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Editorial

Bombay phenotype in Orissa: What could we make out of it?

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In this issue of the journal, Balgir has reported three individuals with Bombay phenotype in the tribal population of North Western Orissa. Initially it may as appear to be a surprising finding but we continue to see a few cases of this phenotype belonging to Orissa. Our Institute had been approached for confirmation of their blood group for transfusion purposes. The blood banks in the tribal areas of Orissa may not have the necessary technology to identify the Bombay blood group and that is an area, which may need strengthening.

This study shows that Bombay ("Oh") phenotype, which was initially thought to be restricted to the western coast of India, may have a wider distribution in other parts of India. Two cases out of 244 Khandayat Bhuyan and one out of 379 Paudi Bhuyan is a very high prevalence by any account and needs further evaluation from a large population-based study. A little history may be relevant here and some detailed research may throw more light on the high prevalence of the Bombay phenotype in the Bhuyan tribe. It is known that the Maratha army reached the interior of Orissa during the height of their glory. An indication of that is still left in front of the Puri Jagannath temple. It is quite possible that this gene might have drifted from the far Konkan coast of western India to the other extreme eastern corners of the country. But how can we say that with certainty? Could molecular biology be of some help? Our study on the mutation of the Bombay phenotypes has shown that a missense mutation T725G in the FUT1 gene is responsible for almost all the Bombay phenotype cases in this part of the country[1] and also in the Indians settled in South Africa and Reunion Island. It will be interesting to see whether the Bombay phenotype found in Orissa can be explained by the same mutation on the backdrop of a different haplotype or by some other mutation. Secretor studies done on these individuals suggest the classical Bombay phenotype.

Long follow-up of more than 50 unrelated Bombay phenotype donors at our Institute has not shown any untoward propensity or resistance to common infectious diseases. Moreover it is generally agreed that Factor VIII and Von Willebrand factor (VWF) levels in plasma are epistatically controlled by ABO blood group antigens so that AB and A blood group individuals have high levels of these proteins compared to B and O blood group individuals.[2] A small study on "Oh" phenotype individuals elsewhere showed that plasma levels of factor VIII and VWF are like any other O group individual.[3] Similar results were found when Factor VIII levels in blood were correlated with salivary secretor status.[4] ABO glycosyltransferases and H-defining fucosyltransferases modify other blood group systems. Newborn children show "i" antigen in cord red cells, which slowly changes into "i" antigen due to continued glycosylation by ABO transferases "i" antigen have been found to be expressed in increasing amount in certain haematological disorders.[5] It may not be out of place to mention here that in Congenital Dyserythropoietic Anemia type II (HEMPAS) give full form here, increased 'i' antigen expression on red cells is characterized by abnormal N-acetylglucoseaminyl transferase activity.[6] However, in the "Oh" phenotype where no fucosyl transferase activity is seen, erythropoiesis is apparently normal probably because there are many different types of fucosyl transferases with different substrate speificites. Increased 'i' antigen expression has also been reported in the Bombay (Oh) phenotype by some workers.[7]

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In a very comprehensive survey, Sathe *et al.*, looked into the distribution of 179 Oh individuals according to their state of origin showing a rather wide distribution although major contributions were from Maharashtra, Karnataka, Goa and Andhra Pradesh.[8]

Whenever any mutation and polymorphism survives, it is often asked whether this mutation / polymorphism has any biological significance. Presently we have no information as to whether this mutation has any adverse eftect or any positive health benefit. A detailed questionnaire on various diseases and long follow-up of these individuals may unravel an hitherto unsuspected side of Bombay phenotype biology.

Finally, what all this means as far as blood bank services are concerned is that a systematic investigation of the population is essential for the identification of Bombay Blood Group individuals and their family members should be tested to identify any other member with this phenotype.

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