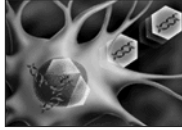


Imunogenetika a imunogenomika



Imunogenetika

Definice

- Klasická
- Aktuální

Imunogenetika

Definice

- Klasická
- Aktuální

Definice

Klasická

**Studium genetických rozdílů
pomocí imunologických
metod**

Definice

Klasická

Genetika krevních skupin

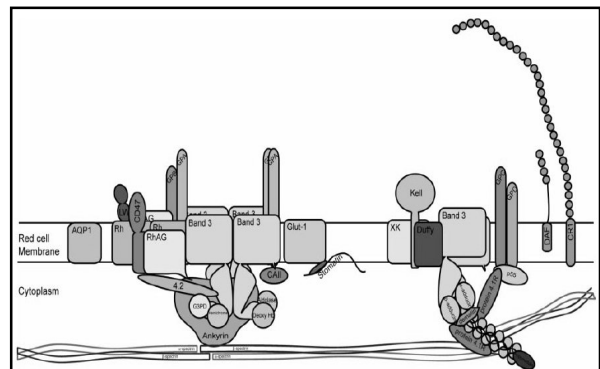


Figure 3. Structure of the human red cell membrane showing the major surface proteins and minor proteins Fy and CR1. Two major membrane complexes linked to the underlying red cell skeleton are depicted. The Band 3 complex containing glycoproteins A (GPA) and B (GPB) and Rh proteins, Rh-associated protein (RfAG), CD47, Lf glycoprotein (intercellular adhesion molecule-4), and the junctional complex comprising glycoproteins G and D (GPC, GPD), Kell glycoprotein, XK glycoprotein, and Duffy (Fy) glycoprotein. Aquaporin 1 (AQP1), the glucose transporter (GLUT1), decay accelerating factor (DAF, CD55), and complement receptor 1 (CR1) are also shown. ABH active oligosaccharides known to be present on all major surface proteins except Rh proteins are not depicted.

Definice

Krevní skupiny

- **Gen:** *krevněskupinový systém*
- **Alela:** krevní (feno)*skupina*
- **Epitop:** krevní *faktor*

KS systémy lidí a zvířat

Dělení

- **Primární a sekundární**
- **Jednoduché a komplexní**
- **Jiná dělení**

KS systémy lidí a zvířat

Funkce

- **Strukturální**
- **Receptorová**
- **Transportní**
- **???**

KS systémy lidí a zvířat

Molekulární podstata

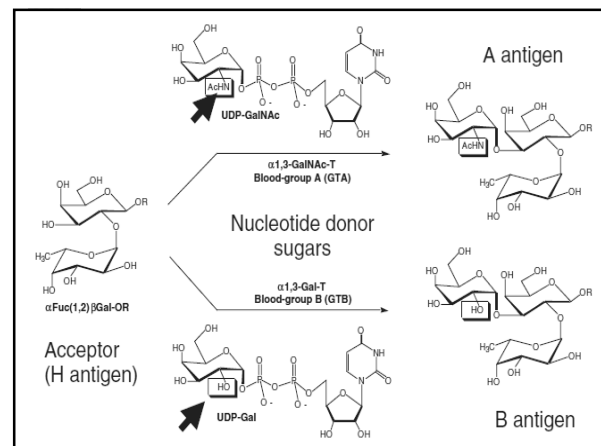
- **Proteiny a jejich geny**
- **Polysacharidy, enzymy a jejich geny**

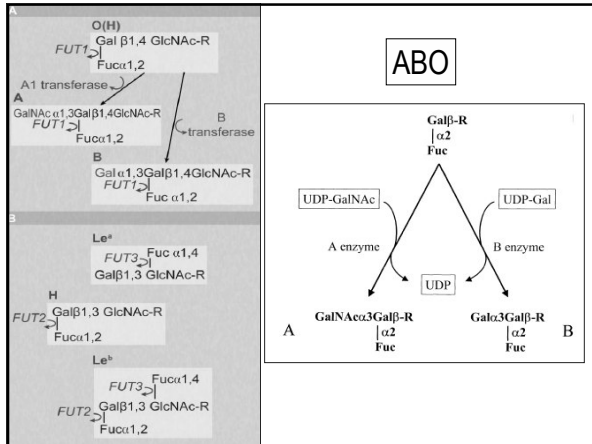
KS systémy lidí a zvířat

Teoretický význam: modely

Příklady

- **ABO vzácné alely**
- **ABO hybridní alely**
- **Genetická organizace Rh**





Genetika ABO

Table 1. Comparison of the Nucleotide Residues that Produce Amino Acid Substitutions Discriminating GTA from GTB

Glycosyltransferase	526 (176)	703 (236)	796 (260)	803 (268)
GTA	C (Arg)	G (Gly)	C (Leu)	G (Gly)
GTB	G (Gly)	A (Ser)	A (Met)	C (Ala)

NOTE: Although 4 other nucleotides are polymorphic between the consensus A101 and B alleles, they do not alter the amino acid sequence of the enzyme.

Polymorfismus ABO

Exon	1	2	3	4	5	6	7	8	9	10	11	12												
nt. position	53	106	188	189	220	261	297	467	526	646	657	681	703	771	796	802	803	829	930	1061	1096	1106		
Consensus	G	G	G	C	C	G	A	T	C	T	C	G	G	C	C	G	G	G	C	G	G	C	G	T
A ¹ -1																								
A ¹ -2																								
A ¹																								
B																								
O ¹																								
O ¹ '																								
Amino acid	18	36	63	63	74	87	99	156	176	216	219	227	235	257	266	268	268	277	310	354				
Consensus	Arg	Val	Arg	Arg	Pro	Val	Thr	Pro	Arg	Phe	His	Pro	Gly	Pro	Leu	Gly	Gly	Val	Leu	Pro				
Change	Leu	Phe	His	His	Ser			Leu	Gly	Ile			Ser				Arg	Ala	Met					

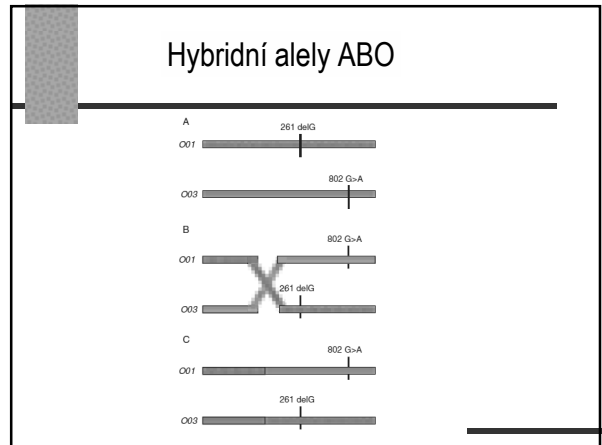
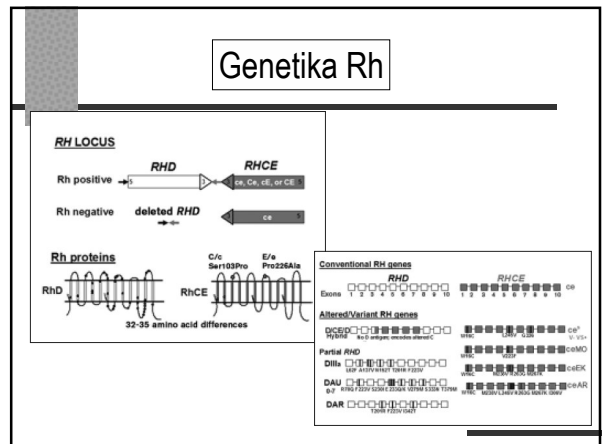
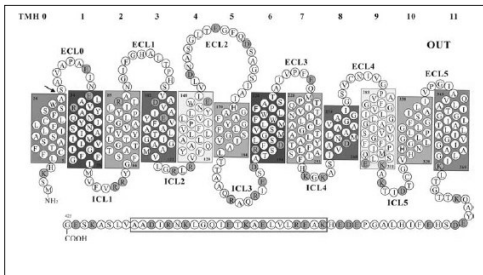


Table 1. Summary of selected ABO genotyping methods

Method ^a	PCR ^b	RE ^c +/-	Detection ^d	N. positions considered ^e											Ref.										
				261	297	467	526	703	796 ^f	802	1061	1096													
PCR-RFLP	4	+	PAGE-ET	x				x	x	x														1	
PCR-RFLP	2	+	AGA-ET	x																					2
PCR-DGGE	1	-	PAGE-ET		*																				3
PCR-ASP	1	-	PAGE- ³² P						x	x															4
PCR-RFLP	2	+	PAGE-ET	x																					5
PCR-RFLP	1	+	PAGE-ET	x																					6
PCR-RFLP	3	+	AGA-ET ^g	x						x	x														7
PCR-RFLP	1	-	PAGE-SS	x																					8
PCR-RFLP ^h	1	+	AGA-ET	x					x																10
PCR-RFLP	2	+	?	x						x															11
PCR-RFLP ⁱ	5	+	PAGE-ET	x						x	x														12
PCR-RFLP	2	+	PAGE-ET	x							x														13
PCR-DGGE	1	-	PAGE-ET	*																					14
PCR-ASP	8	-	AGA-ET	x																					15
PCR-MS	2	-	AGA-ET	x																					16
PCR-ASP	1 ^j	-	?	x																					17
PCR-RFLP	4	+	PAGE-ET	x																					18
PCR-SSO ^k	2	-	SB	x						x	x														19
PCR-SSCP	4	-	PAGE-SS	*	*	*	*	*	*	*	*	*	*	*	*	*	*	*	*	*	*	*	*	*	20
PCR-RFLP	4	+	AGA-ET	x																					21
PCR-ASP	1	-	PAGE-ET	x						x	x														22
PCR-ASP	7	-	AGA-ET	x																					23
PCR-ASP	2	-	PAGE-ET	x							x														24
PCR-SSCP	2	-	PAGE-SS	*	*	*	*	*	*	*	*	*	*	*	*	*	*	*	*	*	*	*	*	*	25
PCR-SSCP	2	-	PAGE-SS	*	*	*	*	*	*	*	*	*	*	*	*	*	*	*	*	*	*	*	*	*	26
PCR-SEQ	4	-	?	*	*	*	*	*	*	*	*	*	*	*	*	*	*	*	*	*	*	*	*	*	27



Rh protein

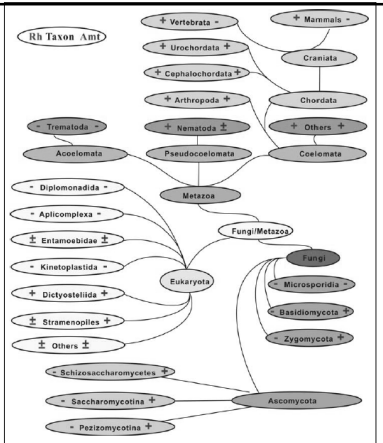


Rh protein: evolve

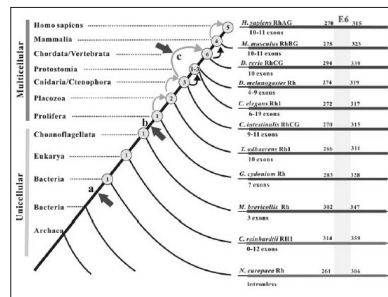
Table 1 Comparison between bacterial and human Rh proteins

Taxonomic name	Bacterial Rh homolog		Human Rh protein family				
	ORF (aa)	% IdSm (aa align)	RhBG	RhBG	RhCG	RhCE	RhD
<i>Proteobacteria β</i>							
<i>Nitrosomonas europaea</i>	425	100/100 (425)	3758 (354)	3651 (407)	3453 (387)	2948 (316)	3149 (277)
<i>Nitrosomonas</i> sp. AL212	457	70/83 (401)	3858 (357)	3955 (407)	3390 (430)	2948 (314)	3351 (283)
<i>Nitrosomonas multiformis</i>	407	73/84 (407)	3657 (355)	4129 (340)	3655 (313)	2847 (313)	2950 (317)
<i>Proteobacteria δ</i>							
<i>Geobacter</i> sp. M21	403	51/66 (362)	3655 (342)	3755 (301)	3351 (336)	2948 (284)	2948 (286)
Planctomycetes							
<i>Kuvenenia mariturgensis</i>	585	50/67 (360)	3355 (368)	3959 (313)	3457 (296)	2546 (321)	2546 (338)
Fimicutes-Chloridia							
<i>Chloridium carboxidovorans</i>	404	50/70 (345)	3555 (376)	3656 (346)	3455 (313)	2949 (311)	2652 (277)
<i>Chloridium celidivorum</i>	401	50/70 (345)	3658 (345)	3660 (319)	3090 (384)	2747 (318)	2852 (289)
<i>Chloridium pygmaeolens</i>	391	63/78 (356)	3757 (352)	3858 (306)	3253 (379)	2748 (340)	2849 (335)
<i>Desulfotomaculum acetoxidans</i>	400	51/68 (346)	3857 (341)	3556 (347)	3455 (292)	2750 (304)	3053 (273)
Acidobacteria							
<i>Acidibacterium bacterium</i>	390	61/75 (388)	3656 (345)	3654 (342)	3454 (312)	2746 (332)	2949 (290)
Archaea							
<i>Archaeoglobus fulgidus</i> Amt1	391	26/44 (222)	2744 (240)	2843 (216)	2843 (153)	4148 (97)	2844 (225)

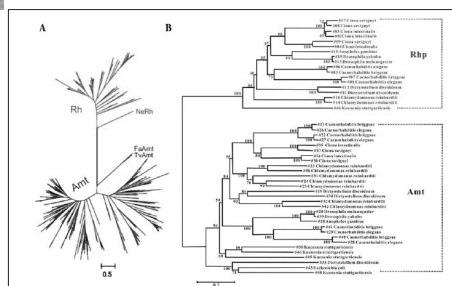
Rh protein: evolve



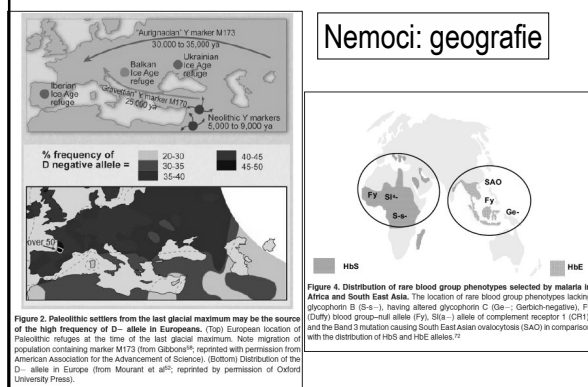
Rh protein: evolve



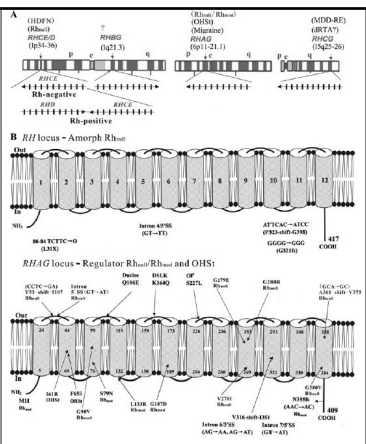
Rh protein: evolve



Nemoci: geografie



Nemoci:
molekulární
podstata



KS systémy lidí a zvířat

Praktický význam

- Krevní transfúze
- Inkompatibilita matka/plod
- Markery mapovací, asociační, populační
- Asociace s nemocemi
- Paternita

KS systémy zvířat

Zvláštnosti a význam

- Komplexní systémy
- Krevní transfúze
- Inkompatibilita matka/plod
- Markery mapovací, asociační, populační
- Asociace s nemocemi
- Paternita

Skot

Locus	Chrom.	Linkage group	Synteny group	Antigens
A	15	5	19	A D H Z
B	29	12	27	B ₁ G ₁ I ₁ K ₁ O ₁ O ₂ Q ₁ O ₁ P ₁ P ₂ Q ₂ Q ₂ T ₁ T ₂ Y ₁ Y ₁ A ₁ B ₁ D ₁ E ₁ E ₂ E ₃ F ₁ G ₁ J ₁ J ₂ K ₁ O ₁ P ₁ Q ₁ Y ₁ B ₁ G ₁ P ₁
C	18		9	C ₁ C ₂ E ₁ R ₁ R ₂ W X ₁ C ₁ L ₁
FIV	21	7		F ₁ V ₁
J	11	2	16	J
L	3		6	L
M	23	1	20	M M'
S	21	7	4	S ₁ U ₁ U ₁ H ₁ U ₁ U ₂ H ₂ S ₂ U ₂
Z	8 (10 ^{rat})	12	18 (5 ^{rat})	Z
R/S'	16		1	R ₁ S ₁
T'	19		21	T'

Psi a kočky

Table 1. Geographic Distribution of Blood Types in Domestic or Nonpurebred Cats

Country	Reference	Type A (%)	Type B (%)
USA (by region)	16		
Northeast	16	96.7	0.3
North central/rocky mountain	16	99.4	0.4
Southeast	16	98.5	1.5
Southwest	16	97.5	2.5
West coast	16	94.9	4.7
Australia	8	73.3	26.3
England	17	97.0	3.0
France	17	85.0	15.0
Switzerland	17	99.6	0.4
Finland	17	100	0.0
Italy	17	98.8	11.2
Netherlands	17	96.1	3.9
Scotland	17	97.1	2.9
Austria	17	97.0	3.0
Germany	18	92.6	6.7
Denmark	18	96.1	1.9

Table 2. Blood Type B Frequencies in United States Purebred Cats*

Purebred Cats	Frequency of Type B Cats
Siamese, Oriental Shorthair, Burmese, Tonkinese, and Russian Blue	0%
Maine Coon, Norwegian Forest Cat	<5%
Abyssinian, Himalayan, Birman, Persian, Somali, and Sphinx	5%-25%
Devon Rex, British Shorthair	25%-50%