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Clinico-Hematological Presentation of Rare Hemoglobin Variant (HB-O Indonesia) in 3 Families

Ravindra Kumar[1] · Purushottam Patel[1] · Anil Gwal[1] · Praveen K. Bharti[2] · Roshan Colah[3] · Shanmugam Rajasubramaniam[1]

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In India, Hemoglobinopathies poses a significant burden on the society. It can be prevented by screening, genetic counseling and antenatal diagnosis. During a community screening program, A rare Hb-O Indonesia (HBA1:c.349G > A) (HbO_{ind}) in 3 unrelated families of the same Jharia community were encountered. This variant has a heterogeneous phenotypic effect thereby necessitating a need for correlating it with associated clinical presentations. Here, red cell indices of individuals carrying HbO_{Ind} (other than the proband) were within the normal range (Table 1).

In case 1, the cause of low Hb level in proband may be due to other etiologies such as iron deficiency anemia or erythropoietin deficiency which is common in geriatric age. These etiologies were not investigated.

In the family 2, proband had microcytic hypochromic anemia along with mild splenomegaly suggesting a suspected clinical manifestation due to HbO_{Ind}. However the other causes of anemia and splenomegaly were not ruled out. On the other hand, proband's sibling, a compound heterozygote for both HbO_{Ind} and HbS, was asymptomatic with near normal red cell indices when tested.

In family 3, proband was symptomatic with mild anemia with microcytic and hypochromic RBCs and complained of frequent abdominal pain that may be due to coinheritance of HbS.

Individuals having HbO_{Ind} either in heterozygous or in compound heterozygous state with HbS presented here show variable hematological presentations warranting detailed molecular and routine clinical analysis. In all 3 cases, the Hb-O Indonesia was identified by hemoglobin electrophoresis at alkaline pH, High performance Liquid chromatography followed by sequencing of alpha globin gene (Fig. 1).

✉ Shanmugam Rajasubramaniam
raja.rmrct@gmail.com

[1] Division of Genetic Disorders, ICMR-National Institute of Research in Tribal Health, Nagpur Road, PO-Garha, Jabalpur, Madhya Pradesh 482003, India

[2] Division of Vector Borne Diseases, ICMR-National Institute of Research in Tribal Health, Jabalpur, Madhya Pradesh, India

[3] ICMR-National Institute of Immunohematology, Mumbai, Maharashtra, India

Table 1 Hematological indices of studied individuals

	Age/sex	HB (g/dl)	RBC (X 10^6/µl)	MCV (fl)	MCH (pg)	HbA (%)	HBA2 (%)	HBF (%)	HbO_{Ind} (%)	HBS (%)
Family 1										
Father	85 year/M	14.0	5.63	71.8	24.9	47.6	2.6	0.3	12.2	25.3
Mother (proband)	80 year/F	5.3	3.17	68.2	16.7	74.7	0.3	1.3	14.4	–
Offspring 1	51 year/M	14.4	4.18	77.8	26.8	59.7	1.4	0.2	29.6	–
Offspring 2	45 year/M	14.8	5.14	83.5	28.8	59.8	1.4	0.3	30.0	–
Offspring 3	40 year/F	13.3	5.13	77.8	25.9	69.8	1.9	0.1	19.4	–
Grand daughter	21 year/F	13.8	4.66	86.9	29.6	74.8	2.2	0.7	14.2	–
Family 2										
Father	40 year/M	15.0	5.19	85.4	28.9	57.3	2.6	0.4	–	30.7
Mother	35 year/F	11.2	4.26	82.6	26.3	71.5	1.6	0.3	11.7	–
Proband	4 year/M	11.1	4.77	72.1	25.3	67.5	1.5	0.5	18.9	–
Sibling	8 year/F	12.5	4.82	76.8	25.9	49.0	1.9	0.7	7.5	28.9
Family 3										
Father	35 year/M	15.2	5.49	87.3	27.6	84.8	2.7	0.3	–	–
Mother	32 year/F	10.9	4.32	77.8	25.2	48.1	2.5	0.6	6.4	32.8
Proband	16 year/F	10.3	4.45	71.4	23.1	49.2	2.2	0.5	11.3	25.6
Sibling 1	13 year/F	10.4	4.32	76.8	24.0	74.2	2.1	0.4	13.0	–
Sibling 2	10 year/F	10.9	4.86	69.0	22.4	48.4	2.4	1.0	11.2	25.5
Sibling 3	7 year/M	11.5	4.38	81.2	26.2	47.2	2.3	0.9	7.2	32.6

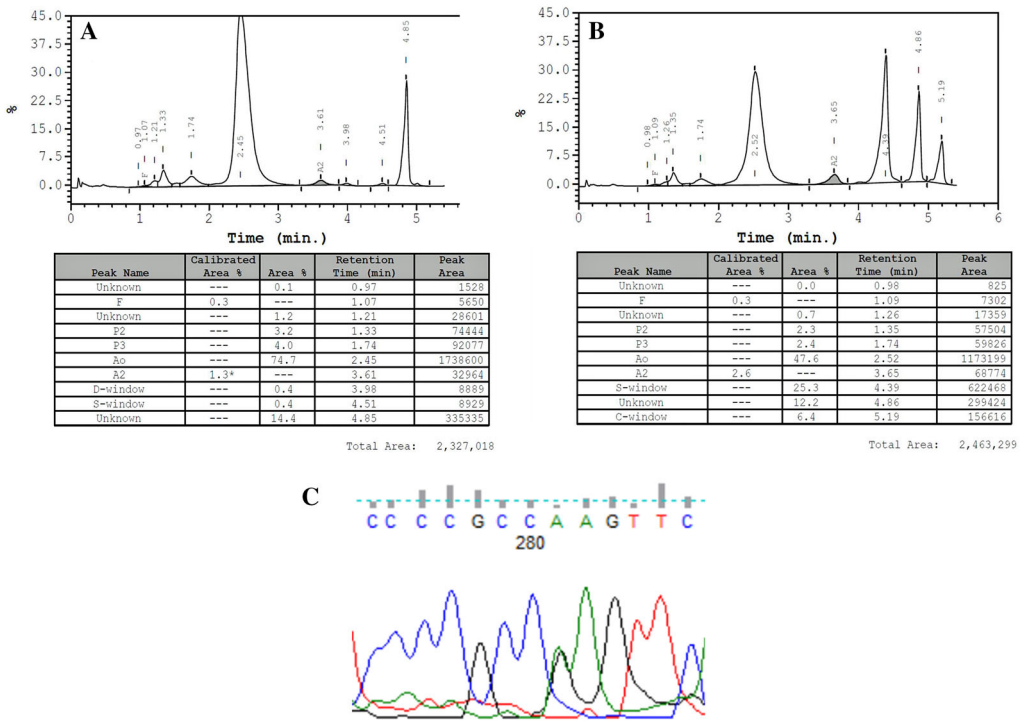

Fig. 1 **a** HPLC chromatogram of proband with HB O_{Ind}, **b** compund heterozygous of HbO Indonesia and HbS, **c** electrophorogram depicting G to A point mutation responsible for HB O_{Ind}

Compliance with Ethical Standards

Conflict of interest All authors declare that they have no conflict of interest.