

## Hereditary Anemias in North West Frontier Province of Pakistan

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Hereditary anemias are common in cities, villages, mountainous area and tribal regions of Pakistan. Thalassemia: major/minor, sickle cell anemia, Hb, C and E diseases and G. 6. P. D. deficiency anemia have been well reported from all over Pakistan<sup>1,2</sup>. Other enzyme deficiency anemias, red cell membrane defect anemia and Hb.H and Bart hemoglobin diseases, though present in this area have not been reported frequently. The magnitude of this hereditary abnormality in the population of Pakistan is tremendous, however, its enormity has been masked by other preventable conditions like infectious diseases and nutritional deficiencies. If Pakistan overcomes the infectious and nutritional problems (hopefully by the year 2000: a target by World Health Organization), hereditary diseases including anemias will become the most important problem to be considered and tackled by the National Planners.

The problem of hereditary anemias in the North West Frontier Province (N.W.F.P.) of Pakistan is worse than the situation in other parts of the country because of its marginal inferiority in the economical and educational standard, which has a big say in the diagnostic facilities and treatment of these anemias. Comparing with Karachi,<sup>6</sup> our experience in Peshawar<sup>3,4</sup> is summarized here. This paper consists of a report of 200 cases of completely "worked up" children with gross anemias between the ages of birth to 12 years, hospitalized in PGMI/LRH, Peshawar for work up and management including blood transfusion. The hemoglobin ranged from 2.5 gram to 8 grams. The break-up of the type of anemias was as follows :- the largest group was nutritional anemias (41%) including iron deficiency anemias, megaloblastic anemia and mixed nutritional anemia. Next bigger group was that of hemolytic anemia (27%). Anemia due to acute or chronic hemorrhage was seen in 7% of cases and aplastic and hypoplastic group in 5.5% cases due to either septran, chloramphenicol, diphtheria or some undetectable causes. One case was due to congenital aplastic (Fancony's) anemia. A miscellaneous group (15%) consisted of leukemias, lymphoma, chronic infection (urinary or tubercular), Kwashiorkor, neuroblastoma, Wilms tumor and sideroblastic anemia. Cases that could not be diagnosed (due to lack of facilities) were 4.5%.

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Hemolytic anemias was the second biggest group of anemias seen in the hospitalized children at our Institute requiring diagnostic work up and repeated blood transfusions. Out of a total of 27% of hemolytic anemias, Thalassemia major was the commonest cause (19%) and G. 6. P. D. deficiency anemia (8%) was the next common cause in this group. Other causes were chronic malaria (4.5%) and thalassemia trait with combination of Hb.S, C&E.

From our this study of 200 hospitalized patients with anemia we had made the following conclusions. Firstly, three major groups of anemias had emerged, namely, iron deficiency group, megaloblastic group and hemolytic group. Secondly Thalassemia major was found to be the most frequent type of hemolytic anemia in the North West Frontier Province of Pakistan followed by G. 6. P. D. deficiency anemia. Since the political crisis in Afghanistan and the influx of Afghan refugees to Pakistan, mainly our province (the immediate neighbouring territory of Afghanistan), the problem has been aggravated because children of Afghan refugees have also similar carriage rate of hereditary anemia besides other diseases.

We have made some interesting observations :-

——— Thalassemias are genetically transmitted but some examples of genetic mutations are seen.

——— Thalassemia major seems to be more severe than in western countries because the typical clinical picture of onset is earlier than six months in many cases and death is also earlier due to severe disease or infections or lack of repeated blood transfusions.

——— The attitude of parents towards the prognosis of disease depends on the social economic status and grade of education. The educated and economically sound parents usually continue with repeated blood transfusion and Desferoxamine therapy. The un-educated parents with poor socio-economic status usually discontinue the management of the child once they know the prognosis.

——— Some time the mother of the affected child may become the victim of the society. Once the father knows about the genetics of the disease, and has many or all thalassemic children, he may decide to remarry for normal children in future.

Thalassemia major was described first in 1925 and 1927 by Cooley and associates<sup>1,2</sup>. The majority of patients have a mediterranean ancestry but now it has been described in a variety of non-mediterranean races with wide geographic distribution including Pakistan. The first report of Sick cell hemoglobin was given by Lehman<sup>5</sup> in a Pathan in 1961. Another report of Beta thalassemia and G. 6. P. D. deficiency anemia and Hb D-Punjab in Pathans was published by

Stern, Kynoché and Lehman<sup>7</sup> in 1968. Since then there have been reports of hemoglobinopathies from different provinces of Pakistan in the last 15 years.<sup>1,2,3</sup> The increased incidence of Thalassemia syndromes in Pathans of tribal areas and North West Frontier Province of Pakistan is explainable historically by the fact that most of the invaders of old India (including Greek invaders like Alexander, the Great) came through "Khyber Pass". "Khyber Pass" is now the gateway to Pakistan in the north and it has been historically in the pathway of hordes invading the subcontinent for thousands of years.

With the tremendous magnitude of hemoglobinopathies in Pakistan, the picture is gloomy. Only diagnostic facilities are available while repeated blood transfusion and iron chelating drugs can be obtained only by the rich people. Ignorance and poverty aggravate the situation. Foetal diagnosis and genetic counselling are not available. Future plans in Pakistan include early diagnosis, intensive blood transfusion, splenectomy when indicated and iron chelating therapy with Desferoxamine. These measures will decrease the mortality and morbidity and improve the life span and quality of living. Each hospital should have a "blood cell" where facilities for diagnosis and treatment should be available free of cost. Finally programme of prevention of hemoglobinopathies including foetal diagnosis, genetic counselling and community control of hereditary anemia should be available in specialised medical institutes of big cities of Pakistan.

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