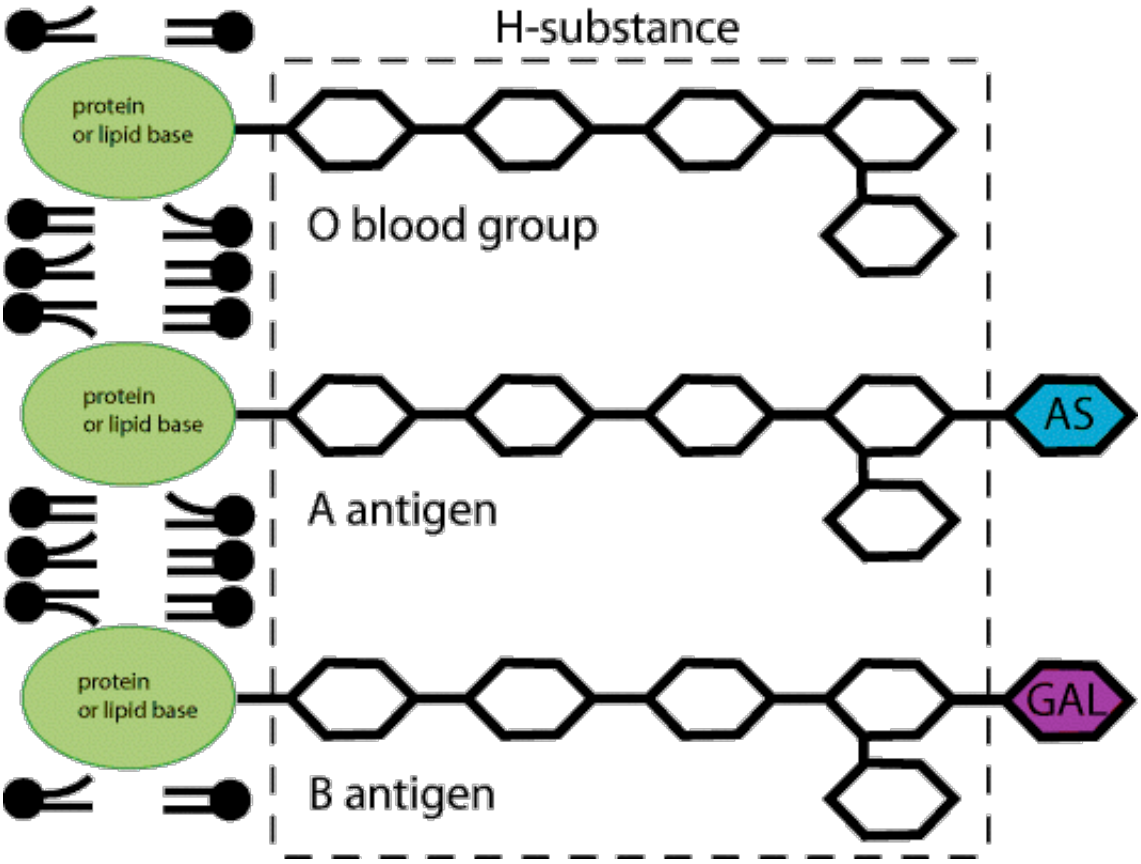
Molecule Genetics of the ABO Blood Group System

The ABO blood group system involves three major alleles, I^A, I^B, and i, which are located on the long arm of chromosome 9. The product of the I^A and I^B alleles are transferase enzymes that attach a specific sugar to a short chain of sugars known as **H substance** (see figure below). The enzyme coded for by the I^A allele adds **AS** (N-acetyl-galactosamine), and the enzyme coded for by the I^B adds **Gal** (galactose). The *i* allele codes for a protein, but this protein has no enzymatic activity.



Thus there are three carbohydrate antigens (A, B, and H). A, B, and AB individuals have transferases that convert the H substance into A and/or B antigens, whereas O individuals lack such enzymes and express only the H substance. The molecular basis for ABO genotypes is now understood. The DNA sequence of the I^A and I^B alleles differs in four nucleotide bases, changing four amino acids that cause differences in A and B transferase specificity. A critical single-base deletion was found in the sequence of the *i* allele, which results in an entirely different, inactive protein incapable of modifying the H substance.

The ABO antigens are expressed on the surface of cells other than the red

blood cells. This means that in any tissue transplant, it is necessary to match the ABO type of donor and recipient.

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