

# **CARATTERISTICHE DEGLI ALLELI HLA-A, -B, -C E -DRB1 E DEGLI APTOTIPI DELLA POPOLAZIONE ITALIANA NAZIONALE E REGIONALE ATTRAVERSO L'ANALISI DEI DATI HLA DI 120.926 DONATORI ISCRITTI ALL'IBMDR**

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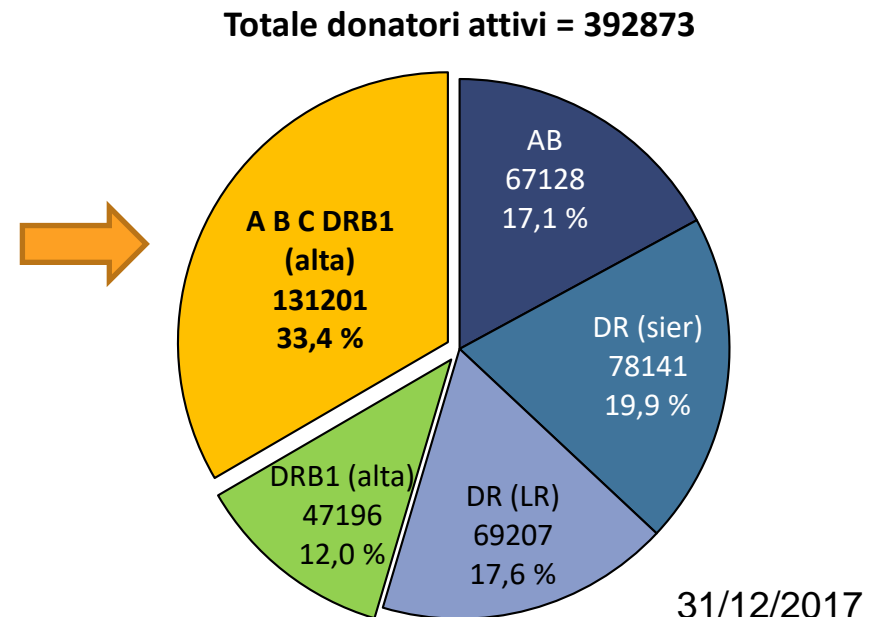
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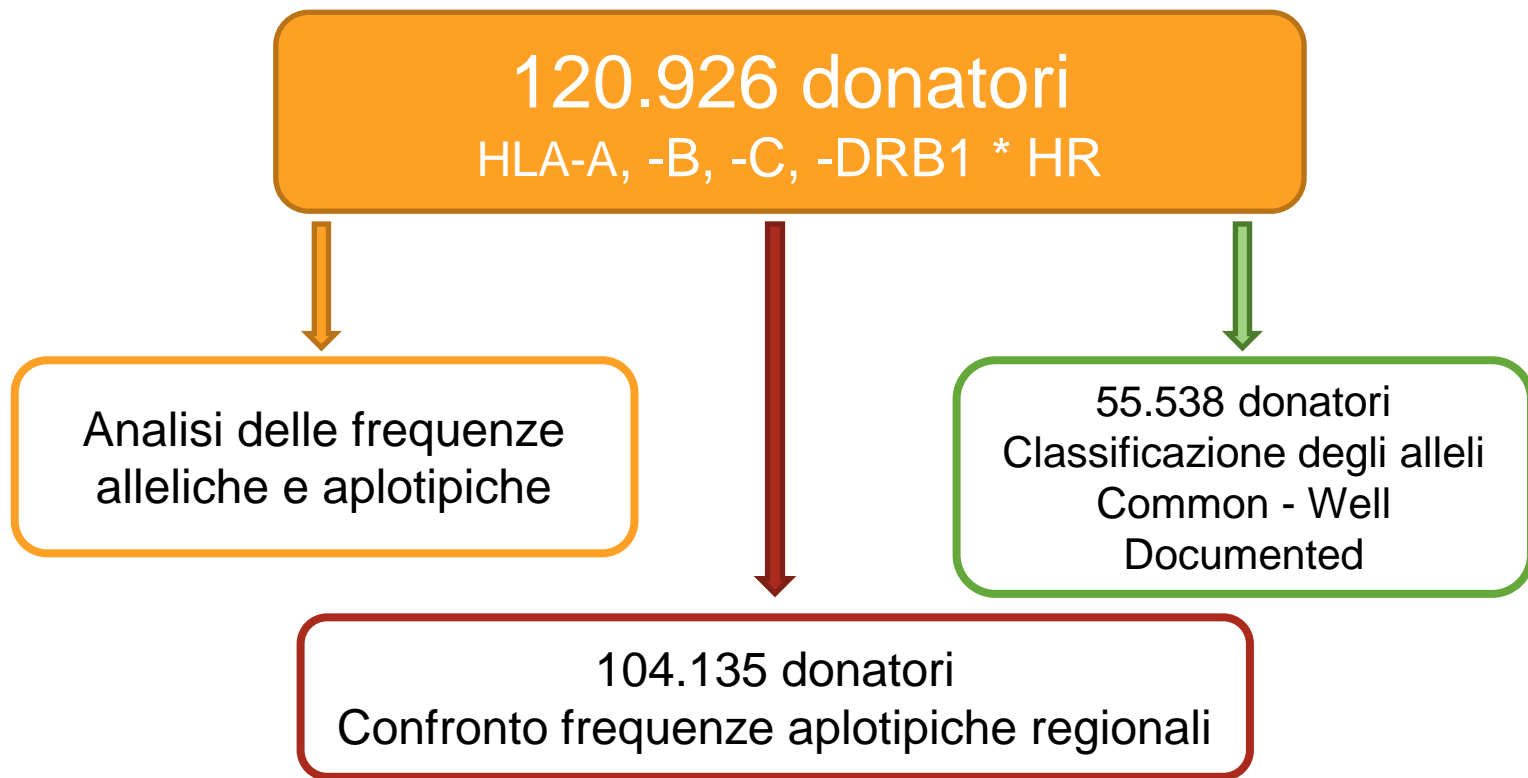
HLA polymorphisms in Italian bone marrow donors: a regional analysis. (1998).

Le caratteristiche genotipiche ed allotipiche del sistema HLA della popolazione Italiana: analisi di 370.000 donatori volontari di cellule staminali emopoietiche iscritti all'IBMDR (2010).

Estimation of human leukocyte antigen class I and class II high-resolution allele and haplotype frequencies in the Italian population and comparison with other European populations (2012).



# Caratteristiche HLA-A, -B, -C e -DRB1 della popolazione italiana



	Sample Size	Gender	Age				Median (25th-75th)
		Males (%)	18-25	26-35	36-45	46-55	
<b>Dataset 1</b>	120,926	45.6	44,466 (36.8%)	49,582 (41.0%)	23,760 (19.6%)	3,118 (2.6%)	28 (23 - 35)
<b>Dataset 2</b>	104,135	45.9	37,459 (36.0%)	43,254 (41.5%)	20,761 (19.9%)	2,661 (2.6%)	28 (24 - 35)
<b>Dataset 3</b>	55,538	46.8	18,496 (33.3%)	22,441 (40.4%)	12,167 (21.9%)	2,434 (4.4%)	29 (24 - 36)

## I primi 20 alleli più frequenti

Rank	HLA-A		HLA-B		HLA-C		HLA-DRB1	
	Allele	Freq(%)	Allele	Freq(%)	Allele	Freq(%)	Allele	Freq(%)
1	02:01g	22.820	51:01g	9.798	04:01g	17.526	07:01g	12.525
2	24:02g	12.286	18:01g	9.521	07:01g	17.108	11:01g	11.632
3	01:01g	11.528	35:01g	8.000	06:02g	9.921	11:04g	10.067
4	03:01g	10.628	08:01g	5.760	12:03g	9.028	03:01g	9.472
5	11:01g	5.973	07:02a	5.239	07:02a	6.582	01:01g	5.987
6	32:01g	5.874					01:01g	5.874
7	26:01g	5.389					01:01g	5.389
8	68:01g	5.135					01:01g	5.135
9	30:01g	4.957					01:01g	4.957
10	23:01g	4.937					02:02g	4.937
11	29:02g	2.447					02:02g	2.447
12	31:01g	2.017					01:01g	2.017
13	30:02g	1.765					01:01g	1.765
14	02:05g	1.642					03:03g	1.642
15	33:01g	1.636					04:01g	1.636
16	25:01g	1.645	58:01g	1.976	17:01g	1.488	11:03g	1.572
17	68:02g	0.802	50:01g	1.954	07:04g	1.253	15:02g	1.396
18	03:02g	0.792	55:01g	1.936	16:02g	1.114	04:02g	1.381
19	29:01g	0.758	39:01g	1.925	15:05g	0.915	04:05g	1.379
20	33:03g	0.623	52:01g	1.628	03:02g	0.689	12:01g	1.335

HLA-A: 02:01g (22,8%) 24:02g (12,3%) 01:01g (11,6%)

HLA-B: 51:01g (9,8%) 18:01g (9,5%) 35:01g (8,0%)

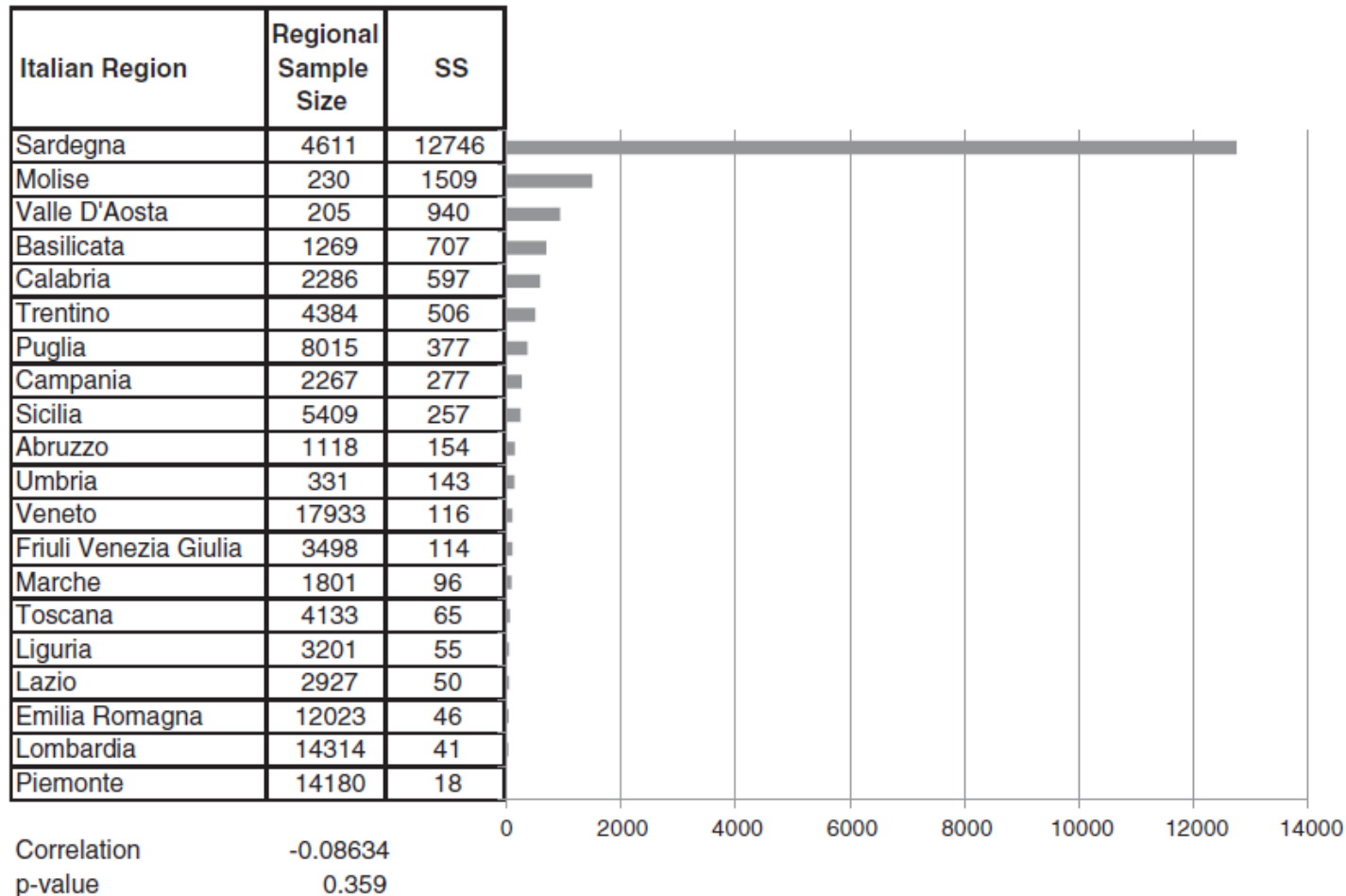
HLA-C: 04:01g (17,5%) 07:01g (17,1%) 06:02g (9,9%)

HLA-DRB1: 07:01g (12,5%) 11:01g (11,6%) 11:04g (10,1%)

## Aplotipi più frequenti

Rank	HLA Haplotype				Frequency X 1000
	A	B	C	DRB1	
1	01:01g	08:01g	07:01g	03:01g	0.025357
2	02:01g	18:01g	07:01g	11:04g	0.011435
3	30:01g	13:02g	06:02g	07:01g	0.010879
4	29:02g	44:03g	16:01g	07:01g	0.010829
5	03:01g	07:02g	07:02g	15:01g	0.010167
6	33:01g	14:02g	08:02g	01:02g	0.009459
7	24:02g	35:02g	04:01g	11:04g	0.009276
8	02:02g	12:01g	05:01g	03:01g	0.008925
9					0.007673
10	01:01g - 08:01g - 07:01g - 03:01g (25,4 x1000)				0.006078
11	02:01g - 18:01g - 07:01g - 11:04g (11,4 x1000)				0.005041
12	30:01g - 13:02g - 06:02g - 07:01g (19,9 x1000)				0.004771
13					0.004577
14					0.004511
15					0.004326
16	11:01g	35:01g	04:01g	14:01g	0.004191
17	24:02g	18:01g	12:03g	11:04g	0.004070
18	02:01g	18:01g	05:01g	03:01g	0.003987
19	02:05g	50:01g	06:02g	07:01g	0.003841
20	02:01g	08:01g	07:01g	03:01g	0.003554

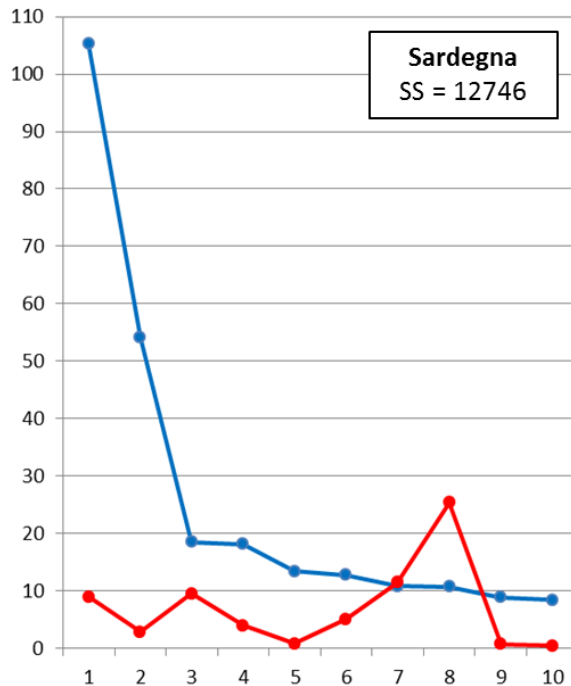
## Confronto frequenze aplotipiche regionali



Distributions of the 10 most common HLA four-locus haplotypes in the Italian geographic regions with a SS (Sum of Squares) > 500.

(Correlation SS /Sample Size -0,086; p-value 0,359)

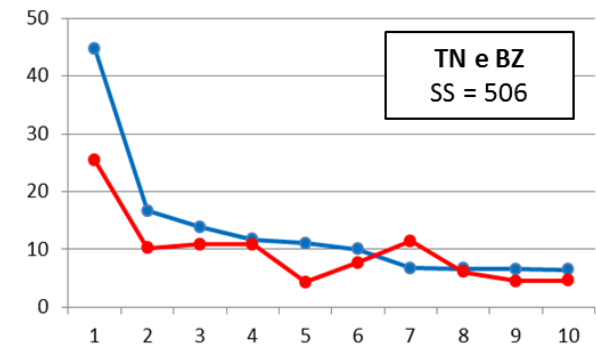
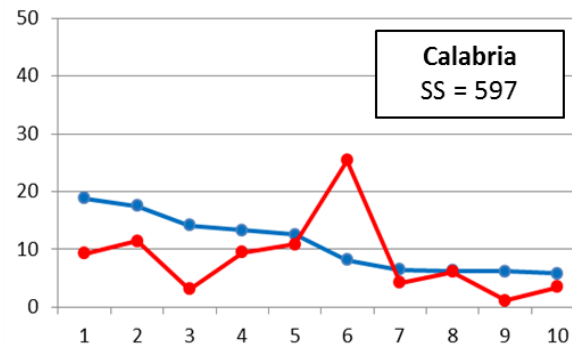
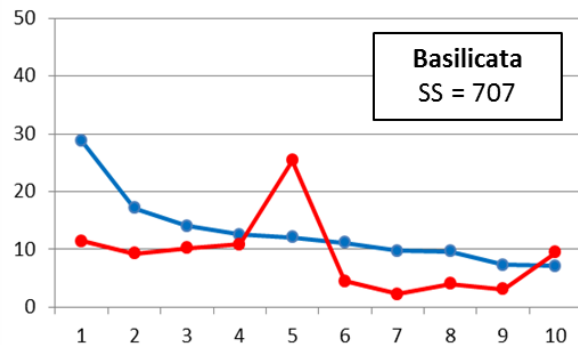
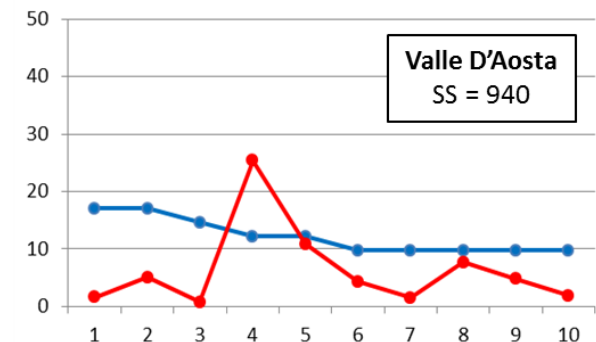
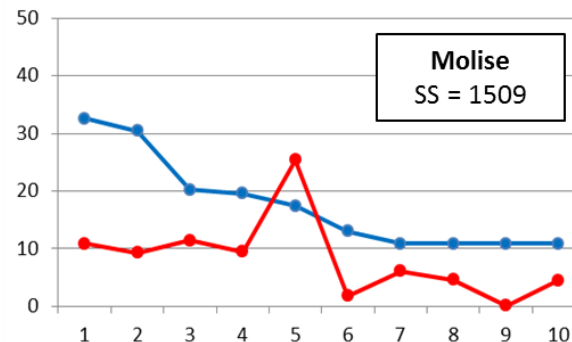
## Confronto frequenze aplotipiche regionali



Distributions of the 10 most common HLA four-locus haplotypes in the Italian geographic regions with a SS (Sum of Squares) > 500.

—●— Region  
—●— Italian national data

(Correlation SS /Sample Size -0,086; p-value 0,359)



## Alleli Common e Well Documented (CWD)

Locus	C	WD	no CWD	CWD	Total number of HLA alleles	% CWD
A	30	38	84	68	152	44.7
B	50	72	126	122	248	49.2
C	24	33	77	57	134	42.5
DRB1	35	36	87	71	158	44.9
Total	139	179	374	318	692	46.0



Number of common (C) and well-documented (WD) alleles (CWD) in 55,538 Italian individuals

54% no CWD



## Alleli Common e Well Documented (CWD)

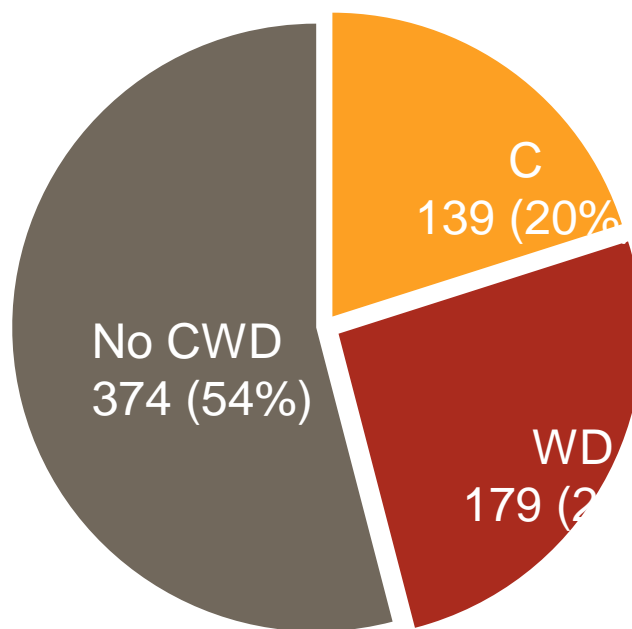
			EFI CWD catalogue		
	Italian CWD according to ASHI		C	WD	No CWD
Overall	C	139	132	7	0
	WD	179	35	75	69
	No CWD	374	5	125	244
	Total	692	172	207	313
Locus A	C	30	28	2	0
	WD	38	5	17	16
	No CWD	84	2	34	48
	Total	152	35	53	64
Locus B	C	50	46	4	0
	WD	72	15	32	25
	No CWD	126	1	42	83
	Total	248	62	78	108
Locus C	C	24	23	1	0
	WD	33	7	13	13
	No CWD	77	2	25	50
	Total	134	32	39	63
Locus DRB1	C	35	35	0	0
	WD	36	8	13	15
	No CWD	87	0	24	63
	Total	158	43	37	78

38%

Comparison between the numbers of Italian common (C) and well-documented (WD) alleles (CWD), ASHI criteria vs. EFI catalogue

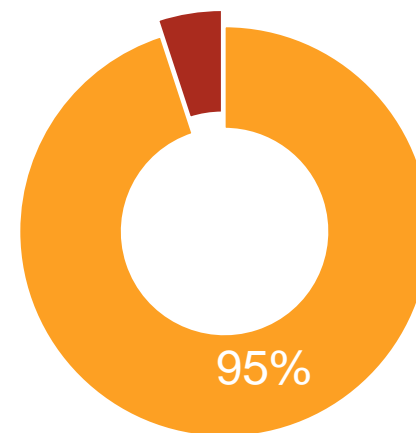
## Alleli Common e Well Documented (CWD)

Italian CWD  
According to ASHI

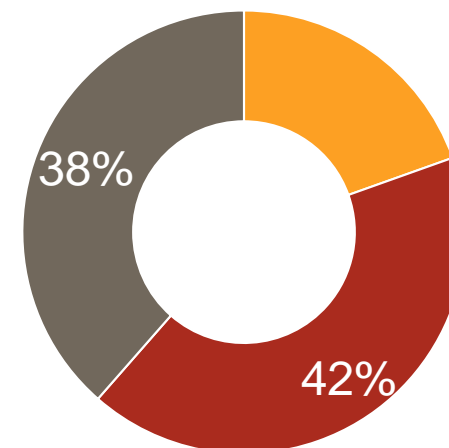


■ C ■ WD ■ No CWD

Catalogo EFI



Catalogo EFI



Catalogo EFI

