A child with beta thalassemia major in tribal area: A case report

Manisha Shukla[1], Department of Biotechnology, Pandit S.N. Shukla University, Shahdol, Madhya Pradesh, India, mshukla@bt.iitr.ac.in; drdemanisha@gmail.com
Phone No.: +91 8839096157
Neha Chauhan[2], Department of Biotechnology, Anna University Chennai, Tamil Nadu, India.

Introduction

Thalassemia refers to blood related, hereditary genetic disorder classified as alpha and beta thalassemia according to the hemoglobulin alpha and beta chain mutation respectively. Abnormal beta chain gene in pair leads to beta-thalassemia major morbidity. Further classified as Intermediate and Minor beta thalassemic condition with single mutated allele in confer to severity. India has the largest number of children with Thalassemia major in the world particularly in tribal population prevalence rate (0.36-13.20%) is higher compared to non-tribals. The manifestation appears as early as 6-24 months with growth retardation, sever hemolytic anemia, fever abdomen pain, delayed development, bone abnormalities and may be leads to mortality in grim cases.

Case report:

This case study is classic example of family inheritance of beta thalassemia. Our finding reports the case of a couple with beta thalassemic minor(hbA2) marks. Born to them major thalassemic girl (5.5 years) and a fetus(terminated) diagnosed with b-thalassemia major at 13 weeks. Village Shadole, Madhya Pradesh, India.

Clinical observations:

At 7th month hospitalization with whitish tinge and paleness of skin (hand and feet), growth retardation. Hb(5gm/dl). Treated with blood transfusion 1st time. Physical observation concludes blood related disorder. At 9th month rehospitalization chronic anemia observed. 2nd blood transfusion with iron chelating therapy with DFO and Deferasirox. Parents HPLC testing conclude b-thal minor. Regular blood transfusion (Hb), bone marrow or hematopoietic stem cell transplantation suggested.

Conclusion:

Early diagnosis, Genotyping literacy among youth and effective management are the way forward to reduce the burden of this malady.

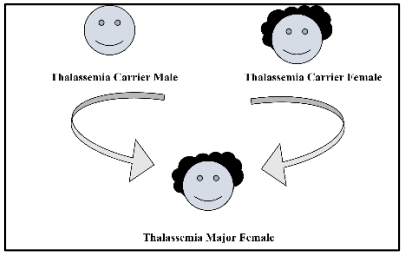


Figure1. Beta thalassemia case presentation

1. **Shukla M et al.** Expression, Purification, and Refolding of Chikungunya Virus Full-Length Envelope E2 Protein along with B-Cell and T-Cell Epitope Analyses Using Immuno-Informatics Approaches. ACS Omega. 2022, 1c05975.***Impact factor: 4.132***
2. **Shukla M et al.** The Role of B-Cells and Antibodies against *Candida* Vaccine Antigens in Invasive Candidiasis. **Vaccines**. 2021 Oct;9(10):1159. ***Impact factor: 3.641***
3. **Shukla M et al.** Expression and purification along with evaluation of serological response and diagnostic potential of recombinant Sap2 protein from *C. parapsilosis* for use in systemic candidiasis. **Journal of Fungi**. 2021, 7 (12), 999.***Impact factor: 5.724***
4. **Shukla M. et al.** Vaccination with secreted aspartyl proteinase 2 protein from *Candida parapsilosis* can enhance survival of mice during *Candida tropicalis* mediated systemic candidiasis. **Infection and Immunity**. 2020 Sep 18;88 (10):e00312-20.***Impact factor: 3.441***
5. **Sharma SK et al.** Isolation and characterization of emerging Monkeypox virus from India. Journal of Medical Virology. Article ID: 10.1002/jmv.28911. ISSN:0146-6615.**Impact factor: 20.69**



Currently, she is working as Assistant Professor in the Department of Biotechnology at Pandit S. N. Shukla University Shahdol, Madhya Pradesh, India. Her area of interests is immunology, molecular biology, infectious and genetic disease. She worked on various project related to viruses (COVID-19, monkey pox viruses, CCHF, KFD etc.) for 11 months as a Research Associate in DRDE-DRDO Gwalior, India. She obtained her PhD from Indian Institute of Technology Roorkee, India. During her doctoral degree, she worked on fungal vaccine (*Candida albicans* and *non-albican candida species*). Genetic Diseases and their prominence in the Indian population have always drawn her attention and particularly the existence of beta thalassemia and sickle cell anemia in tribal populations of India.