RBCs, whereas congenital cataracts occur in those i adults in whom all 3 IGnT-enzymes are defective, but not in analogs in which the IGnTC form is defective (Table 2).

The molecular genetics of the human I locus revealed in the present study provide a new view of the formation and expression of the I antigen. The architecture of the I-gene structures and the results obtained from the molecular analysis of the i adults answer many questions about the human I gene, the adult i phenotype, and the relationship with congenital cataracts. The most interesting deduction is that the I β6GlcNacT activity of the human I gene (possibly from the IGnTB gene) may play an essential role in maintaining lens transparency. It should be noted, however, that the lenses used for the RT-PCR analysis were from adults, and the question of whether the expression of IGnTB in the lens occurs during human embryonic development is left for future consideration. In addition, although the results of the present investigation are highly suggestive, direct evidence implicating an I-gene defect in the development of congenital cataracts is still lacking. The requisite evidence may be obtained through a gene knockout experiment in a mouse model. Further investigation to elucidate the functional role of I β6GlcNacT activity for maintenance of lens transparency will be significant.

References


Erratum

In the letter by Alvarez et al entitled “Cytogenetic characterization reveals that the SAM-1 erythroid cell line is derived from K-562 cells,” which appeared in the November 1, 2002, issue of Blood (Volume 100:3435), the cell line previously reported to be a derivative of K562 should have been TI-1.