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IMAGES

An Uncommon Variant Hemoglobin: Hb Ty Gard Detected from Gujarat, India

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More than 30 abnormal hemoglobins have been described in individuals of Indian and Pakistani origins which include 18 alpha chain variants, 14 beta chain variants, 2 delta chain variants, and one gamma chain variant as described in the HbVar database [1]. A number of abnormal hemoglobin variants have been characterized, mostly occurring at very low gene frequencies. We are reporting here a variant hemoglobin with relevant molecular characterization as identified by our Thalassemia carrier screening program among various high-risk population groups of Gujarat. The preliminary test was done with complete blood count (Sysmex KX-21), hemoglobin analysis with high performance liquid chromatography (VARIANT II Hemoglobin Testing System) followed by a confirmatory molecular analysis with Sanger sequencing (ABI 3730 DNA Analyzer, Applied BioSystems).

Case 1

A 15 years old male who was originally born in Ranghat village, North 24 Parganas district, West Bengal and moved to Jamnagar district, Gujarat with his parents where

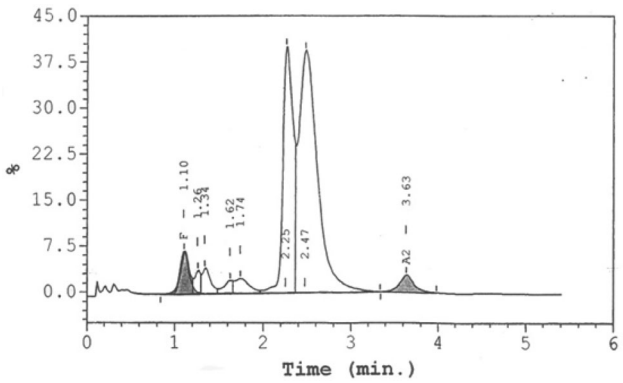

Fig. 1 HPLC chromatographs showing the variant hemoglobin

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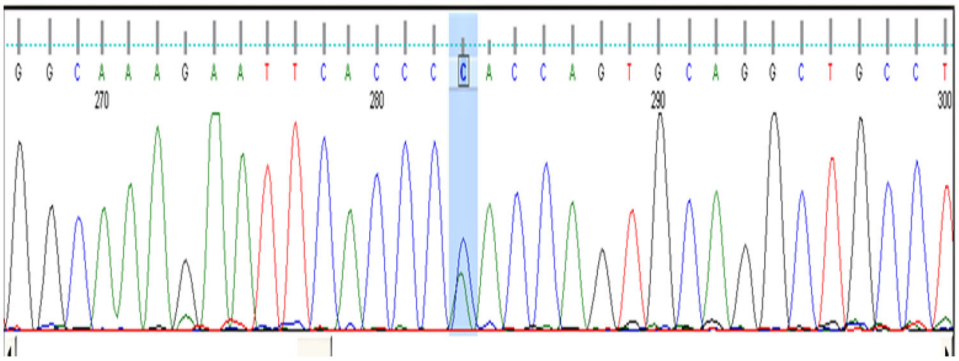

Fig. 2 DNA sequencing of the β globin gene showed a mutation in codon 124 CCA -> CAA, thus identifying the variant as Hb Ty Gard [α A2β2 124 (H2) Pro -> Gln]

he has been staying for the last 15 years and has no previous history of anemia or blood transfusion. Results of the hematological data are normal: Hb—15.7 g/dl, RBC—5.04 × 106/μl, HCT—47.8%, MCV—94.8 fl, MCH—31.2 pg, MCHC—32.8 g/dl and RDW (CV)—13.1%. His Hb F is high (4.7%) while Hb A2 is normal (2.8%). The A_O window is showing two distinct peaks which are highly abnormal and alerted the hematopathologist for the presence of a variant hemoglobin. The retention time of A_O window is 2.47 min (Fig. 1). DNA sequencing identifies the variant as Hb Ty Gard (Fig. 2).

The study is a part of Thalassemia carrier screening program among high-risk populations residing in Jamnagar district, Gujarat where the highest frequency of Beta-Thalassemia is reported among Halai Lohanas (17.20%) and comparably low frequency among Sindhi Lohanas (5.66%) [2]. The program has a population genetic research agenda as a contribution to basic research and ancestry targeted carrier screening with family counselling in a community setting is an important component.

The occurrence of different types of variant hemoglobins in India has been well documented in urban populations and in certain tribal areas. Most of the variants were discovered in clinical symptom-free patients. Hb Ty Gard is one of the high oxygen affinity hemoglobins. Higher fetal hemoglobin level, as we found in our case is reported to be able to compensate defective β-globin products and significantly reduce the severity of sickle cell anemia [3]. Further screening and characterization of these variant hemoglobins among high-risk populations for β-Thalassemia is important as it is likely to have adverse effects with advancing age for individuals who have such variant hemoglobin.

Compliance with Ethical Standards

Conflict of interest All authors declare that they do not have any conflict of interest.

Ethical Approval The work was approved by the Ethical Committee, Department of Anthropology, University of Delhi.

Informed Consent Informed consent was obtained from the individual include in the case.

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