GERMAN DATA ON THE LOCI LOW-DENSITY-LIPOPROTEIN RECEPTOR, GLYCOPHORIN A, HEMOGLOBIN γ^G, D7S8, GROUP-SPECIFIC COMPONENT AND HLA-DQα

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Summary

This paper reports on the allele and genotype frequencies for six polymerase chain reaction-based loci. The loci Low-Density-Lipoprotein Receptor, Glycophorin A, Hemoglobin γ^G, D7S8 and Group-Specific Component have been typed in samples from Düsseldorf (n=371), Aachen (n=107) and Saarland (n=100) (Germany). The HLA-DQα system was examined in a sample from Düsseldorf (n=579), Aachen (n=107) and Ulm (n=168). Typing was carried out by using the AmpliType® PM PCR amplification and typing kit and the HLA-DQα Forensic DNA Amplification and Typing Kit (AmpliType™, Perkin Elmer Cetus). The loci were tested for possible divergences from Hardy-Weinberg expectations (HWE). The gene frequencies found can be used in forensic analyses and paternity tests.

Materials and Methods

DNA extraction was performed from German Caucasians. For amplification and visualization the AmpliType® PM and the HLA-DQα PCR amplification and typing kits were used. Preparation of the samples, hybridization and visualization have been previously described (Perkin Elmer). Gene frequencies have been calculated by gene counting, the power of exclusion was calculated according to Krüger et al 1968.

Results and Discussion

AmpliType® PM Kit: The distributions of observed and expected genotypes are shown in table1. No deviations from the Hardy-Weinberg equilibrium were found. In the case of Glycophorin A (Aachen and Saarland) no χ^2-test was practicable because of df = 0, but comparison of observed and expected values results in good agreement.
The gene frequencies found in our three samples are shown in table 2 together with the data of 3 other German samples.
The AVACH values for the Düsseldorf sample [pooled German data (2166 genomes) in Italics] are LDLR (18.62 %; *18.50 %*), GYPA (18.62 %; *18.62 %*), HBGG (19.79 %; *19.87 %*), D7S8 (18.08 % ;*18.24 %*) and GC (29.39 %; *30.06 %*) which result in a AVACH value of (69.27 %; *69.61 %*) for the five loci together.
AmpliType® HLA-DQα Kit: The distributions of observed and expected genotypes are shown in table3. The Ulm sample is not with the Hardy-Weinberg equilibrium. Because we could exclude possible mistakes (foreigners eg) we think that this fact is caused by the size of the sample. The AVACH value for the Düsseldorf sample is 59.78%, for the Aachen sample 53.05 %. The pooled German data result in an AVACH value of 60.02 %.
For all six systems the AVACH value (pooled data) is 87.90 %.

	Düsseldorf				Aachen				Saarland			
	observed		expected		observed		expected		observed		expected	
	n	%	n	%	n	%	n	%	n	%	n	%
Low-Density-Lipoprotein Receptor (LDLR)												
A	80	21.56	75.17	20.26	24	22.43	23.36	21.83	23	23	20.70	20.70
B	117	31.54	112.17	30.24	31	28.97	30.36	28.38	32	32	29.70	29.70
AB	174	46.90	183.65	49.50	52	48.60	53.27	49.79	45	45	49.60	49.60
Σ	**371**	**100.00**	**370.99**	**100,00**	**107**	**100.00**	**106.99**	**100,00**	**100**	**100.00**	**100.00**	**100.00**
	χ^2= 1.03 df=1 30 > p > 50				χ^2=0.06 df=1 80 > p > 90				χ^2=0.86 df=1 30 > p > 50			
Glycophorin A (GYPA)												
A	110	29.65	112.72	30.38	34	31.78	31.44	29.38	29	29	28.09	28.09
B	72	19.41	74.72	20.14	25	23.36	22.44	20.97	23	23	22.09	22.09
AB	189	50.94	183.55	49.48	48	44.86	53.12	49.64	48	48	49.82	49.82
Σ	**371**	**100.00**	**370.99**	**100.00**	**107**	**100.00**	**107.00**	**99.99**	**100**	**100.00**	**100.00**	**100.00**
	χ^2=0.33 df=1 50 > p > 70				χ^2=0.99 df=1 30 > p > 50				χ^2=0.13 df=1 70 > p > 80			
Hemoglobin γ^G (HBGG)												
A	95	25.61	93.75	25.27	22	20.56	21.09	19.71	26	26	27.04	27.04
B	92	24.80	88.79	23.93	34	31.78	31.98	29.89	20	20	21.16	21.16
C	0	0.00	0.02	0.01	0	0.00	0.01	0.01	1	1	0.04	0.04
AB	178	47.98	182.47	49.18	49	45.79	51.93	48.54	51	51	47.84	47.84
AC	5	1.35	3.02	0.81	2	1.87	0.88	0.82	1	1	2.08	2.08
BC	1	0.27	2.94	0.79	0	0.00	1.09	1.02	1	1	1.84	1.84
Σ	**371**	**100.01**	**370.99**	**99.99**	**107**	**100.00**	**106.98**	**99.99**	**100**	**100.00**	**100.00**	**100.00**
	χ^2=0.24 df=1 50 > p > 70				df=0				df=0			
D7S8												
A	145	39.08	140.14	37.77	38	35.51	35.35	33.04	32	32	35.40	35.40
B	60	16.17	55.11	14.85	22	20.56	19.35	18.08	13	13	16.40	16.40
AB	166	44.74	175.76	47.37	47	43.93	52.30	48.88	55	55	48.20	48.20
Σ	**371**	**99.99**	**371.01**	**99.99**	**107**	**100.00**	**107.00**	**100.00**	**100**	**100.00**	**100.00**	**100.00**
	χ^2=1.14 df=1 20 > p > 30				χ^2=1.10 df=1 20 > p > 30				χ^2=1.99 df=1 10 > p > 20			
Group-Specific Component (GC)												
A	30	8.09	24.58	6.63	7	6.54	8.41	7.86	8	8	10.56	10.56
B	7	1.89	7.86	2.12	5	4.67	1.58	1.48	2	2	2.72	2.72
C	141	38.01	132.24	35.64	39	36.45	38.28	35.78	21	21	26.01	26.01
AB	32	8.63	27.81	7.50	6	5.61	7.29	6.81	9	9	10.73	10.73
AC	99	26.68	114.02	30.73	40	37.38	35.89	33.54	40	40	33.15	33.15
BC	62	16.71	64.50	17.38	10	9.35	15.55	14.53	20	20	16.83	16.83
Σ	**371**	**100.01**	**371.01**	**100.00**	**107**	**100.00**	**107.00**	**100.00**	**100**	**100.00**	**100.00**	**100.00**
	χ^2=4.58 df=3 20> p > 30				χ^2=3.21 df=2 20 > p > 30				χ^2=4.07 df=2 10 > p > 20			

Table 1 Polymarker kit: frequency distribution of observed and expected genotypes in the samples from Düsseldorf (n=371), Aachen (n=107) and Saarland (n=100).

Population		Germany Düsseldorf	Germany Saarland	Germany Aachen	Germany Mainz	Germany N.Bavaria	Germany Hannover	*Germany*
Genomes		742	200	214	310	300	400	*2166*
References		this study	this study	this study	Schneider et al	Hausmann et al	Rothämel et al	
Locus	Alleles							
LDLR	A	0.4501	0.4550	0.4673	0.4480	0.3770	0.3825	*0.4293*
	B	0.5499	0.5450	0.5327	0.5520	0.6230	0.6175	*0.5707*
GYPA	A	0.5512	0.5300	0.5421	0.5290	0.5870	0.5500	*0.5499*
	B	0.4488	0.4700	0.4579	0.4710	0.4130	0.4500	*0.4501*
HBGG	A	0.5027	0.5200	0.4439	0.5190	0.5000	0.5500	*0.5092*
	B	0.4892	0.4600	0.5467	0.4740	0.4830	0.4500	*0.4819*
	C	0.0081	0.0200	0.0093	0.0060	0.0170	0.0000	*0.0088*
D7S8	A	0.6146	0.5950	0.5748	0.5900	0.6000	0.5950	*0.5997*
	B	0.3854	0.4050	0.4252	0.4100	0.4000	0.4050	*0.4003*
GC	A	0.2574	0.3250	0.2804	0.3230	0.2930	0.3050	*0.2890*
	B	0.1456	0.1650	0.1215	0.1130	0.1570	0.1425	*0.1414*
	C	0.5970	0.5100	0.5981	0.5650	0.5500	0.5525	*0.5698*

Table 2 Polymarker kit: gene frequencies in German samples

	Düsseldorf				Aachen				Ulm			
	observed		expected		observed		expected		observed		expected	
	n	%	n	&	n	%	n	%	n	%	n	%
1.1-1.1	13	2.25	11.76	2.03	1q	0.94	1.70	1.59	2	1.19	2.50	1.49
1.1-1.2	36	6.22	36.77	6.35	3	2.80	5.81	5.43	5	2.98	9.27	5.52
1.1-1.3	8	1.38	8.27	1.43	0	0.00	0.38	0.35	3	1.79	3.54	2.11
1.1-2	24	4.15	20.94	3.62	8	7.48	3.66	3.42	16	9.52	6.46	3.85
1.1-3	29	5.01	24.93	4.31	4	3.74	2.90	2.71	3	1.79	4.03	2.40
1.1-4	42	7.25	50.59	8.74	10	9.35	10.85	10.14	10	5.95	12.69	7.55
1.2-1.2	31	5.35	28.74	4.96	6	5.61	4.94	4.62	9	5.36	8.60	5.12
1.2-1.3	9	1.55	12.93	2.23	1	0.94	0.64	0.60	6	3.57	6.56	3.91
1.2-2	30	5.18	32.74	5.65	5	4.67	6.23	5.83	11	6.55	11.99	7.41
1.2-3	42	7.25	38.98	6.73	5	4.67	4.95	4.62	6	3.57	7.46	4.44
1.2-4	79	13.64	79.10	13.66	20	18.69	18.49	17.28	30	17.86	23.52	14.00
1.3-1.3	0	0.00	1.45	0.25	0	0.00	0.02	0.02	3	1.79	1.25	0.74
1.3-2	9	1.55	7.36	1.27	0	0.00	0.41	0.38	3	1.79	4.57	2.72
1.3-3	10	1.73	8.77	1.51	0	0.00	0.32	0.30	1	0.60	2.85	1.70
1.3-4	22	3.80	17.79	3.07	2	1.87	1.20	1.13	10	5.95	8.97	5.34
2-2	11	1.90	9.32	1.61	1	0.94	1.97	1.84	2	1.19	4.18	2.49
2-3	20	3.45	22.20	3.83	1	0.94	3.12	2.91	7	4.17	5.20	3.10
2-4	42	7.25	45.06	7.78	13	12.15	11.65	10.89	12	7.14	16.40	9.76
3-3	12	2.07	13.22	2.28	1	0.94	1.24	1.15	1	0.60	1.62	0.96
3-4	50	8.64	53.65	9.27	11	10.28	9.25	8.64	14	8.33	10.21	6.08
4-4	60	10.36	54.43	9.40	15	14.02	17.28	16.15	14	8.33	16.10	9.59
Σ	**579**	**99.98**	**579.00**	**99.98**	**107**	**100.03**	**107.01**	**100.00**	**168**	**100.02**	**167.97**	**100.01**
	χ^2=7.39 df=14 90 > p > 95				χ^2=6.25 df= 5 20 > p > 30				χ^2=25.65 df=11 0.1 > p > 1			

Table 3 HLA-DQα:Observed and expected genotype frequencies in Düsseldorf, Aachen and Ulm (this study)

	Alleles			1.1	1.2	1.3	2	3	4
	Population	Ref.	Genomes						
1	Germany Düsseldorf	this study	1158	0.1425	0.2228	0.0501	0.1269	0.1511	0.3066
2	Germany Wuppertal	Huckenbeck et al	298	0.1409	0.2315	0.0772	0.0906	0.1678	0.2920
	Germany Aachen	this study	214	0.1262	0.2150	0.0140	0.1355	0.1075	0.4019
	Germany Ulm	this study	336	0.1220	0.2262	0.0863	0.1577	0.0982	0.3095
3	Germany Mainz	Schneider et al	318	0.154	0.230	0.069	0.104	0.135	0.308
4	Germany Southwest	Reinhold et al	604	0.140	0.210	0.093	0.136	0.112	0.308
5	Germany Munich	Weichhold et al	426	0.106	0.185	0.080	0.134	0.160	0.336
	Germany		***3018***	***0.1368***	***0.2160***	***0.0650***	***0.1244***	***0.1414***	***0.3165***

Table 4 HLA-DQα system: gene frequencies in Germany; ***Germany***: pooled data without Ulm

References

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