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Screening for 3.7 and 4.2 deletion mutations in Sudanese patients suspected with alpha thalassemia

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Background & Aim: Alpha-thalassemia is the genetic disorders that have high prevalence in human population around all over the world, characterized by microcytic and hypochromic anemia. The carriers for the disease present with a mild anemia and like these patients in the rural medical centres especially in Sudan can be miss diagnosed as iron deficiency anemia, because of low facilities to do further investigations for differentiation, so those patients could take iron therapy without response which exposes them to the risk of hemochromatosis. The disease was not known in Sudanese and there were no published data. The 3.7 and 4.2 alpha gene deletion mutations are the most common types of the alpha thalassemia mutations in West Africa. This study aimed to screen the participant samples for the 3.7 and 4.2 deletion mutations at the molecular level and to correlate the findings with the CBC parameters in order to find out indicative criteria in routine hematological parameters that can help in diagnosis of alpha thalassemia in Sudanese patients which should be confirmed later by genetic investigations.

Method: This is a cross sectional study targeted 98 patients of highly suspected to have alpha thalassemia based on the microcytosis and hypochromasia of their RBCs, no past history of malaria (Plasmodium falciparum infection), normal serum ferritin level and free of any chronic diseases were selected to be screen for the 3.7 and 4.2 alpha gene deletion mutations by single tube multiplex GAP-PCR.

Result: The revealed of these 98 patients 7 were carriers for the 3.7 deletion mutation in the alpha globin genes and only one patient was 3.7 homozygous deletion mutation and all samples were negative for the 4.2 deletion mutation. The study revealed the 3.7 deletion mutation was found in Sudanese tribes originated from West Africa which are Four, Hawsa and Rezagat Tribes. The results showed the carrier patients of the 3.7 deletion mutation RBCs and HCT were significantly increased P-value <0.05, the RBCs were 7.23±0.78×1012/L in the adult male and 7.21±0.67×1012/L in adult female while in the children were 5.07±0.87×1012/L. The MCV and MCH were clearly decreased, but the MCHC slightly decreased. The Hb level revealed mild decrease without statistical significance P-value >0.05 in the adult males were 11.7±0.57 g/dl and 11.25±0.64 g/dl while in the children were 11.6±2.95 g/dl. The ferritin level was normal and the RDW CV clearly increased. The quantitative Hb electrophoresis was normal in addition to the presence of many target cells in peripheral picture and no one of these carriers presence with clinical manifestations indicating for anemia, but the homozygous 3.7 deletion mutation patient was anemic and his basic hematological parameters were as follows RBCs 1.38×1012/L, Hb 4.99 g/dl, HCT 11%, MCV 79.7 fl, MCH 35.5 pg, MCHC 44.5 g/dl, RDW CV 17.7% and the ferritin level were 1,807 mg/dl and this elevation due to the blood transfusion.

Conclusion: The study confirmed the presence of the alpha thalassemia in Sudanese population for the type 3.7 deletion mutation in the tribes that belong to the western reside of the Sudan and which is basically originated from the West Africa where the disease was already known and this transmission due to the migration.

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