p<0.001), had significant differences and 2 parameters, included Hb(p=0.179) and MCHC (p=0.432), had no differences with the normal population.

Conclusion: If the patient presented with microcytic hypochromic anemia whether or not combined with other hematology diseases, hemoglobin electrophoresis should be detected to distinguishing thalassaemia even in the north. It is the education to physician.

Keywords: Thalassemia; North of China; Education

Abbreviations: SF: serum ferritin, IDA: iron deficiency anemia, AA: aplastic anemia , HL: Hodgkin's lymphomas, MM: multiple myeloma, MDS: myelodysplastic syndrome, MCV: mean corpuscular volume, MCH: mean corpuscular hemoglobin, MCHC: mean corpuscular hemoglobin concentration, RDW-CV: red cell distribution width, RDW-SD: standard deviation of red cell distribution width , Hb:hemoglobin, SI: serum iron, TIBC: total iron binding capacity, EPO: erythropoietin, TfR: transferrin receptor.

Introduction

Thalassemia is an inherited disease with a defect in the synthesis rate of one or more globin chains. In China, it was mostly found in the southern part, but sporadic thalassemia was not uncommon in north of China [1]. The clinical data of 21 northern birthplace thalassemia patients were collected and analyzed since 2013. Most of them had symptoms with fever, headache or urine color depth, only 2 patients had fatigue. Some patients only exhibited abnormal on blood routine examination at onset in health examination. They were diagnosed as carrier thalassemia with ranged from 9 to 70 years old at onset, included 8 males and 13 females with 6 (28.6%) thalassemia and 15 (71.4%) thalassemia patients. The main data were showed in table 1, included erythrocyte parameters, serum ferreting (SF) and gene mutations.

Table 1: The erythrocyte parameters, SF and gene mutations in 21 thalassemia patients

No	diagnosis	RBC(4-5.5)×10∧12/L	Hb(120-160)g/L	MCV(80-100)fl	MCH(27-34)pg	MCHC(320-360)g/L	RDW-CV(11-14.1)%	RDW-SD(39-46)fl	SF(11-306.8)ng/ml	Gene mutations
1	α thalassemia	5.31	97	63.8	18.3	286	21.9	48.9	ND	ND
2	α thalassemia	4.72	95	72.5	20.1	278	23.5	57.3	ND	ND
3	α thalassemia +AA	3.24	74	78.7	22.8	290	22.8	55.6	2335	ND
4	β thalassemia	6.38	130	64.9	20.4	314	17.4	35.3	163.4	β^{CD17}/β^{N}
5	α thalassemia	6.6	139	68.8	21.1	306	16.8	36.4	ND	α α /--sea
6	β thalassemia	5.95	114	60.8	19.2	315	16.7	32.5	412.3	βIVS-2-654/βN
7	β thalassemia	5.88	116	61.9	19.7	319	15.3	34.2	111.6	ND
8	β thalassemia+IDA	6.04	108	57.1	17.9	313	15.4	31	4.2	ND
9	α thalassemia	5.05	94	69.7	18.6	267	23	55.3	40.1	ND
10	β thalassemia	6.62	132	66	19.9	302	18.6	37.7	82	ND
11	β thalassemia+MM	4.35	79	59.8	18.2	304	22.8	47.7	ND	ND
12	β thalassemia+MDS	-	40	62.4	18.8	301	20.8	42.5	203.54	ND
13	β thalassemia	4.12	91	69.2	22.1	319	16.1	37.9	ND	βCD41-42/βN
14	β thalassemia	-	117	63.9	20.1	315	16.4	36.8	701.1	βCD41-42/βN
15	β thalassemia	5.1	102	63.3	20	316	16	36.5	53	βIVS-2-654/βN
16	β thalassemia+HL	3.68	75	64.1	20.4	318	24.4	52.5	2184	ND
17	β thalassemia	-	110	80.2	25.2	316	22.5	60.1	ND	βCD17/β N
18	β thalassemia	4.45	113	78.7	25.4	323	20.6	57.3	573.6	βIVS-2-654/βN
19	β thalassemia+IDA	5.62	102	57.8	18.1	314	15.5	30.5	3.9	βCD17/β N
20	β thalassemia	5.7	107	63	18.8	298	14.8	31.6	69.6	βCD17/β N
21	β thalassemia	5.23	100	62.3	19.1	307	14.7	31.7	35.9	βCD17/β N

There were 2 cases with iron deficiency anemia (IDA), 1 case with plastic anemia (AA), 1 case with Hodgkin's lymphomas (HL), 1 case with multiple myeloma (MM) and 1 case with myelodysplastic syndrome (MDS). The patient's leukocytes and platelets were in the normal range except the one combination with AA. They did not need treatment, except combination with other hematology diseases.

Case Report with Results and Discussion

Patients had no significant differences in age and gender with the normal population. Erythrocyte parameters were lower in mean corpuscular volume(MCV), mean corpuscular hemoglobin(MCH) and mean corpuscular hemoglobin concentration (MCHC)[2], higher in red cell distribution width (RDW-CV) and standard deviation of red cell distribution width (RDW-SD) than normal population, except hemoglobin (Hb) (table 2).

Table 2: The analysis of erythrocyte parameters in thalassemia patients compared with normal population.

	Hb(g/L)	MCV(fl)	MCH(pg)	MCHC(g/L)	RDW-CV(%)	RDW-SD(fl)
normal range	120-160	80-100	27-34	320-360	11-14.1	39-46
α thalassemia	96.33±22.92@	68.88±6.61#	19.85±1.84$	288.5±15.02^	21.8±2.50&	50.2±7.80*
β thalassemia	103.8±22.59@@	65.04±6.56##	20.34±2.26$$	312.67±7.32^^	17.68±3.03&&	39.21±9.68**
normal population	138.41±12.27	90.73±3.18	30.38±1.15	335±7.92	12.61±0.50	41.62±1.82

Notes: Hb: hemoglobin, MCV: mean corpuscular volume, MCH: mean corpuscular hemoglobin, MCHC: mean corpuscular hemoglobin concentration, RDW-CV: coefficient of variation of red cell distribution width, RDW-SD: standard deviation of red cell distribution width. a thalassemia compared with normal population: Hb $^{@}$p=0.304, MCV $^{\#}$p=0.034, MCH $^{\$}$p=0.08, MCHC $^{\wedge}$p=0.03, RDW-CV & p<0.001, RDW-SD *p<0.001; thalassemiacompred with normal population: Hb $^{@@}$p=0.179, MCV $^{\#\#}$p=0.031, MCH $^{\$\$}$p=0.044, MCHC $^{\wedge\wedge}$p=0.432, RDW-CV p<0.001, RDW-SD **p<0.001.

Iron overload was common in thalassemia patients through inhibiting iron regulatory protein-iron modulin. But only a few patients expressed increased SF in our data, the others showed normal on serum iron (SI) and SF. There were 2 β thalassemia patients with IDA on decreased SI and SF and increased total iron binding capacity (TIBC), that would be misdiagnosed as pure IDA. IDA presented by iron-deficiency could reduce α globin chain synthesis, when the supply of β-globin chains are limited in β thalassemia, β-globin chains compete more effectively for α-globin chains than δ-globin chains, resulting in reduced HbA2. The SF is related to HbA2 level, and decreased SF relates to decreased HbA2 [3], that is positively associated with HbA2 and iron deficiency. β thalassemia characterized by the increased HbA2 could show the normal or even lower HbA2 with IDA, that would cover up β thalassemia. Hematological phenotypic analysis was a method for screening thalassemia. According to the international standards for screening thalassemia, patients with MCH<27pg and MCV<78fl should experience hemoglobin analysis and gene mutation detection. But now IDA patients with MCH<27 pg, MCV<78 fl should be suggested to examine hemoglobin analysis at onset and after iron therapy to avoid thalassemia misdiagnosis.

One β thalassemia patient with MM was diagnosed at 58 years old. The microcytic hypochromic anemia with Hb 60g/L was found when she was 27. When she diagnosed MM with microcytic hypochromic anemia in our hospital, Erythrocyte parameters exhibited by Hb 79g/LMCV 59.8fl and MCH 18.2pg. Then α thalassemia diagnosed after hemoglobin electrophoresis simultaneously. Thalassaemia was particularly tended to persistent stimulation of the reticuloendothelial system with chronic haemolysis. An initial antigen exposure resulted in immunocyte proliferation. After an oncogenic stimulus or mutation induced a sub-population clone proliferation, MM might happen.

There was a MDS patient happened thalassaemia in the data. During the physical examination in 2008, the blood routine examination of this patient indicated normal with WBC 8.3×109/L, Hb 148g/L, MCV 89.2fl, MCH 32.7pg, MCHC 366g/L, Plt 140×109/L. In 2016, the blood routine examination showed microcytic hypochromic anemia as WBC 5.35×109/L, Hb 40g/L, MCV 62.4fl, MCH 18.8pg, MCHC 301g/L, Plt 245×109/L. The patient was diagnosed as MDS-MLD by bone marrow examination, and β thalassaemia by hemoglobin electrophoresis. According to the blood routine examination in 2008, β thalassaemia was not belong to congenital, but was acquired. C. hoyle also reported a case of MDS combined with acquired β thalassaemia. However, the most of MDS patients were combined with acquired **α** thalassaemia, named ATMDS. It had been found that 43% of MDS patients were accompanied by microcytic cells had a ATRX mutation [4]. The main molecular mechanism of ATMDS was inactivating mutations of the ATRX gene, included somatic missense mutation or splicing defect in the ATRX gene, which caused the expression resting of immature α-globulin gene in the late period of erythropoiesis. Maybe, acquired β thalassemia in MDS patient might exist another gene mutation, that need further research. And there was one β thalassaemia patient accompanied with HL in our data,which might because of infectious diseases, disfunction of the complement system or oxidative injury by iron overload to neoplastic clone amplification.

In addition, SF (11-306.8 ng/ml) (p=0.003), folic acid (p=0.014), erythropoietin (EPO) (p=0.031), soluble transferrin receptor (TfR)(p<0.001) and transferrin (p=0.023) showed significant differences with the normal population (table 2). Gene mutations were all heterozygous mutation, included aa/--sea heterozygous mutation, CD17 heterozygous mutation, CD41-42 heterozygous mutation, and IVS-2-654 heterozygous mutation, of which CD17 heterozygous was the most common one.

Conclusion

If the patient presented with microcytic hypochromic anemia whether or not combined with other hematology diseases, hemoglobin electrophoresis should be detected to distinguishing thalassaemia even in the north [5] , especially along with abnormal parameters included folic acid, EPO, TfR, transferrin. It should pay attention to the patients with IDA presented with significantly reduced MCV and MCH, hemoglobin electrophoresis should be redone after iron supplementation to prevent missed diagnosis of thalassemia.

Author Contribution

Shijun Li wrote this paper and submitted it. Mingyuan Sun, Bo Jiang and Weiwei Zheng collected the data of patients. Junyuan Qi was the correspondent of this paper.

Disclosure of Conflicts of Interest

The authors report no conflicts of interest. The authors alone are responsible for the content and writing of this article.

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Ethics Approval Statement

The study protocol was approved by the Ethical Committee of Tianjin fourth central hospital, and the informed consent was gaven to all patients or the legal guardians if patient was under 18.

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