

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 α -globin family and three functional genes in the β -globin gene family. α -globin and β -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).

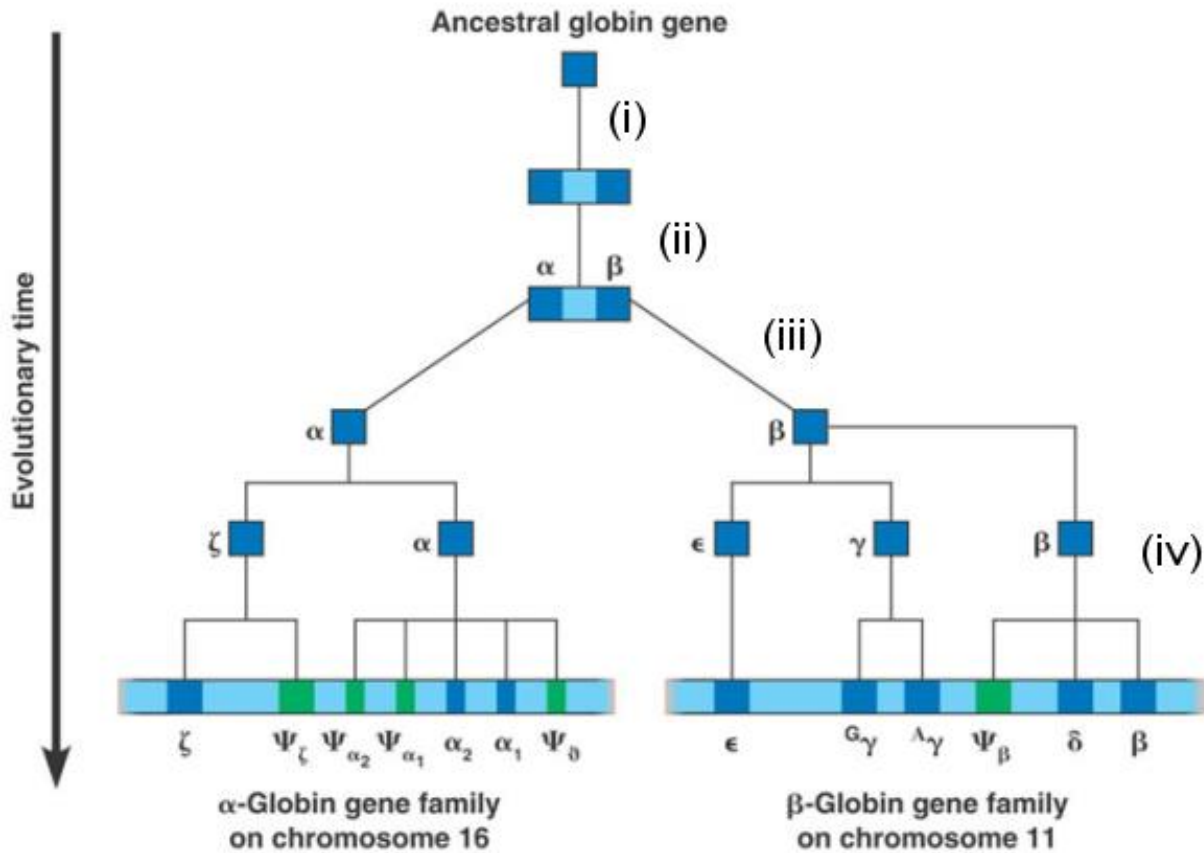


Figure 1 Evolutionary history of human globin genes

1. What event (i) during evolutionary time could explain the appearance of two copies of the globin gene?
2. Provide an explanation for the appearance of the original two globin genes, α and β (ii).
3. Explain how the two original globin genes, α and β , are now found on separate chromosomes (iii).

4. a) Explain the appearance of the members of both the α and β globin gene families.
 b) Why are some of the versions now pseudogenes?

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

Table 1 Percentage of Similarity in Amino Acid Sequence Between Human Globin Proteins						
		α -Globins		β -Globins		
		α	ζ	β	γ	e
α -Globins	α	100	58	42	39	37
	ζ	58	100	34	38	37
β -Globins	β	42	34	100	73	75
	γ	39	38	73	100	80
	e	37	37	75	80	100

families.

5. How does the amino acid similarity between globin proteins support the idea that the genes 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 α -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. Suggest an explanation for the observation that both proteins are found in mammals but birds have only lysozyme.