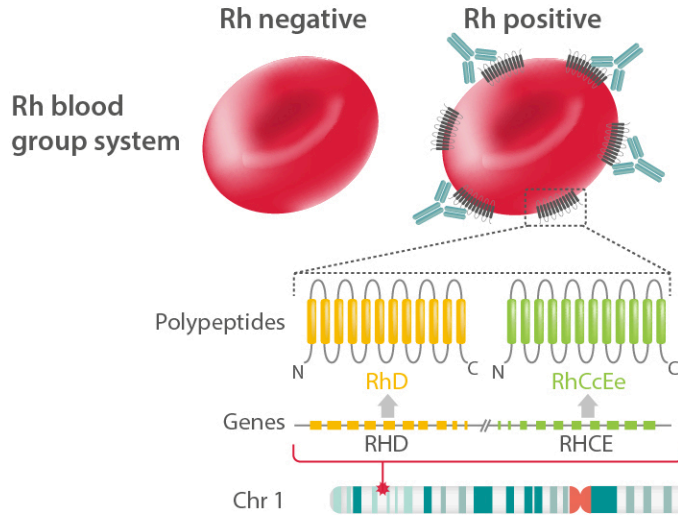


Somatic mosaicism with partial haploid loss of 1p involving the RH locus responsible for Rh blood group anomaly



- Rh antigens are highly immunogenic
- Antibodies against Rh antigens may elicit hemolytic transfusion reactions or hemolytic disease of the fetus and newborn
- D and c are the clinically most important Rh antigens
- All Rh antigens reside on RhD and RhCcEe polypeptides encoded by the RHD and RHCE genes, respectively
- RHD and RHCE genes are mapped to the short arm of chromosome 1 (p34-36)

Two unrelated female Caucasoid propositae without history of transfusion or HSC transplantation



RHD and RHCE genotyping + RHD zygosity determination of blood-derived DNA

A
53%
Normal

B
50%
Normal

- Proportion of c-positive red cells
- C+D+E-e+ Rh phenotype

Somatic mosaicism with partial haploid loss of 1p



Presence of two cell populations in which one lost one 1p segment

Presence of two cell populations in which one lost one allele