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Q8(b)

Describe the genetics and inheritance patterns of the ABO and Rh blood groups in man.

UPSC Civil Services Mains 2024 · Anthropology GS 1 · 15 marks · Mendelian genetics in man-family study, single factor, multifactor, lethal, sub-lethal and polygenic inheritance in man.

9.2 Mendelian genetics in man-family study, single factor, multifactor, lethal, sub-lethal and polygenic inheritance in man.

Core demand of the question

- Describe the genetics of ABO and Rh blood groups
- Explain genotype-phenotype relations and inheritance patterns
- Cover transfusion and haemolytic-disease significance
- Add population-anthropological use and cautions

Revision summary

ABO lies on chromosome 9 and encodes glycosyltransferases acting on H antigen.

I^A and I^B are codominant; both dominate common O.

Bombay phenotype hh lacks H antigen and demonstrates epistasis.

Rh is a chromosome-1 complex centred on RHD and RHCE, not literally one allele pair.

Maternal IgG anti-D can cause haemolytic disease after sensitisation; prophylaxis prevents most cases.

Blood-group frequencies trace populations but cannot define races or prove unique paternity.

Introduction

ABO and Rh are inherited red-cell antigen systems and classic markers of human variation. ABO illustrates multiple alleles and codominance; Rh, especially the D antigen, illustrates a genetically complex system with major maternal-foetal significance.

Body

ABO system

Karl Landsteiner discovered ABO agglutination. The **ABO** locus on chromosome 9 encodes glycosyltransferases. I^A adds N-acetylgalactosamine to the H antigen; I^B adds galactose; common i/O alleles produce inactive enzyme. I^A and I^B are codominant and both dominate O:

- **A phenotype:** I^AI^A or I^Ai; anti-B in plasma.
- **B:** I^BI^B or I^Bi; anti-A.
- **AB:** I^AI^B; both antigens and neither antibody.
- **O:** ii; neither A nor B antigen and both antibodies.

A and B parents can therefore have an O child only if both are heterozygous; paternity can sometimes be excluded, never uniquely proved, by ABO alone. **Bombay phenotype (hh)**, first reported in Mumbai, lacks H antigen and can type deceptively as O-an important epistatic exception.

Rh system

The Rh complex lies on chromosome 1, principally **RHD** and **RHCE**. Presence of D usually defines Rh-positive; absence or inactivation defines Rh-negative. "Positive dominant over negative" is a useful pedigree shorthand, but C/c and E/e antigens, variant D alleles and gene structure make Rh more complex than one two-allele locus.

Unlike naturally occurring ABO antibodies, anti-D usually follows sensitisation. An Rh-negative mother carrying an Rh-positive foetus may form IgG anti-D; in a later incompatible pregnancy it can cross the placenta and cause haemolytic disease of the foetus/newborn. Anti-D immunoglobulin prophylaxis greatly reduces this risk.

ABO and Rh frequencies helped early population anthropology map clines and affinity, including Indian caste and tribal samples. Drift, founder effect, selection and gene flow shape frequencies, but no blood group defines a race. DNA markers now provide finer histories.

Flow diagram

H antigen antigen -> IA adds IA adds A sugar sugar

H antigen -> IB adds IB adds B sugar sugar

H antigen -> i inactive

RHD present -> Rh positive

RHD absent or inactive -> Rh negative

Rh negative -> Maternal sensitisation

Maternal sensitisation -> IgG anti-D haemolysis

Conclusion

ABO is a clean example of multiple allelism, codominance and epistasis; Rh is a multi-antigen system whose D incompatibility matters in pregnancy. Both remain valuable teaching and clinical markers, but population frequencies are not racial essences.

Facts & figures

Karl Landsteiner

Discovered the ABO blood-group system and established the immunological basis of safer transfusion.

Bombay phenotype

The hh genotype prevents H antigen formation, so A or B transferases have no substrate; first recognised in Mumbai.

ABO codominance

I^AI^B individuals express both A and B antigens; this is codominance, not blending.

Anti-D prophylaxis

Rh immunoglobulin given to an unsensitised Rh-negative mother prevents immune response to foetal D-positive cells.

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