International Journal of Contemporary Research In Multidisciplinary

Case Report

Macrocytic Anemia as the Initial Manifestation of Hypothyroidism in an Infant: A Case Report

Dr. Sheikh Almas [1*], Dr. Sabavath Arun [1], Dr. Kalyadapu Nikhil [1], Dr. Pulendhar Reddy [1]
[1] MBBS, Osmania Medical College, Hyderabad, Telangana, India

Corresponding Author: *Sheikh Almas

DOI: https://doi.org/10.5281/zenodo.17069057

Abstract

Hypothyroidism is a well-recognized cause of anemia in both children and adults, most often producing normocytic or microcytic anemia. Macrocytosis, however, is much less commonly encountered in hypothyroid states, especially in infancy, where nutritional deficiencies such as vitamin B12 or folate deficiency remain the foremost considerations. We report the case of an 8-month-old infant who presented with failure to thrive, developmental delay, and macrocytic anemia and was subsequently diagnosed with primary hypothyroidism. The case highlights the importance of considering endocrine disorders in the evaluation of unexplained macrocytosis, as timely thyroid hormone replacement can lead to complete haematological recovery in addition to improving neurodevelopmental outcomes.

Manuscript Information

- **ISSN No:** 2583-7397
- **Received:** 09-07-2025
- **Accepted:** 23-08-2025
- **Published:** 06-09-2025
- **IJCRM:**4(5); 2025: 20-22

- **Plagiarism Checked:** Yes
- **Peer Review Process:** Yes

How to Cite this Article

Almas S, Arun S, Nikhil K. Reddy P. Macrocytic Anemia as the Initial Manifestation of Hypothyroidism in an Infant: A Case Report. Int J Contemp Res Multidiscip. 2025;4(5):20-22.

Access this Article Online

www.multiarticlesjournal.com

KEYWORDS: Anemia, Congenital Hypothyroidism, Developmental delay

INTRODUCTION

Anemia is among the most common clinical problems in infancy, and its etiological spectrum varies widely. Microcytic hypochromic anemia, usually from iron deficiency, is most frequently encountered, followed by macrocytic anemia secondary to folate or vitamin B12 deficiency. In the paediatric age group, macrocytosis is often a red flag prompting extensive evaluation for nutritional deficiencies, bone marrow failure syndromes, or inborn errors of metabolism [1].

Hypothyroidism is known to be associated with anemia, through mechanisms that include reduced erythropoietin secretion, diminished bone marrow activity, and impaired iron utilization [2,3]. While most of these cases are either normocytic or microcytic, a subset of patients may develop macrocytosis. Such presentations are well documented in adults with long-standing

untreated hypothyroidism, but reports in infants remain scarce [4,5].

CASE DESCRIPTION

An 8-month-old female infant was brought to the outpatient department with poor weight gain and constipation. Her parents reported that she had been excessively sleepy and less active compared to her peers. She was born at term following an uneventful pregnancy and normal vaginal delivery with a birth weight of 2.9 kg. There was no history of perinatal complications, neonatal jaundice, or sepsis, and newborn screening had not been performed. The infant was predominantly breastfed until six months of age, after which complementary feeds were introduced. Her dietary intake was adequate, and there was no history suggestive of restricted vegetarian diet, malabsorption, or chronic illness. Family history was unremarkable, and the parents were non-consanguineous.

On examination, the child appeared pale, lethargic. Anthropometric measurements revealed significant growth faltering with weight below the 3rd centile and length on the 10th centile, while head circumference was normal. She had coarse facies, dry skin, periorbital puffiness, sparse, brittle hair, and an open anterior fontanelle. Systemic examination was unremarkable.

Laboratory evaluation revealed hemoglobin of 7.8 g/dL with macrocytosis (MCV 105 fL), low reticulocyte count, and a peripheral smear showing macrocytosis without hypersegmented neutrophils. White cell and platelet counts were normal (Table 1). Biochemical work-up showed normal iron, vitamin B12, folate, and ferritin levels, as well as normal liver, renal, and electrolyte profiles. Endocrine evaluation demonstrated markedly elevated TSH (125 mIU/L) and low free T4 (<0.2 ng/dL), confirming primary hypothyroidism. In view of these findings and the absence of nutritional deficiency, bone marrow examination was not pursued. The child was started on oral levothyroxine. Parents were counselled on medication adherence and the importance of neurodevelopmental stimulation. After 3months, the infant showed marked improvement in activity, feeding, and bowel habits. Haematological parameters improved with haemoglobin rising to 10.5 g/dL and normalization of MCV. Thyroid profile also improved with free T4 reaching the normal range. By 3 months of follow-up, the child demonstrated improved motor activity, initiation of babbling, and a better sleep–wake pattern, reflecting both haematological and neurodevelopmental recovery following timely initiation of thyroid hormone replacement.

Table 1: Investigations of the patient

Parameter	Observed	Reference range
Hemoglobin (gm/dL)	7.8	11-15
Total leucocyte count (cells/microL)	7080	4000-11000
Neutrophils (%)	31	40-70
Lymphocytes (%)	59	20-40
Eosinophils (%)	3%	1-6
Monocytes (%)	7	2-8
Platelet count (X 10^5/microL)	2.56	1.5-4.5
Mean corpuscular volume (fL)	105	76-93
Mean corpuscular hemoglobin concentration (g/dL)	31.6	32-36
Mean corpuscular hemoglobin (pg)	32.8	27-32
Urea (mg/dL)	30	20-40
Creatinine (mg/dL)	0.3	0.2-0.7
Sodium (mEq/L)	135	135-145
Potassium (mEq/L)	4.2	3.5-5.5
Chloride (mEq/L)	104	98-107
Thyroid Stimulating Hormone (microU/mL)	125	0.4-4.94
Free T4 (ng/dL)	< 0.2	0.7-1.48

DISCUSSION

The relationship between hypothyroidism and anemia has long been recognized. Prevalence studies suggest that up to 30–40% of patients with untreated hypothyroidism may develop some form of anemia [2,5]. The majority are normocytic normochromic due to reduced erythropoietin synthesis and bone marrow suppression. Microcytic anemia results primarily from associated iron deficiency, which may occur due to menorrhagia in older females or impaired intestinal absorption [2]. Macrocytosis in hypothyroidism is less common, accounting for less than 10% of hypothyroidism-associated anemia [4,5]. Its mechanisms are incompletely understood. Hypothyroid states slow down haematopoiesis and impair DNA synthesis, leading to macrocytosis that resembles megaloblastic anemia [6]. However, unlike classical megaloblastic anemia, hypersegmented neutrophils are usually absent and vitamin B12 and folate levels remain within normal range. Several reports in adults have described macrocytic anemia as the presenting feature of hypothyroidism. However, paediatric data remain limited, with only isolated case reports [7]. Our case adds to the sparse literature documenting such an association in infancy. Importantly, the normalization of haematological parameters following thyroxine replacement alone strongly supports a causal relationship between hypothyroidism and macrocytosis in this patient.

From a diagnostic perspective, this case emphasizes two critical lessons:

1. **Broadened differential diagnosis of macrocytosis:** In infants, nutritional deficiencies, bone marrow failure

syndromes, and metabolic disorders are typically prioritized. Endocrine disorders such as hypothyroidism should also be kept in mind, especially when accompanied by systemic features like developmental delay, coarse facies, and growth faltering.

2. **Avoidance of unnecessary invasive procedures:** Recognition of clinical hypothyroidism in our patient helped avoid bone marrow aspiration or extensive metabolic testing, streamlining the diagnostic process.

From a therapeutic perspective, early initiation of levothyroxine is crucial not only for correcting anemia but also for optimizing neurodevelopmental outcomes. Delayed diagnosis of congenital or infantile hypothyroidism can result in irreversible intellectual disability [8]. Thus, cases like this reinforce the need for universal newborn screening and heightened clinical vigilance in settings where such screening is unavailable.

CONCLUSION

Macrocytic anemia in infants is most attributed to nutritional deficiencies, but may rarely be the first clue to underlying hypothyroidism. Clinicians should maintain a high index of suspicion for endocrine causes in infants presenting with anemia, especially when accompanied by systemic features of hypothyroidism. Timely recognition and initiation of thyroxine therapy can reverse hematological abnormalities and significantly improve developmental trajectory.

REFERENCES

1. Horton L, Coburn RJ, England JM, Himsworth RL. The hematology of hypothyroidism. *Q J Med*. 1976;45(177):101–123. PMID:1252402
2. Fein HG, Rivlin RS. Anemia in thyroid diseases. *Med Clin North Am*. 1975;59(5):1133–1145. doi:10.1016/s0025-7125(16)31876-1
3. Das KC, Mukherjee M, Sarkar TK, Dash RJ, Rastogi GK. Erythropoiesis and erythropoietin in hypothyroidism and hyperthyroidism. *J Clin Endocrinol Metab*. 1975;40(2):211–220. doi:10.1210/jcem-40-2-211
4. Jha A, Waghdhare S, Reddi R, Madan R. An unusual association between hypothyroidism and macrocytic anemia. *Thyroid Res Pract*. 2017;14(2):75–77. doi:10.4103/trp.trp_42_16
5. Erdogan M, Kösenli A, Ganidagli S, Kulaksizoğlu M. Characteristics of anemia in subclinical and overt hypothyroid patients. *Endocr J*. 2012;59(3):213–220. doi:10.1507/endocrj.ej11-0289
6. Christ-Crain M, Meier C, Huber P, Zulewski H, Staub JJ, Müller B. Effect of restoration of euthyroidism on peripheral blood cells and erythropoietin in women with subclinical hypothyroidism. *Hormones (Athens)*. 2003;2(4):237–242. doi:10.14310/horm.2002.1167
7. Shukla A, Bhatnagar S, Aggarwal A, Kaur R. Hypothyroidism presenting as macrocytic anemia in children: A rare association. *Indian J Pediatr*. 2012;79(6):832–833. doi:10.1007/s12098-011-0582-7
8. Rastogi MV, LaFranchi SH. Congenital hypothyroidism. *Orphanet J Rare Dis*. 2010;5:17. doi:10.1186/1750-1172-5-17