Pediatric Hematology Abstract Categories

Red Blood Cell Disorders
OP 17

ASSESSMENT OF VITAMIN B12 AND HOMOCYSTEINE LEVELS IN PREGNANT WOMEN ADMITTED FOR DELIVERY AND CORD BLOOD SAMPLES OF THEIR NEWBORN BABIES: A MULTICENTER STUDY

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Objective: Vitamin B12, an essential micronutrient, plays a vital role in various physiological processes, particularly during pregnancy and fetal development. The growing popularity of vegetarian diets and socioeconomic barriers to consuming animal-based products contributes to Vitamin B12 deficiency becoming a global issue. Understanding the B12 status in pregnant women and its potential impact on newborns is of utmost significance as it can have far-reaching implications for both maternal and infant health. This research aims to investigate the vitamin B12 and homocysteine levels in pregnant women admitted for delivery and analyze corresponding cord blood samples from their newborn babies. **Methodology:** This prospective study was conducted in three tertiary care hospitals and included pregnant women aged ≥16 years admitted for delivery and their newborns ≥34 weeks. The demographic data and the results of complete blood counts performed within the previous 24 hours before birth were recorded. The levels of vitamin B12 and homocysteine were measured in blood samples and cord blood samples taken from pregnant women and their newborns, respectively. The study parameters were compared between the two groups based on the mothers' and babies' homocysteine and B12 levels. **Results:** The study included 615 Turkish and 217 foreign pregnant women. Anemia affected 36% of pregnant, with a higher frequency in mothers with B12 deficiency. The mean B12 level in pregnant women was 157±75.3 pg/ml, with 14.8% having elevated homocysteine levels. The levels of B12 and homocysteine of the newborns were 234.7±13.2 pg/ml and 9.13±5.75 mol/L, respectively. Vitamin B12 deficiency was found in 48.9% of newborns, while homocysteine levels were slightly elevated or elevated in 19.1%; both findings were significantly more common in babies born to B12-deficient mothers. **Conclusion:** In our study, vitamin B12 deficiency was significant in pregnant mothers and their neonates, with a substantial connection to cord blood homocysteine levels. Further study is needed to determine the impact of this deficit on mother and newborn health. Implementing approaches to timely detecting Vitamin B12 deficiency and, if necessary, providing adequate Vitamin B12 supplementation during pregnancy could play a pivotal role in enhancing the health and well-being of both the mother and the child.

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OP 18

A GHOSAL HEMATODIAPHYSEAL DYSPLASIA CASE; EXCELLENT RESPONSE TO NON-STEROIDAL ANTI-INFLAMATORY DRUG TREATMENT

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Objective: Ghosal hematodiaphyseal dysplasia (GHDD) is a very rare autosomal recessive disease caused by prostaglandin metabolism disturbances due to biallelic mutations on chromosome 7q33-34 which lead to decrease in thromboxane synthase function. Previously long-term corticosteroid was the only treatment for GHDD. Currently, non-steroidal anti-inflammatory drugs (NSAIDs) as a targeted therapy are preferred alternatively. Here, a genetically confirmed GHDD case responsive to ibuprofen is presented. **Case report:** A 9-year-old girl presented to our clinic with severe normocytic anemia, swelling, and pain in her lower limbs. In physical and radiologic examination metadiaphyseal dysplasia was diagnosed. The diagnosis of GHDD was confirmed with genetic analysis. The patient was treated with ibuprofen (30 mg/kg/day) with excellent response to both pain and hematologic parameters in 15 days period. **Conclusion:** Ghosal

hematodiaphyseal dysplasia is a very rare disease. The patients manifest with metadiaphyseal dysplasia, severe anemia, chronic fatigue, and inflammation. Previously long-term corticosteroid was the only treatment for GHDD with multiple significant long-term complication risks. NSAIDs, in this case, ibuprofen, are the current and new treatment options with relatively safe side effect profiles. But only a few cases with short-term follow-up were reported in the literature.

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OP 19

THE SIGNIFICANCE OF NEXT-GENERATION SEQUENCING IN NON-IMMUNE HEMOLYTIC ANEMIAS AMONG NORMOCHROMIC-NORMOCYTIC ANEMIAS

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Objective: Next-generation sequencing studies increased the exact diagnosis of unexplained normochromic-normocytic anemias and other anemias. Targeted next-generation sequencing studies allow the diagnosis of cytoskeleton defects, atypical cases, and some enzyme deficiencies. We aimed to compare the children with non-immune hemolytic anemia (n=13), and the others without non-immune hemolytic anemia (n=19) in the means of demographics, diagnosis, detected mutations, and laboratories. **Methodology:** In this study, the children who were examined in the Pediatric Hematology-Oncology Clinic of Duzce University School of Medicine and had unexplained anemia (n=29) underwent next-generation studies. The demographics, laboratory values, and genetic findings of two groups (non-immun hemolytic anemia and the others) were compared. We also found two novel mutations, one with hereditary spherocytosis and one with hereditary elliptocytosis. Mean, standard deviation, median minimum, maximum, frequency and ratio values were used in descriptive statistics of the data. The distribution of variables was measured with the Kolmogorov-Simirnov test. Independent sample t test and mann-whitney u test were used to analyze quantitative independent data. The chi-square test was used to analyze qualitative independent data. SPSS 28. 0 program was used in the analysis Results **Conclusion:** The demographics and the laboratory results are explained in Table 1. Comparing the non-immune hemolytic anemia patients (n=13) with the others (n=19), we found that membrane disorders rates, identified mutations associated with anemia, mean cell volume, mean cell hemoglobin, thrombocyte, reticulocyte, and absolute reticulocyte levels were higher, hemoglobin and erythrocyte levels were lower in the non-immun lower in the non-immune hemolytic anemia group (Table 2). The novel mutations are shown

Table 1) Demographics of the patients with unexplained anemia

		Min-Max.	Median	Mean.±s.d./n-%
Age (years)		0.1 - 17.0	0.1	5.3 ± 4.8
Age at onset of symptoms (years)		0.0 - 17.0	0.0	2.1 ± 4.2
Gender	Girl			9 6.8%
	Boy			23 17.4%
Nonimmune Hemolytic Anemia (+)	(+)			13 9.8%
	(-)			19 14.4%
Nonimmune Hemolytic Anemia (+)	(-)			22 16.7%
	(+)			10 7.6%
Identified Anemia Mutation	(-)			29 22.0%
	(+)			3 2.3%
Other defined mutations	(-)			23 17.4%
	(+)			9 6.8%
Enzyme Deficiency	(-)			27 20.5%
	(+)			5 3.8%
Erythrocyte count (10^9)		1.7 - 4.6	1.7	3.3 ± 0.9
Hct (%)		14.7 - 38.0	14.7	27.9 ± 6.3
Hb (g/dl)		4.7 - 27.0	4.7	9.9 ± 3.7
MCV (fL)		14.7 - 111.0	14.7	84.3 ± 15.8
MCH (pg)		22.0 - 97.0	22.0	30.6 ± 12.6
MCHC (g/dl)		30.0 - 36.1	30.0	32.9 ± 1.5
RDW (%)		10.9 - 21.7	10.9	15.1 ± 2.6
Thrombocyte count (x10^3/ml)		94.0 - 567.0	94.0	354.1 ± 112.2
Reticulocyte (%)		0.1 - 23.7	0.1	3.9 ± 5.7
Adjusted reticulocyte count (%)		0.0 - 12.7	0.0	2.8 ± 3.3
Transfusion rates (/y.l)		0.0 - 4.0	0.0	0.6 ± 1.3
Total bilirubin (mg/dl)		0.0 - 14.0	0.0	4.0 ± 4.9
İndirect bilirübin (mg/dl)		0.0 - 13.2	0.0	2.8 ± 4.1
Ferritin (ng/ml)		14.5 - 1392.0	14.5	192.0 ± 315.8

Abbreviations: Hct: Hematocrite, Hb: Hemoglobin, MCV: Mean cell volüme, MCHC: mean cell hemoglobin concentration, RDW: red cell distribution width

Table 2) Comparing the children with versus without non-immune hemolytic anemia

		non-immune hemolytic anemia (+)		non-immune hemolytic anemia (-)		p	
		Mean.±SD/n-%	Median	Mean.±SD/n-%	Median		
Age (years)		5.1 ± 6.2	1.5	5.4 ± 3.9	5.0	0.247	m
Age at onset of symptoms		2.4 ± 5.7	0.0	1.9 ± 3.0	0.0	0.544	m
Gender	Female	6 46.2%		3 15.8%		0.061	X^2
	Male	7 53.8%		16 84.2%			
Immun hemolytic anemia	(-)	6 46.2%		16 84.2%		**0.023**	X^2
	(+)	7 53.8%		3 15.8%			
Identified anemia mutation	(-)	10 76.9%		19 100%		**0.028**	X^2
	(+)	3 23.1%		0 0.0%			
Other defined mutation	(-)	9 69.2%		14 73.7%		0.783	X^2
	(+)	4 30.8%		5 26.3%			
	(+)	3 23.1%		2 10.5%			
Erythrocyte (10^2/ml)		2.9 ± 0.8	3.2	3.5 ± 0.9	3.7	**0.030**	m
Hct (%)		26.2 ± 6.4	28.0	29.0 ± 6.1	29.8	0.234	m
Hb (g/dl)		8.6 ± 2.1	9.2	10.8 ± 4.3	10.2	0.058	m
MCV (fL)		90.8 ± 9.5	90.0	79.9 ± 17.9	82.0	**0.021**	m
MCH (pg)		35.1 ± 18.9	30.6	27.6 ± 3.2	27.0	**0.030**	m
MCHC (g/dl)		33.0 ± 1.7	32.6	32.9 ± 1.4	32.6	0.835	t
RDW (%)		15.4 ± 3.5	14.0	15.0 ± 1.8	15.0	0.673	m
Thrombocyte (x10^3/ml)		412 ± 104	384	314 ± 102	314	**0.013**	t
Reticulocyte (%)		6.6 ± 7.1	3.2	1.1 ± 0.6	1.2	**0.001**	m
Adjusted reticulocyte count (%)		4.4 ± 4.0	2.3	0.9 ± 0.4	1.0	**0.001**	m
Transfusion rates (/year)		0.9 ± 1.6	0.0	0.2 ± 0.6	0.0	0.150	m
Total bilirübin (mg/dl)		4.8 ± 4.7	3.3	3.2 ± 5.1	0.4	0.124	m
İndirect bilirübin (mg/dl)		4.0 ± 4.6	2.3	1.5 ± 3.0	0.2	0.065	m
Ferritin (ng/ml)		295 ± 186	236	144 ± 356	23	**0.005**	m

Abbreviations: Hct: Hematocrite, Hb: Hemoglobin, MCV: Mean cell volüme, MCHC: mean cell hemoglobin concentration, RDW: red cell distribution width, ^{t}t test / ^{m} Mann-whitney u test / $^{X^2}$ Ki-kare test