Figure 1.

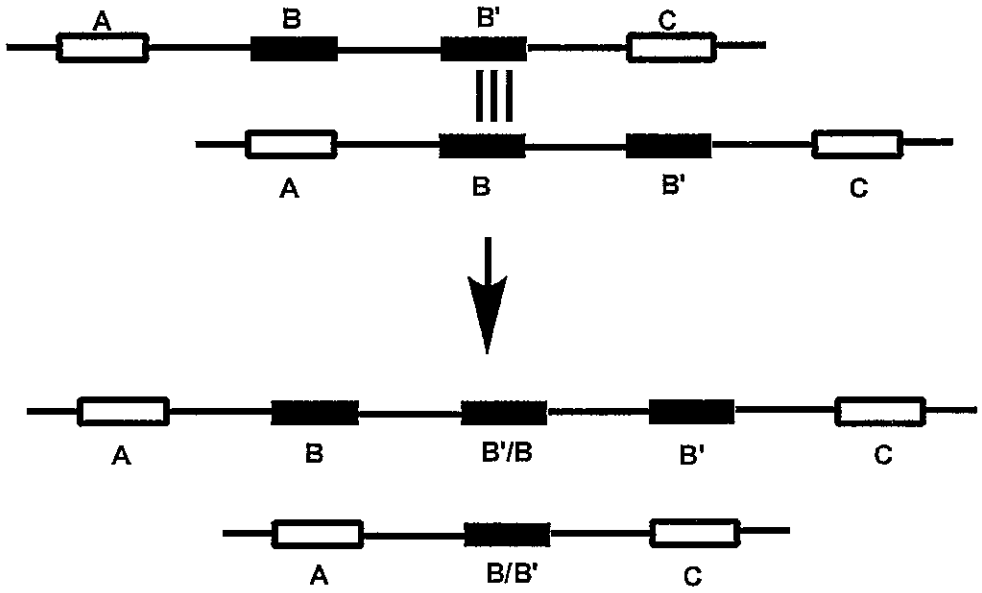


Figure 2.

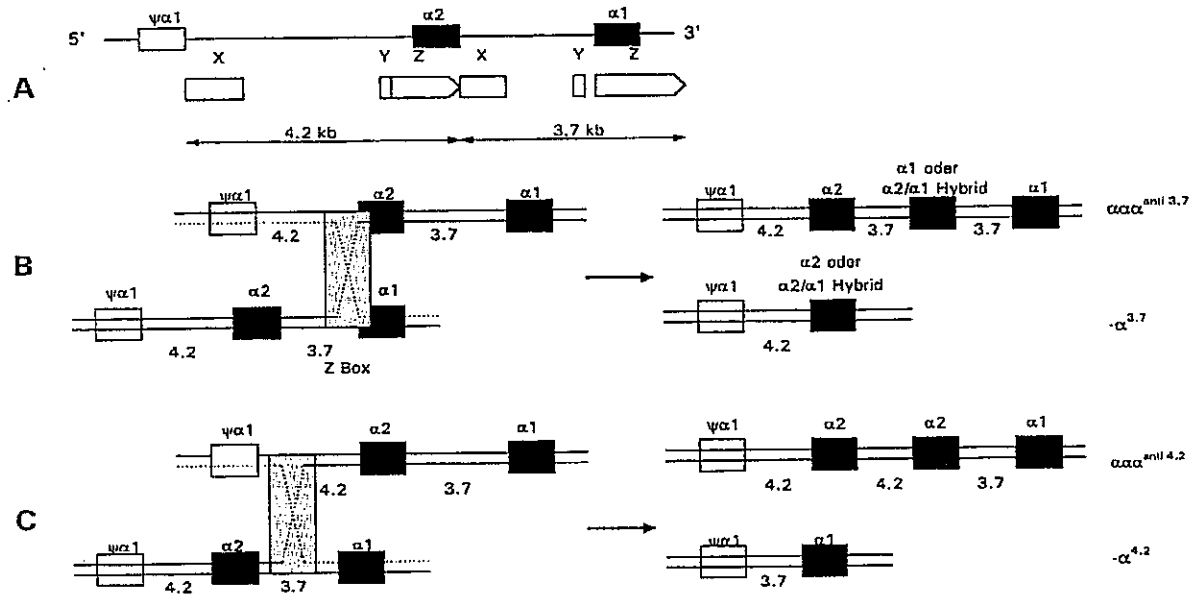
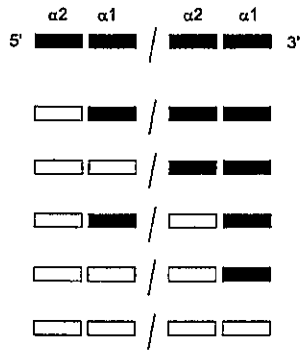
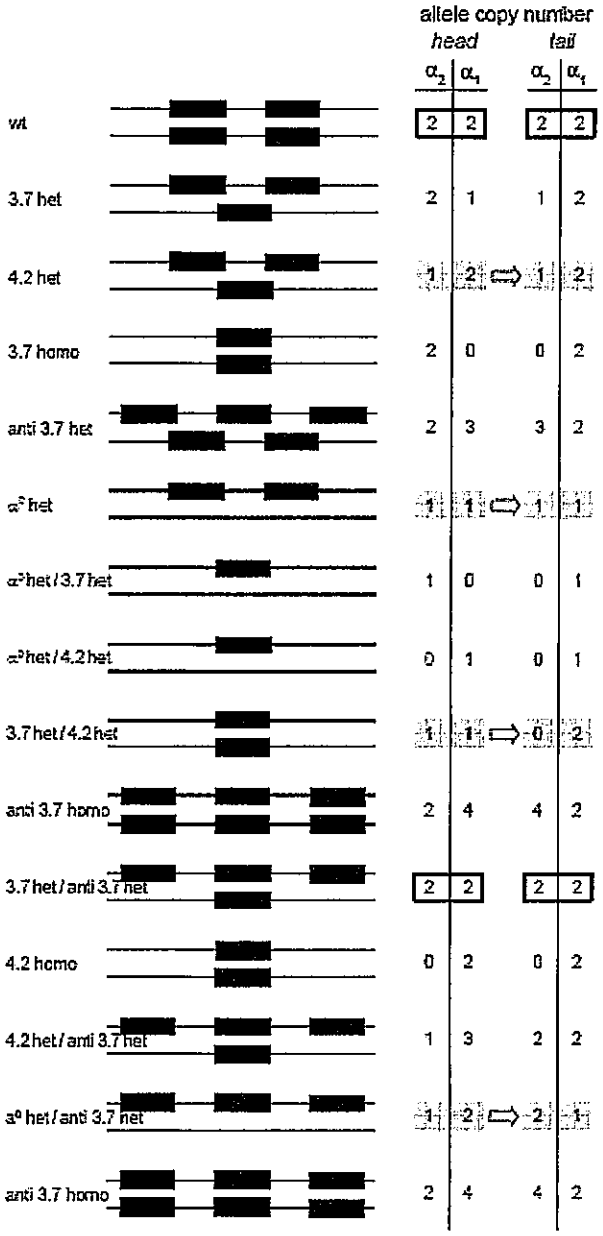

A
5'
ψα1
α2
α1
3'
X
Y
Z
X
Y
Z
4.2 kb
3.7 kb
B
ψα1
α2
α1
4.2
3.7
Z Box
α1 oder
α2/α1 Hybrid
α2 oder
α2/α1 Hybrid
αααanti 3.7
-α3.7
C
αααanti 4.2
-α4.2

Figure 3.



Genotype	Diagnosis	Clinic
αα/αα	normal	
αα/αα	α^+-thalassemia heterozygote	Unapparent form
– –/αα	a^0-thalassemia heterozygote	Thalassemia minor
– α/– α	α^+-thalassemia homozygote	Thalassemia minor
– –/– α	α^+/α^0-thalassemia mixed heterozygote	HbH disease
– –/– –	α^0-thalassemia homozygote	HbBart's hydrops fetalis Syndrom

Figure 4:


allele copy number
head
tail
α2 α1 α2 α1
wt
2 2 2 2
3.7 het
2 1 1 2
4.2 het
1 2 1 2
3.7 homo
2 0 0 2
anti 3.7 het
2 3 3 2
α0 het
1 1 1 1
α0het/3.7het
1 0 0 1
α0het/4.2het
0 1 0 1
3.7het/4.2het
1 1 0 2
anti 3.7 homo
2 4 4 2
3.7het/anti3.7het
2 2 2 2
4.2 homo
0 2 0 2
4.2het/anti3.7het
1 3 2 2
α0 het/anti 3.7 het
1 2 2 1
anti 3.7 homo
2 4 4 2