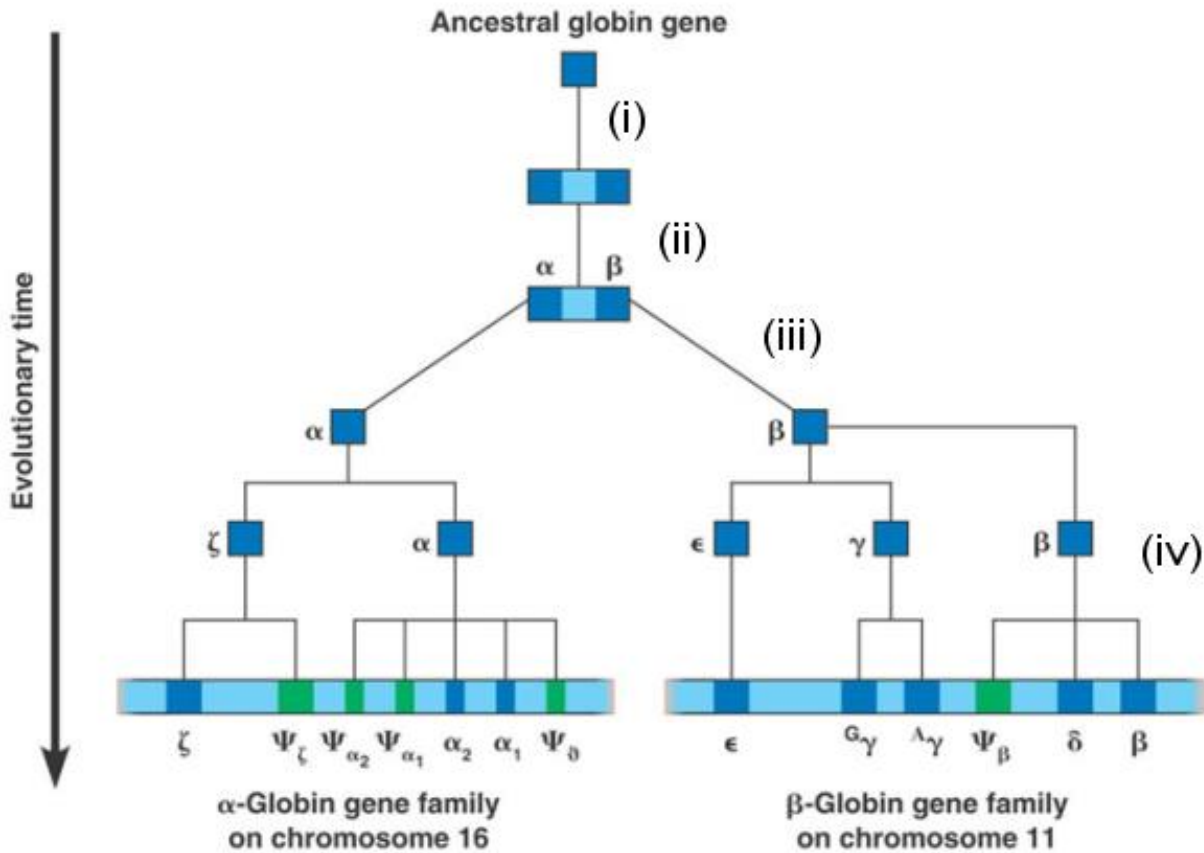


## Evolution of Human Globin Genes

Human hemoglobin is made of two alpha chains and two beta chains and the genes that encode the proteins are not identical.

Figure 1 shows the evolution of the human globin genes. There are currently two functional genes in the  $\alpha$ -globin family and three functional genes in the  $\beta$ -globin gene family.  $\alpha$ -globin and  $\beta$ -globin are the primary adult globins expressed. The others are expressed during embryonic and fetal development. Notice that the functional genes are interspersed with pseudogenes, or nonfunctional copies (indicated in the diagram in green).



**Figure 1** Evolutionary history of human globin genes

1. [SP 1, SP 2] Identify a mechanism that could explain the appearance of two copies of the globin gene (i).
2. [SP 1, SP 2] Describe a mechanism to account for the appearance of the original two globin genes,  $\alpha$  and  $\beta$  (ii).
3. [SP 1, SP 2] Propose a mechanism to account for the fact that the two original globin genes,  $\alpha$  and  $\beta$ , are now found on separate chromosomes (iii).

4. [SP 1] a) Provide an explanation for the existence of the members of both the  $\alpha$  and  $\beta$  globin gene families.
- b) Propose a reason that some of the versions are now pseudogenes.

Table 1 shows the similarity (in percent) in amino acid sequence between globin proteins in the two families.

<b>Table 1 Percentage of Similarity in Amino Acid Sequence Between Human Globin Proteins</b>						
		$\alpha$ -Globins		$\beta$ -Globins		
		$\alpha$	$\zeta$	$\beta$	$\gamma$	e
$\alpha$ -Globins	$\alpha$	100	58	42	39	37
	$\zeta$	58	100	34	38	37
$\beta$ -Globins	$\beta$	42	34	100	73	75
	$\gamma$	39	38	73	100	80
	e	37	37	75	80	100

5. [SP 6] Use the amino acid similarity between globin proteins in table 1 to justify the claim that the gene families originated from gene duplications.

The globin genes provide an example of very similar genes but mutation can result in novel genes also. Lysozyme is an enzyme found in both birds and mammals that protects against bacterial infection by damaging bacterial cell walls, killing the cell. The production of  $\alpha$ -lactalbumin is increased in response to prolactin and increases the production of lactose for inclusion in milk. The two proteins are quite similar in amino acid sequence.

6. [SP 1] Provide an explanation for the observation that both proteins are found in mammals but birds have only lysozyme.