



# DESAFÍO DE PARENTESCO AVANZADO

XVII Jornadas del GHEP-ISFG

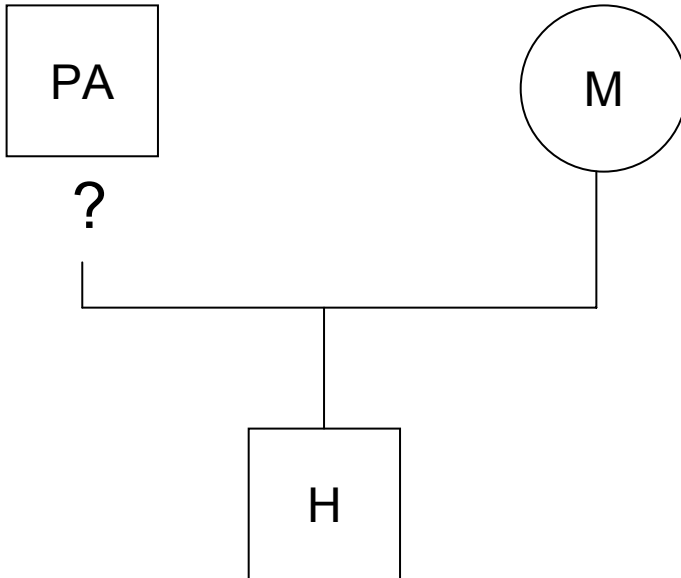
San Andres – Colombia

4-6 junio de 2012

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*Argentina*

# Caso propuesto: trío típico



Marcador (Marker)	PA (AF)	H (Ch)	M (M)
CSF1PO	12	12-13	12-13
D13S317	9-12	8-9	8-12
D16S539	9-10	9-14	12-14
D18S51	14	12-14	12-16
D21S11	32-33,2	32-32,2	28-32,2
D3S1358	15	16	15
D5S818	10-11	9-10	9-11
D7S820	10-11	9-11	9-11
D8S1179	12-14	13-14	13
FGA	24	23-24	23-25
Penta_E	16-20	11-16	11-14
Penta_D	9-13	9-13	11-13
TH01	7	6-7	6-7
TPOX	10-11	8-11	8
VWA	16-18	18	16-18
Amelogenina	XY	XY	XX

1. Son estos resultados consistentes con Paternidad Biológica?
2. Indicar los LR para cada marcador

# LR por marcador

Marcador	PA	H	M	X	Y	LR
CSF1PO	12	12-13	12-13	$1 \cdot \frac{1}{2} + 0 \cdot \frac{1}{2}$	$f_{12} \cdot \frac{1}{2} + f_{13} \cdot \frac{1}{2}$	$1/(f_{12}+f_{13})$
TH01	7	6-7	6-7	$1 \cdot \frac{1}{2} + 0 \cdot \frac{1}{2}$	$f_7 \cdot \frac{1}{2} + f_6 \cdot \frac{1}{2}$	$1/(f_6+f_7)$
D18S51	14	12-14	12-16	$1 \cdot \frac{1}{2} + 0 \cdot 0$	$f_{14} \cdot \frac{1}{2} + f_{12} \cdot 0$	$1/ f_{14}$
FGA	24	23-24	23-25	$1 \cdot \frac{1}{2} + 0 \cdot 0$	$f_{24} \cdot \frac{1}{2} + f_{23} \cdot 0$	$1/ f_{24}$
D7S820	10-11	9-11	9-11	$\frac{1}{2} \cdot \frac{1}{2} + 0 \cdot \frac{1}{2}$	$f_9 \cdot \frac{1}{2} + f_{11} \cdot \frac{1}{2}$	$\frac{1}{2}(f_9+f_{11})$
D13S317	9-12	8-9	8-12	$\frac{1}{2} \cdot \frac{1}{2} + 0 \cdot 0$	$f_9 \cdot \frac{1}{2} + f_8 \cdot 0$	$\frac{1}{2} f_9$
D16S539	9-10	9-14	12-14	$\frac{1}{2} \cdot \frac{1}{2} + 0 \cdot 0$	$f_9 \cdot \frac{1}{2} + f_{14} \cdot 0$	$\frac{1}{2} f_9$
D21S11	32-33,2	32-32,2	28-32,2	$\frac{1}{2} \cdot \frac{1}{2} + 0 \cdot 0$	$f_{32} \cdot \frac{1}{2} + f_{32.2} \cdot 0$	$\frac{1}{2} f_{32}$
D5S818	10-11	9-10	9-11	$\frac{1}{2} \cdot \frac{1}{2} + 0 \cdot 0$	$f_{10} \cdot \frac{1}{2} + f_9 \cdot 0$	$\frac{1}{2} f_{10}$
Penta_E	16-20	11-16	11-14	$\frac{1}{2} \cdot \frac{1}{2} + 0 \cdot 0$	$f_{16} \cdot \frac{1}{2} + f_{11} \cdot 0$	$\frac{1}{2} f_{16}$
Penta_D	9-13	9-13	11-13	$\frac{1}{2} \cdot \frac{1}{2} + \frac{1}{2} \cdot 0$	$f_9 \cdot \frac{1}{2} + f_{13} \cdot 0$	$\frac{1}{2} f_9$
D8S1179	12-14	13-14	13	$\frac{1}{2} \cdot 1 + 0 \cdot 0$	$f_{14} \cdot 1 + f_{13} \cdot 0$	$\frac{1}{2} f_{14}$
TPOX	10-11	8-11	8	$\frac{1}{2} \cdot 1 + 0 \cdot 0$	$f_{11} \cdot 1 + f_8 \cdot 0$	$\frac{1}{2} f_{11}$
VWA	16-18	18	16-18	$\frac{1}{2} \cdot \frac{1}{2}$	$f_{18} \cdot \frac{1}{2}$	$\frac{1}{2} f_{18}$

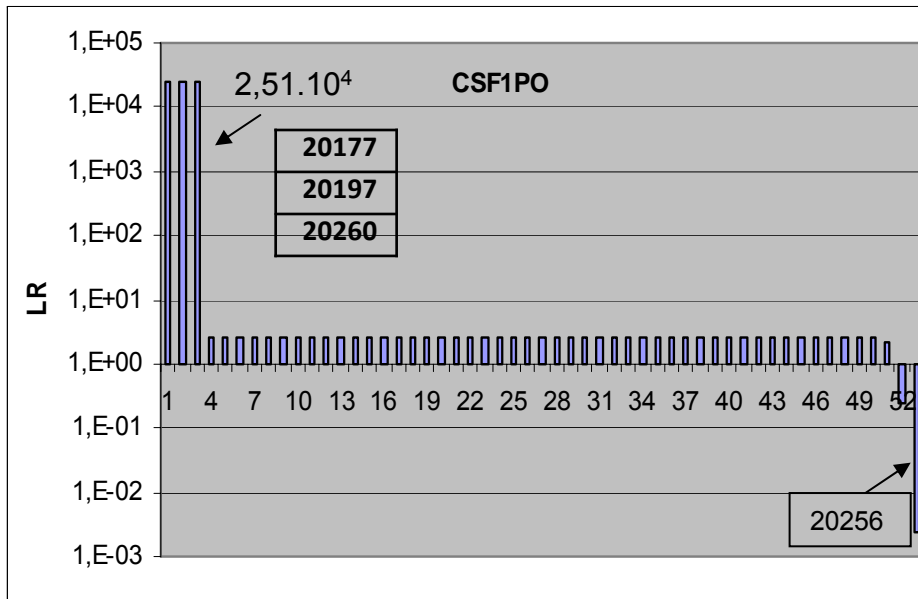
# LR por marcador

Marcador	LR	Valor
CSF1PO	$1/(f_{12}+f_{13})$	2,5189
TH01	$1/(f_6+f_7)$	2,4894
D18S51	$1/f_{14}$	6,1958
FGA	$1/f_{24}$	6,9541
D7S820	$\frac{1}{2}(f_9+f_{11})$	1,5596
D13S317	$\frac{1}{2} f_9$	7,6805
D16S539	$\frac{1}{2} f_9$	4,1220
D21S11	$\frac{1}{2} f_{32} (f_{min})$	51,54
D5S818	$\frac{1}{2} f_{10}$	7,4738
Penta_E	$\frac{1}{2} f_{16}$	10,9890
Penta_D	$\frac{1}{2} f_9$	2,3375
D8S1179	$\frac{1}{2} f_{14}$	2,3191
TPOX	$\frac{1}{2} f_{11}$	1,8025
VWA	$\frac{1}{2} f_{18}$	3,2342

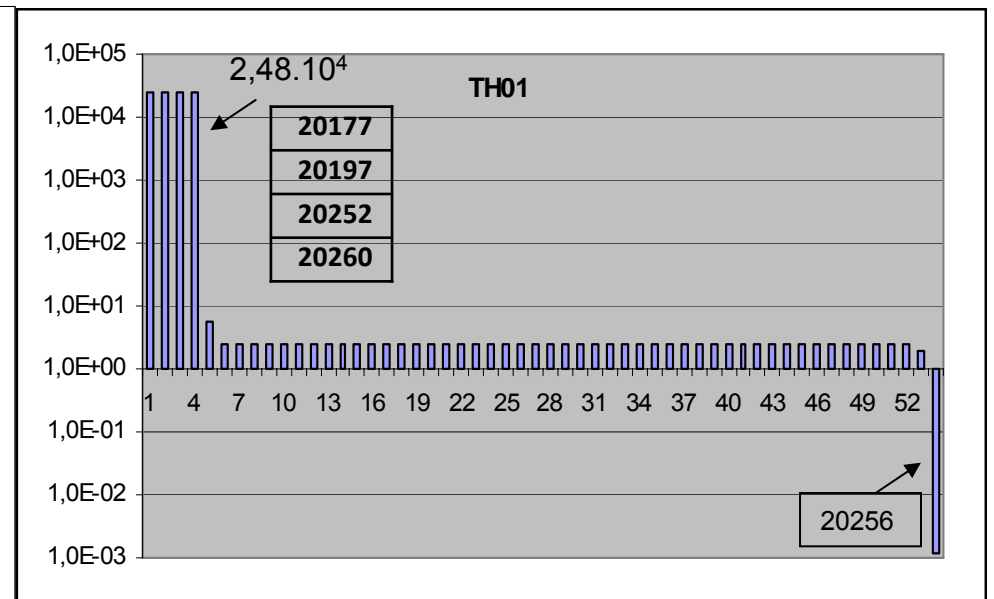
# CSF1PO y TH01

Marcador	PA	H	M	LR	Consenso	Valor correcto
CSF1PO	12	12-13	12-13	$1/(f_{12}+f_{13})$	2,5189	2,5189
TH01	7	6-7	6-7	$1/(f_6+f_7)$	2,4894	2,4894

$$2,5189 \text{ E}+04 = 25.189$$



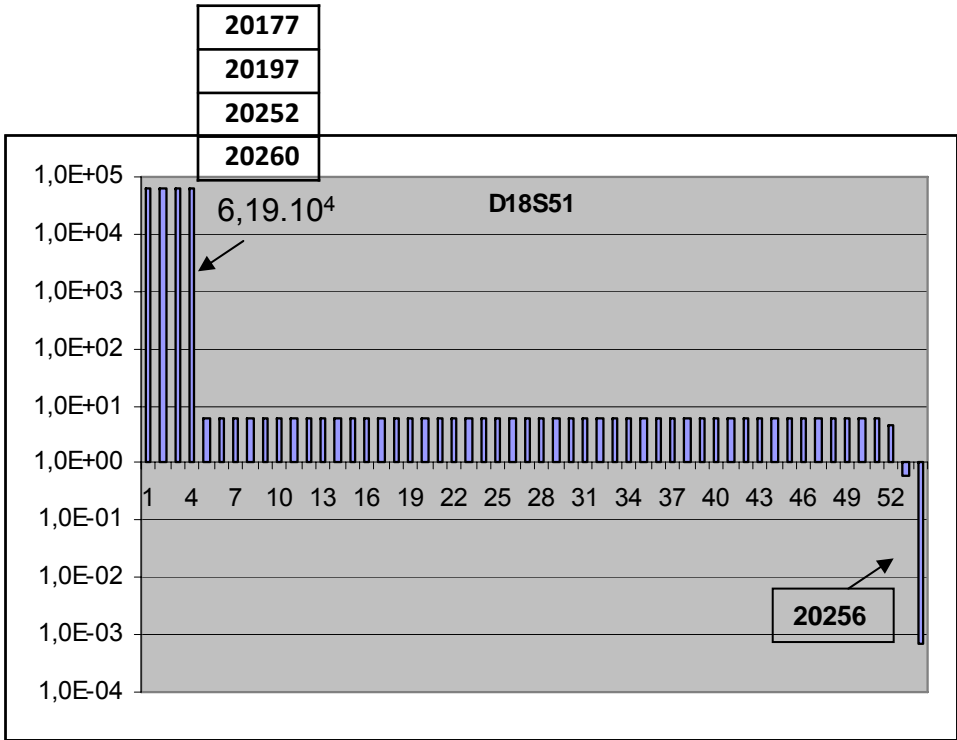
Consenso= 43/53= 81%



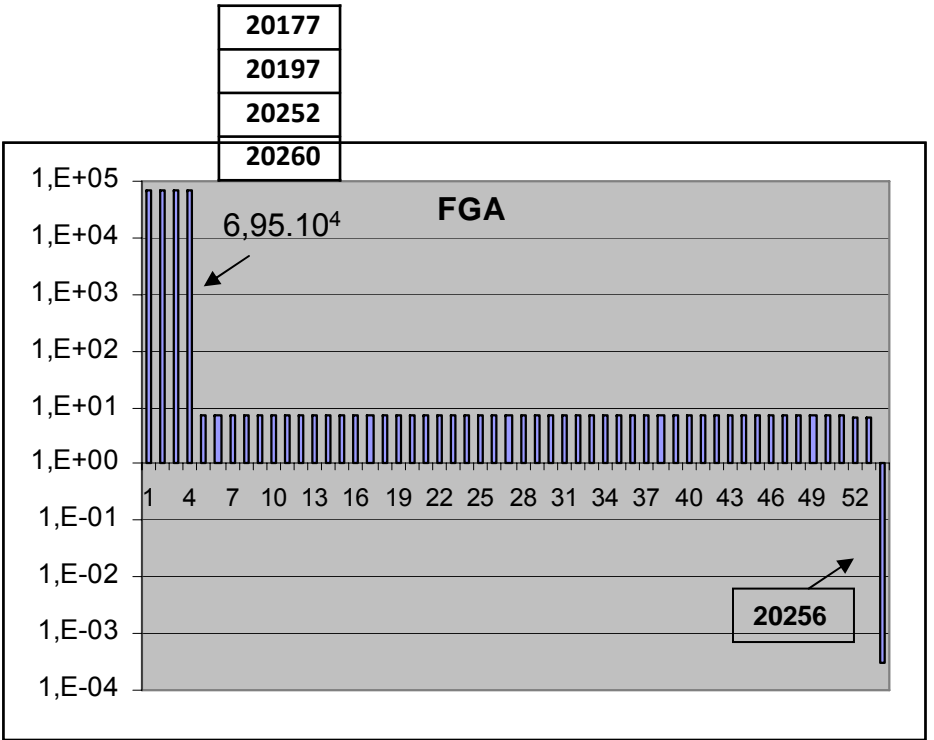
Consenso= 43/53= 81%

# D18S51 y FGA

	PA	H	M	Valor correcto	Consenso
D18S51	14	12-14	12-16	6,1958	6,1958
FGA	24	23-24	23-25	6,9541	6,9541



Consenso=  $38/53= 72\%$

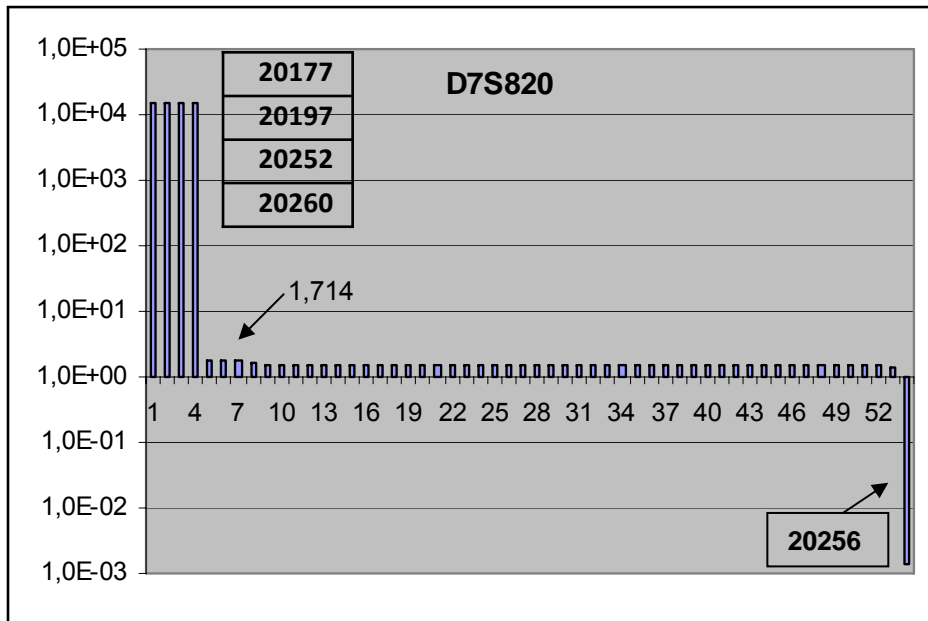


Consenso=  $48/53= 90\%$

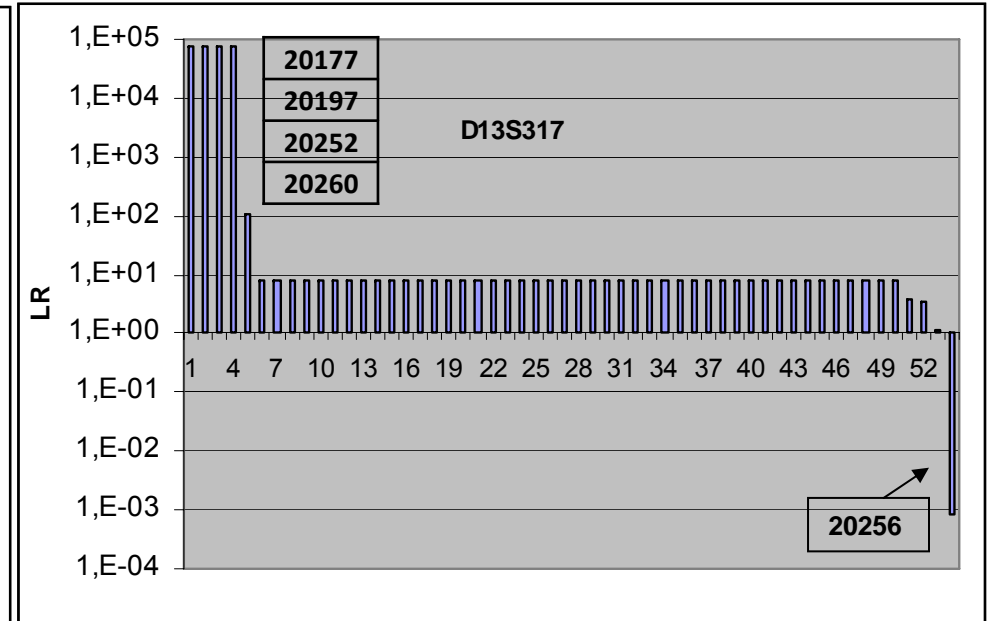
# D7S820 y D13S317

	PA	H	M	Valor correcto	Consenso
D7S820	10-11	9-11	9-11	1,5596	1,5596
D13S317	9-12	8-9	8-12	7,6805	7,6805

Consenso= 38/53= 72%



Consenso= 43/53= 81%

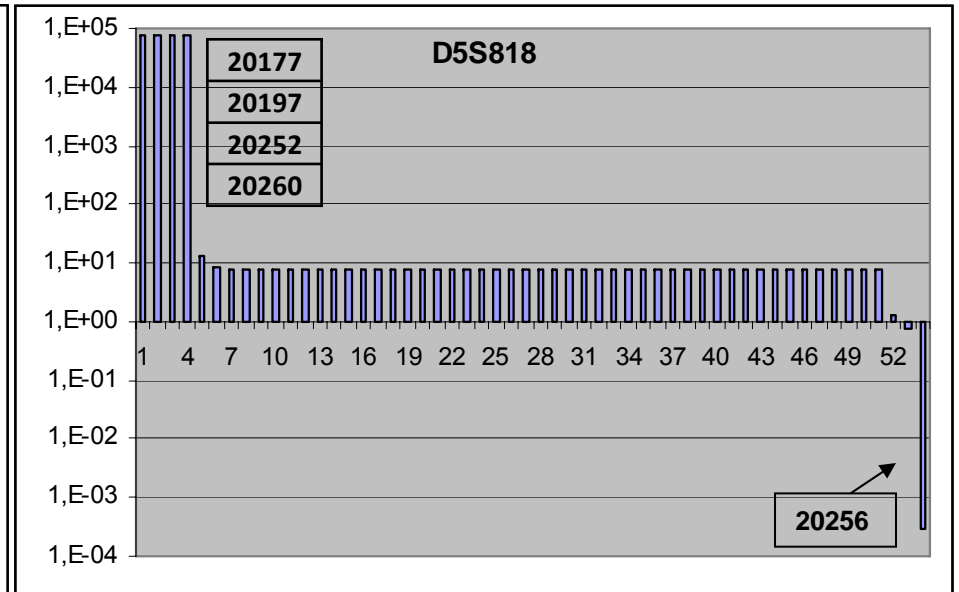
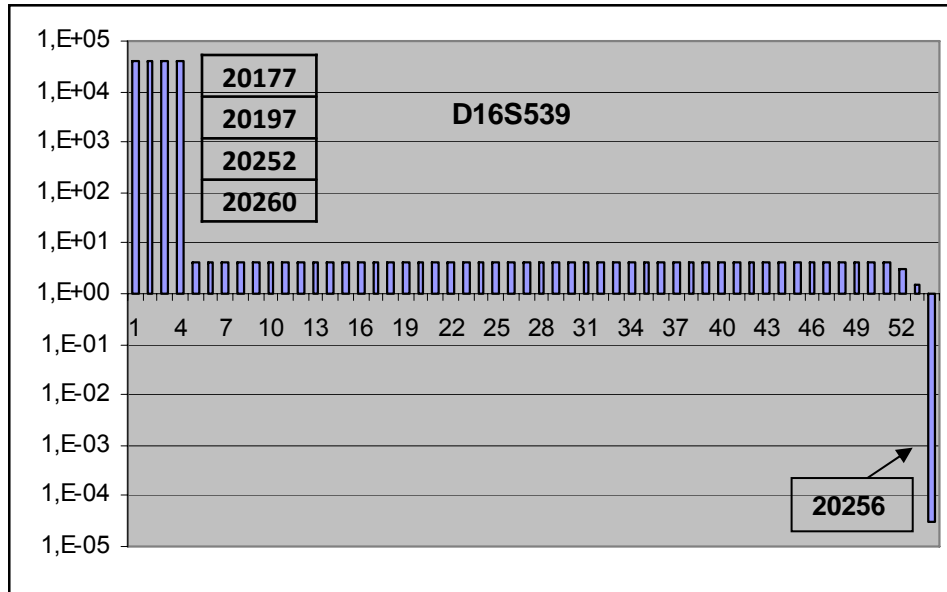


# D16S539 y D5S818

	PA	H	M	Valor correcto	Consenso
D16S539	9-10	9-14	12-14	4,1220	4,1220
D5S818	10-11	9-10	9-11	7,4738	7,4738

Consenso= 46/53= 87%

Consenso= 44/53= 83%



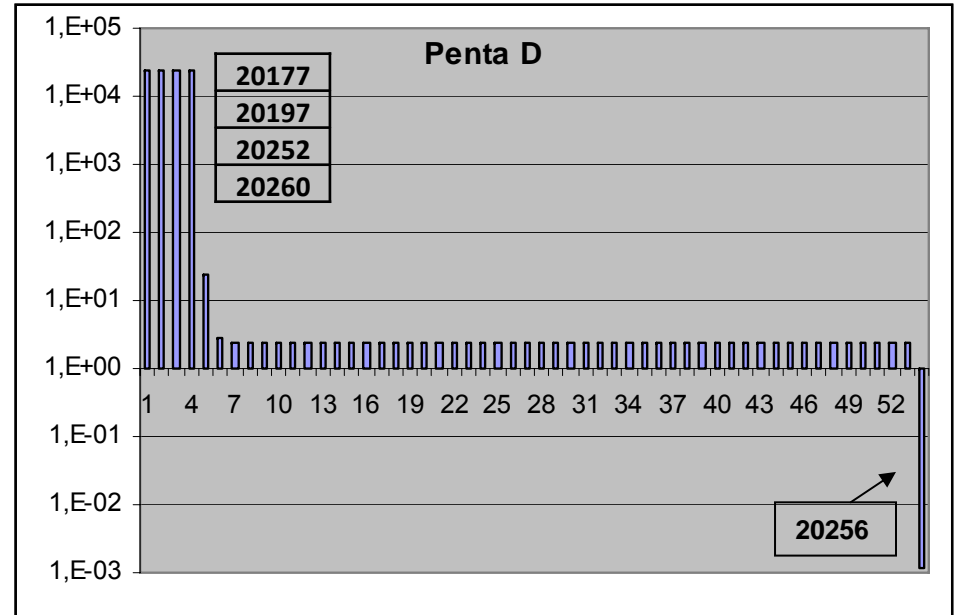
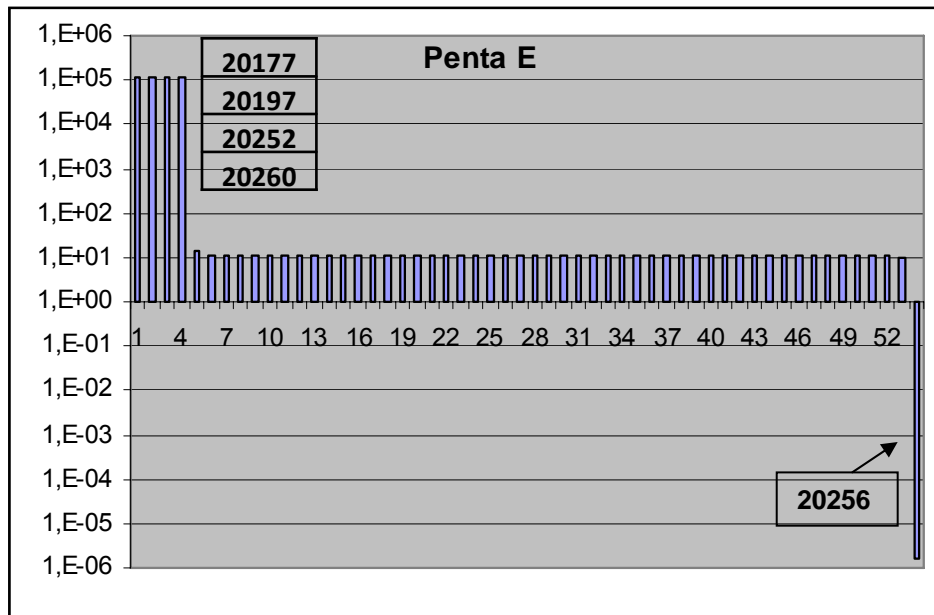


# Penta-E y Penta-D

	PA	H	M	Valor correcto	Consenso
Penta_E	16-20	11-16	11-14	10,9890	10,9890
Penta_D	9-13	9-13	11-13	2,3375	2,3375

Consenso= 46/53= 87%

Consenso= 46/53= 87%

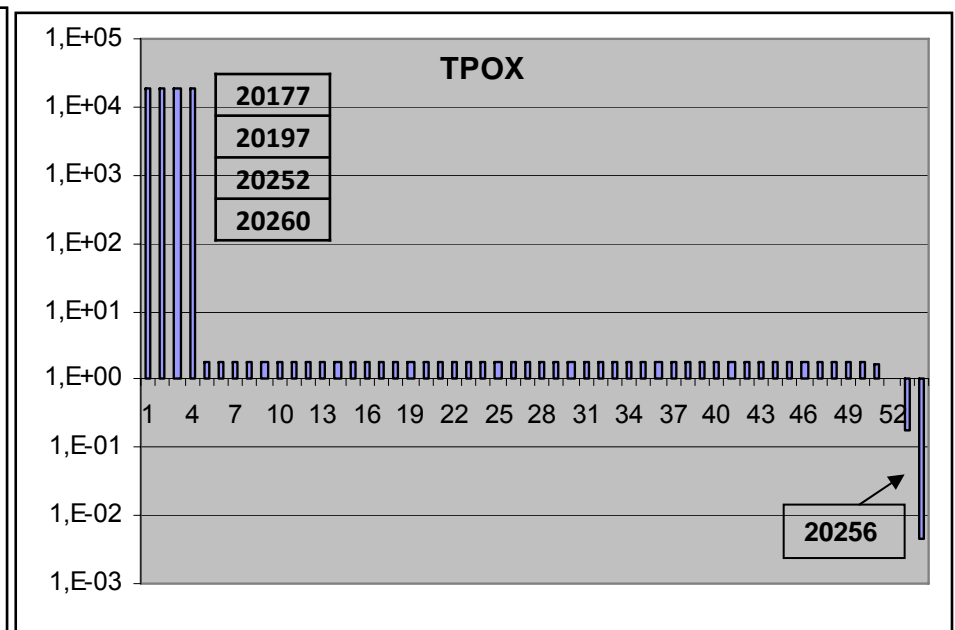
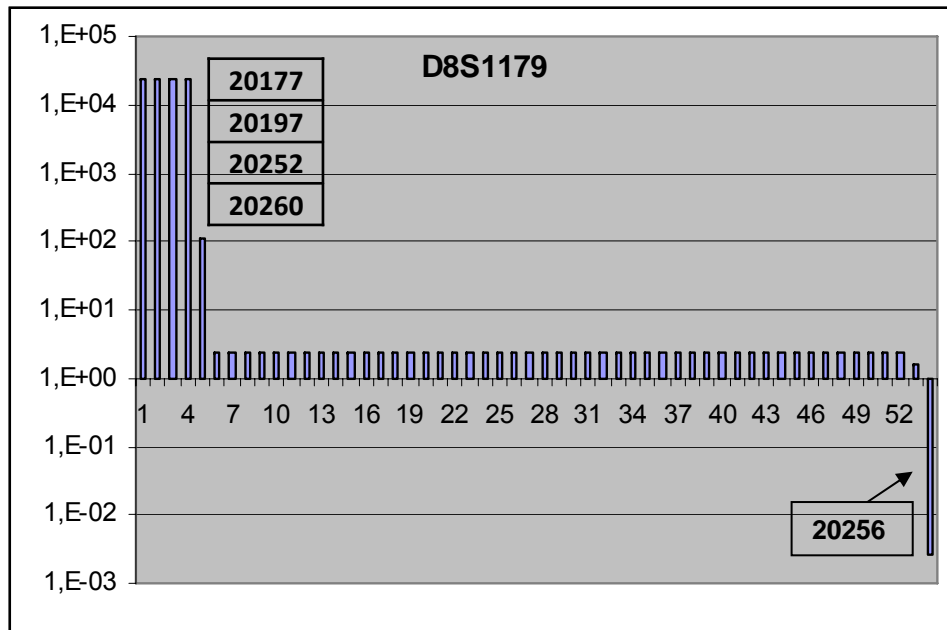


# D8S1179 y TPOX

	PA	H	M	Valor correcto	Consenso
D8S1179	12-14	13-14	13	2,3191	2,3191
TPOX	10-11	8-11	8	1,8025	1,8025

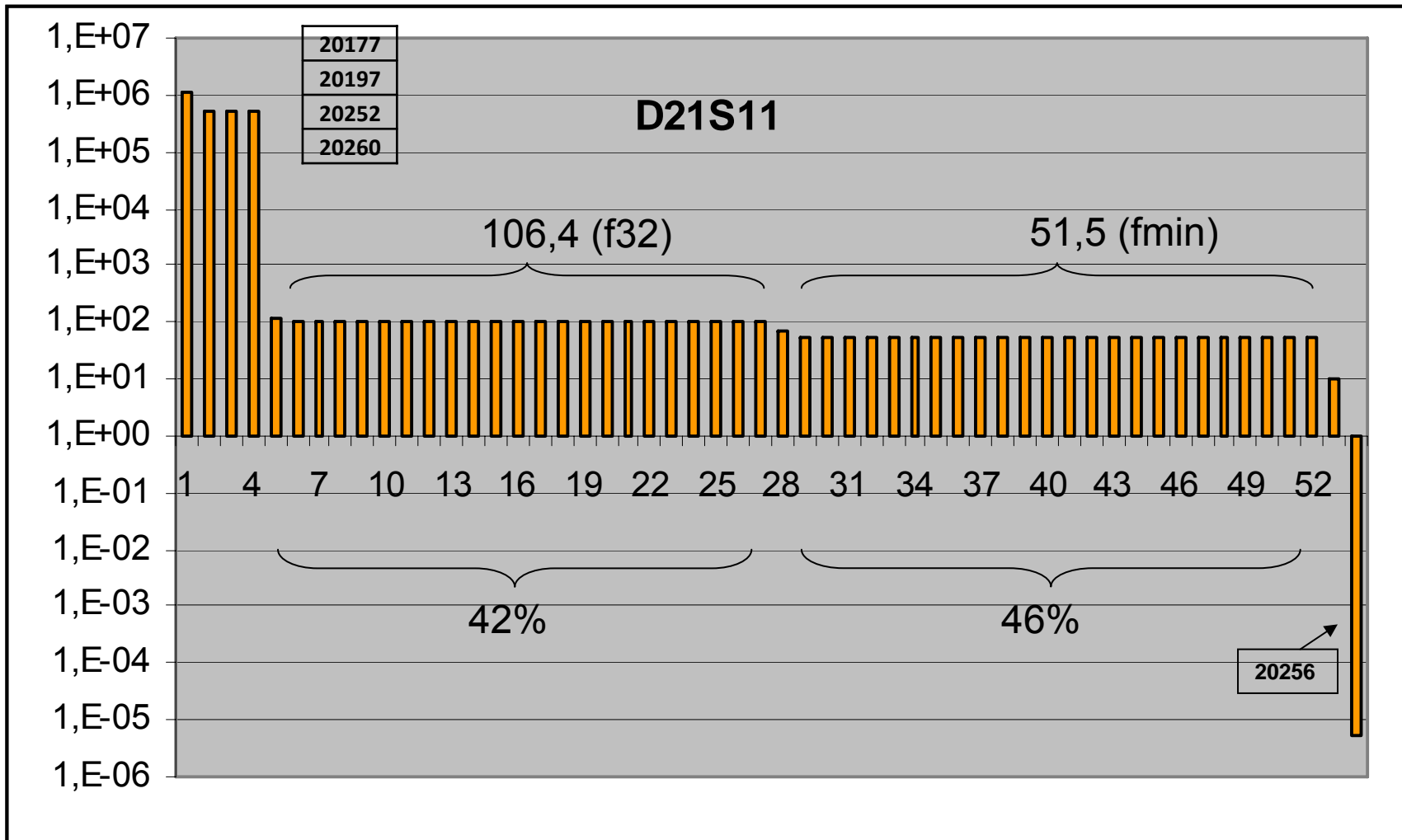
Consenso= 44/53= 83%

Consenso= 45/53= 85%



# D21S11

	PA	H	M	Valor correcto	Cons1	Cons2
D21S11	32-33,2	32-32,2	28-32,2	51,5	51,5	106,4
					1/fmin (0.0097)	1/f32 (0,0047)



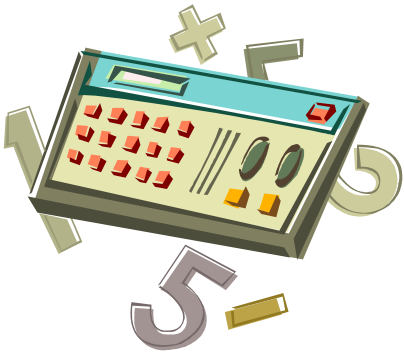
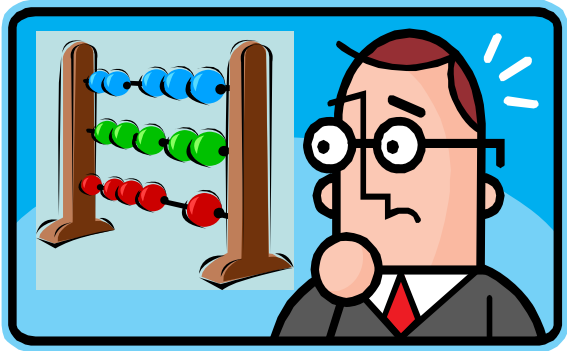
D3S1358			
	PA	H	M
D3S1358	15	16	15

PA  
15/15  
15/0

?

M  
15/15  
15/0

H



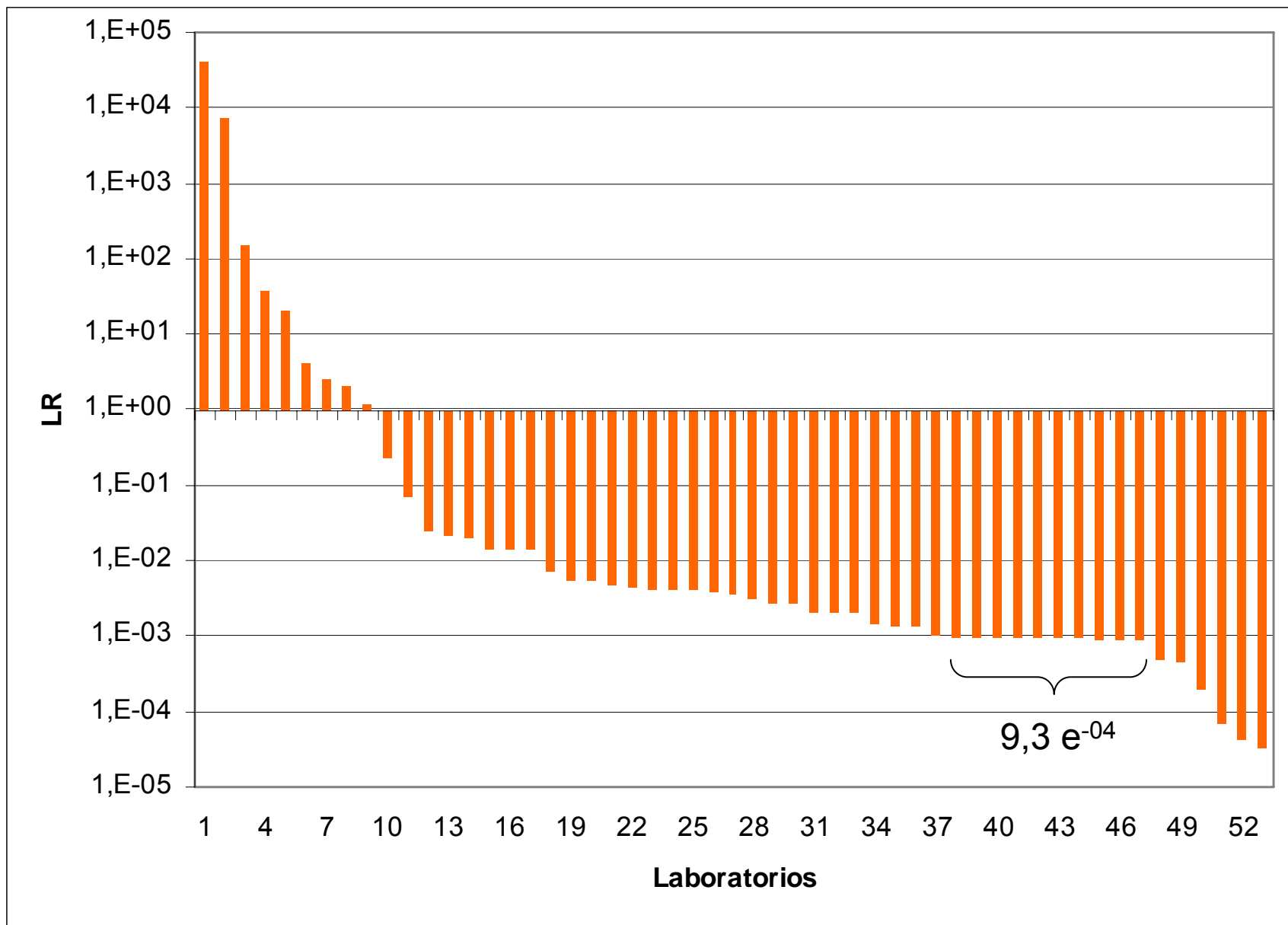
$$f_{15} = 0,2770$$

$$f_{16} = 0,2392$$

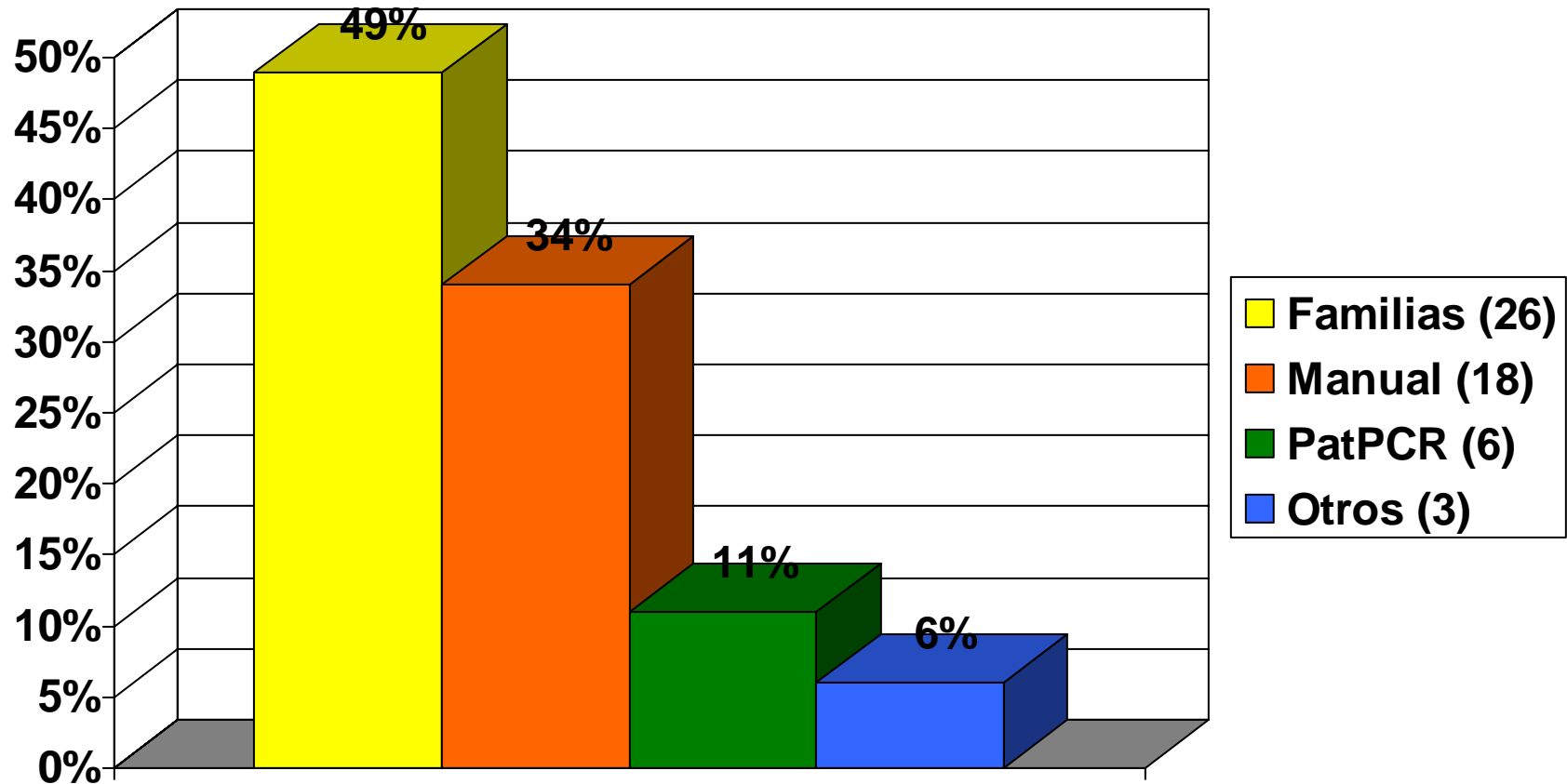
$$\mu = 0,001$$

$$f_0 = 0,005$$

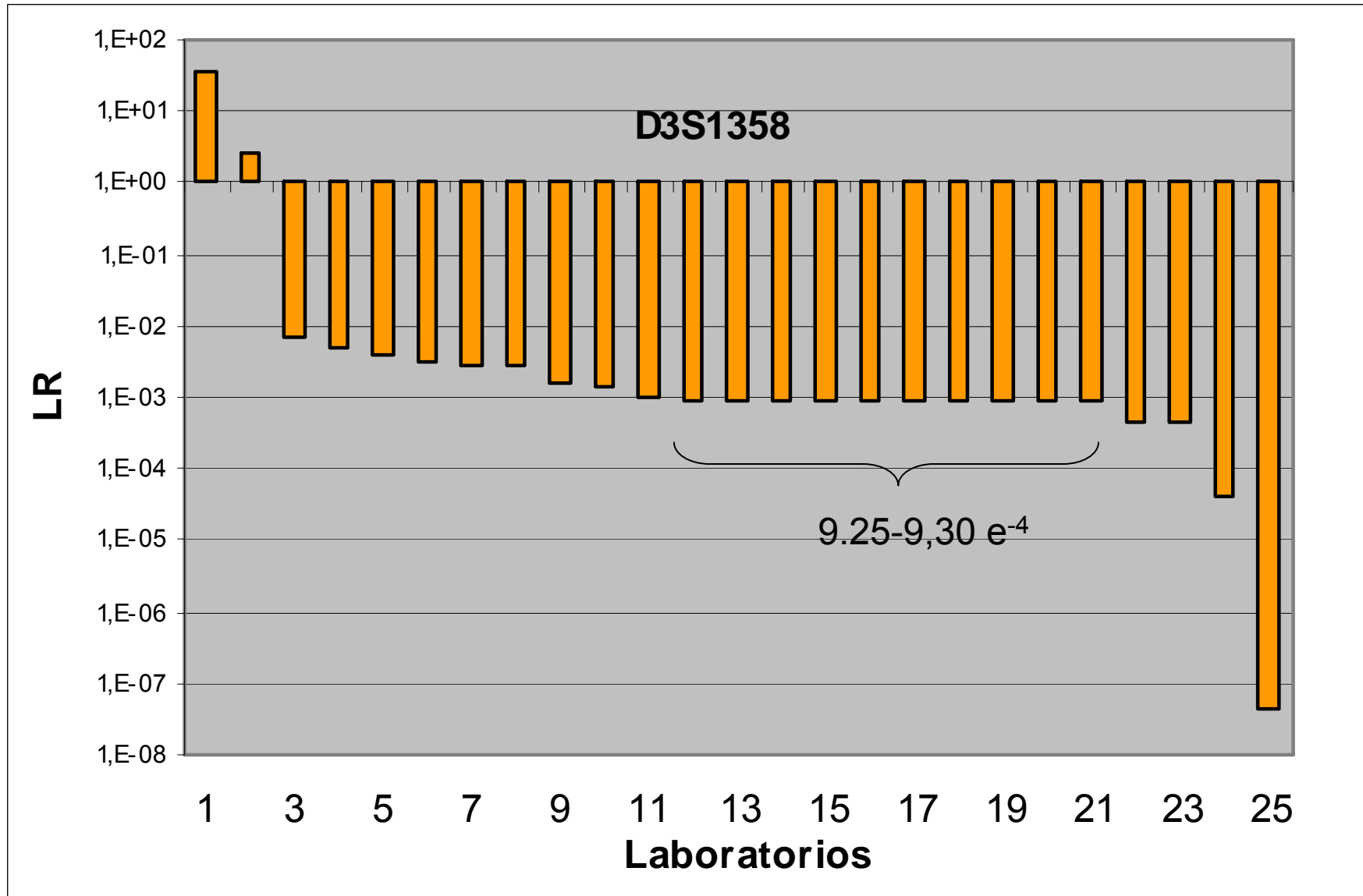
# D3S1358: Valores de LR informados



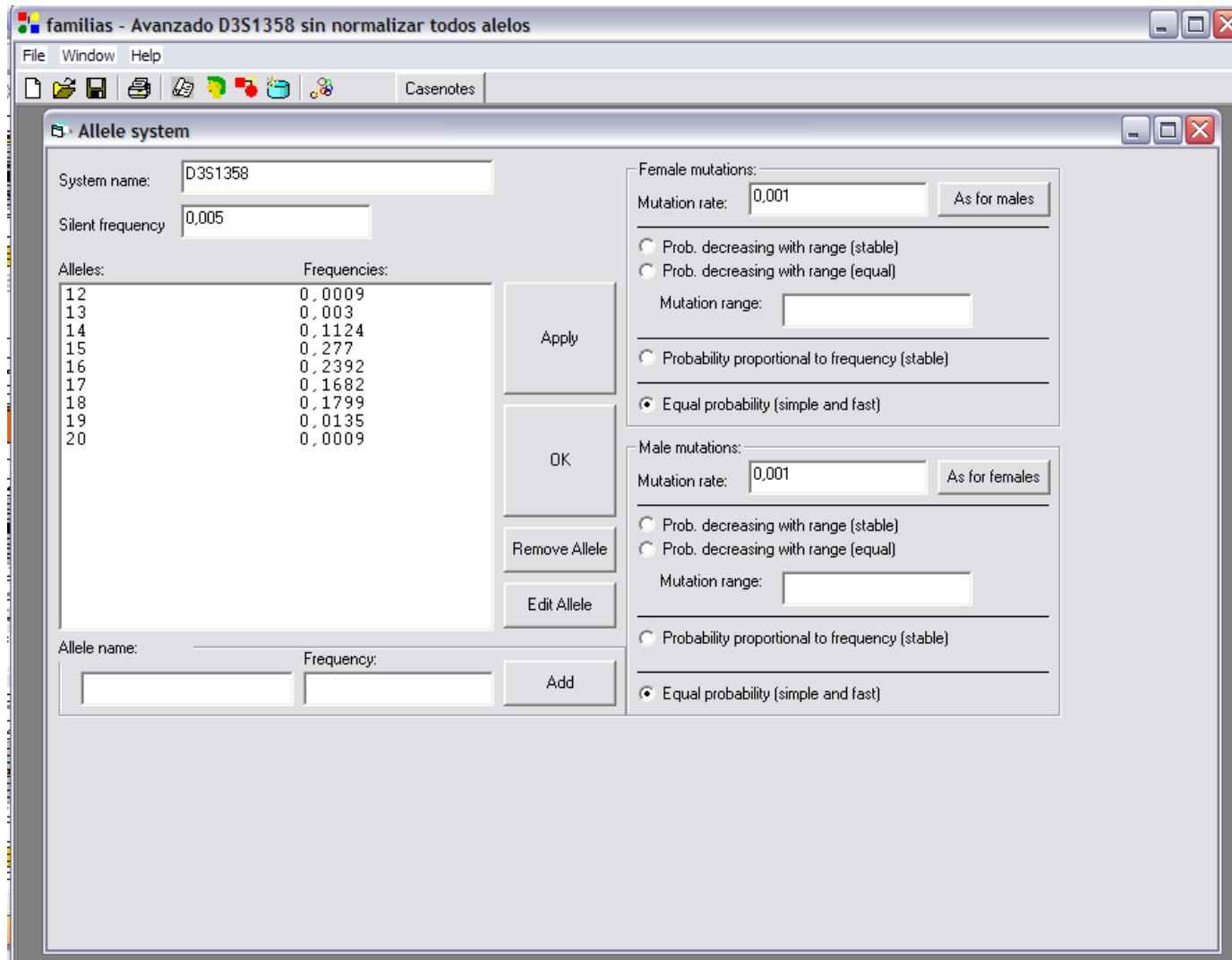
# Tratamiento Estadístico (n=53)



# LR con Software Familias

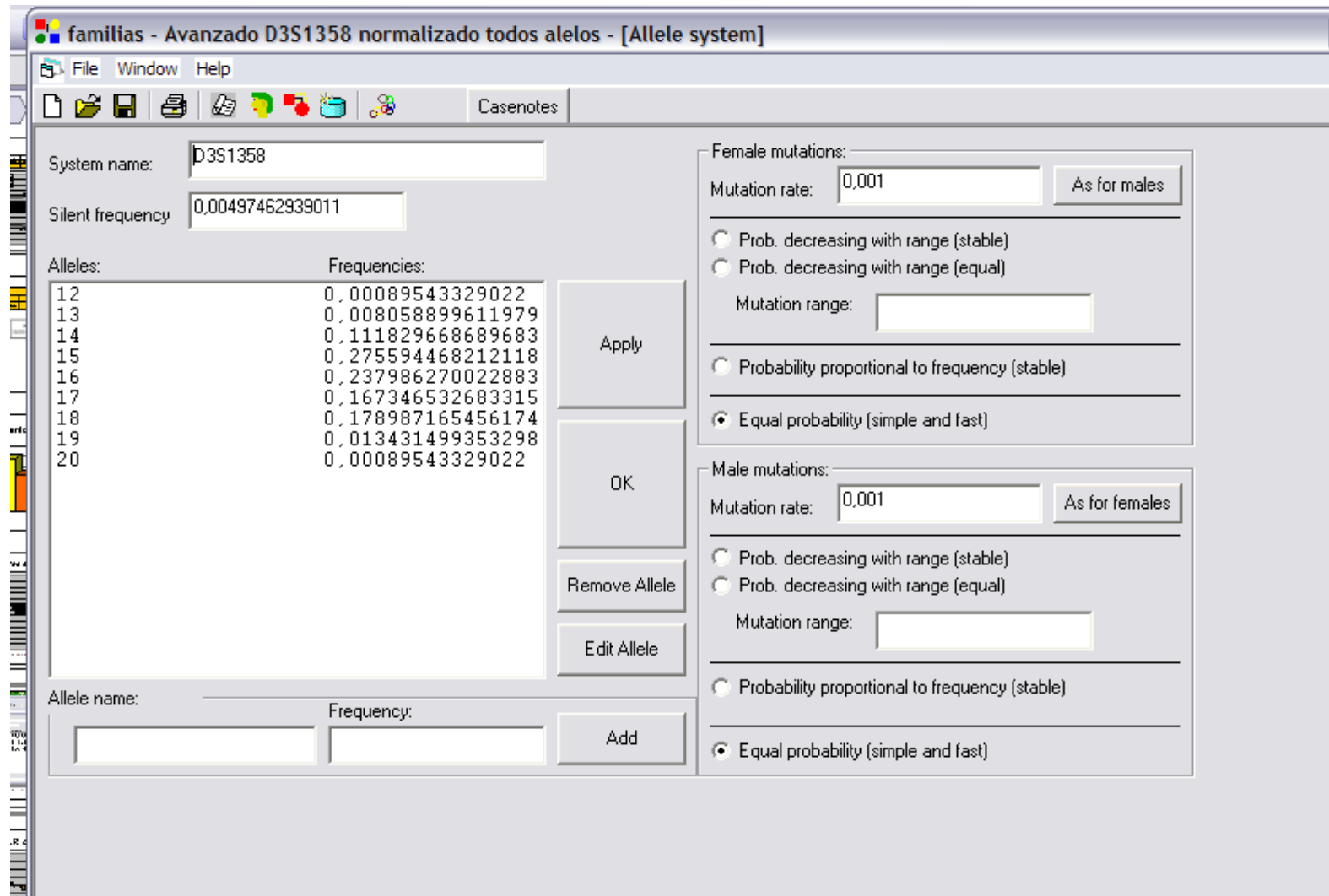


# Influencia de la normalización de frecuencias en software Familias: frec sin normalizar

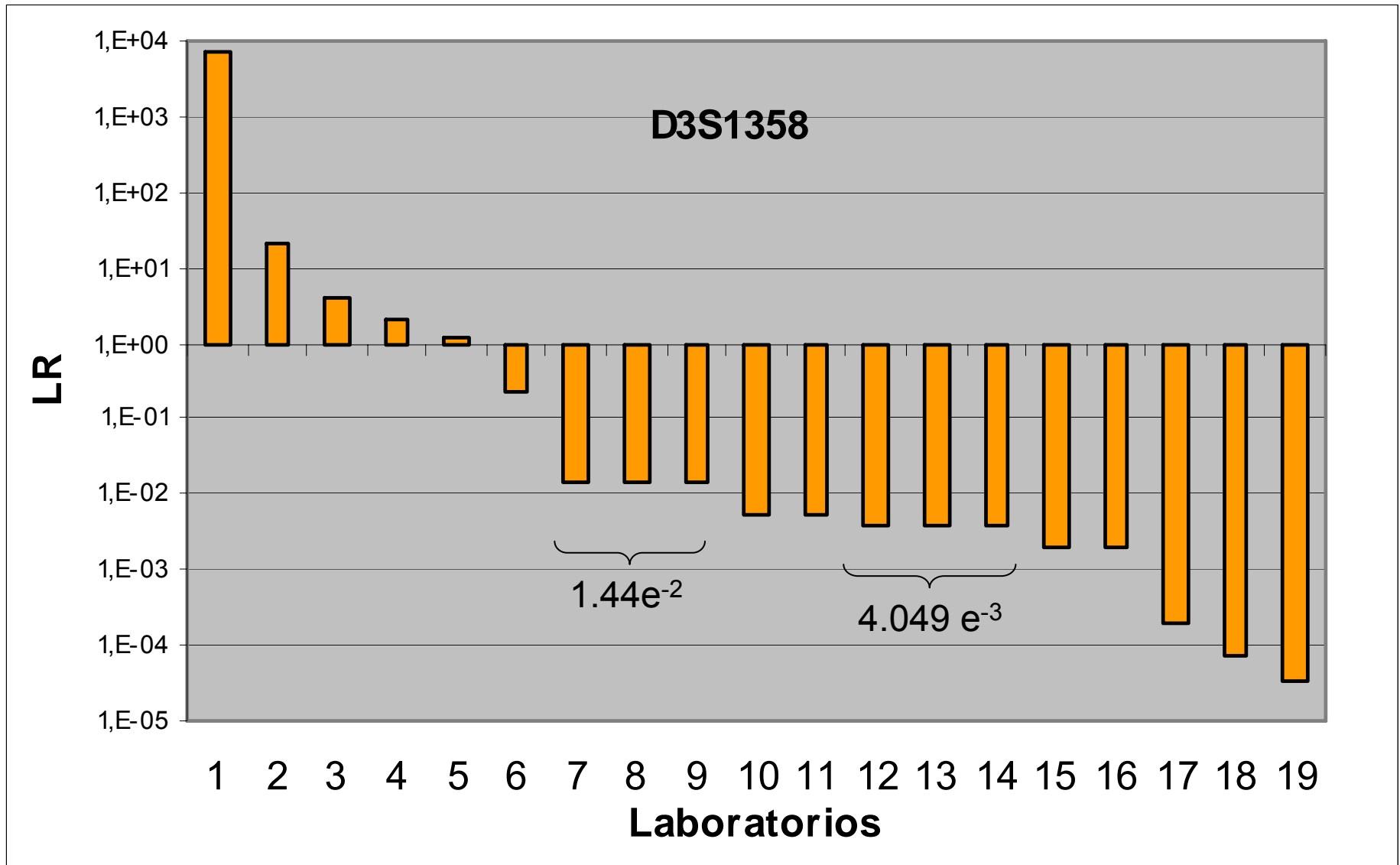




# Influencia de la normalización de frecuencias en software Familias: frec normalizadas



# LR cálculo manual



D3S1358			
	PA	H	M
D3S1358	15	16	15

Genotipos			Probabilidades
$G_{PA}$	$G_H$	$G_M$	$X = \sum (\text{Pr}G_{PA} \cdot \text{Pr}G_M) \cdot (\text{Pr}_{PA \rightarrow 1} \cdot \text{Pr}_{M \rightarrow 2})$
15/15	16/16	15/15	
15/0	16/16	15/15	
15/15	16/16	15/0	
15/0	16/16	15/0	
15/0	0/16	15/15	$Y = \sum (\text{Pr}G_{PA} \cdot \text{Pr}G_M) \cdot (\text{Pr}_{HA \rightarrow 1} \cdot \text{Pr}_{M \rightarrow 2} + \text{Pr}_{HA \rightarrow 2} \cdot \text{Pr}_{M \rightarrow 1})$
15/15	16/0	15/0	
15/0	16/0	15/0	
15/15	16/0	15/15	

Genotipos			$X = \sum (\text{Pr}G_{PA} \cdot \text{Pr}G_M \cdot (\text{Pr}_{PA \rightarrow 1} \cdot \text{Pr}_M \rightarrow 2))$	
$G_{PA}$	$G_H$	$G_M$		X
15/15	16/16	15/15	$f15^2 f15^2 (1 \cdot u/2 \cdot 1 \cdot u/2)$	$f15^4 \cdot u^2/4$
15/0	16/16	15/15	$2f15 \cdot fo \cdot f15^2 \cdot (1/2 u/2 \cdot 1 \cdot u/2)$	$2f15^3 fo \cdot u^2/8$
15/15	16/16	15/0	$f15^2 \cdot 2f15 fo (u/2 \cdot 1/2 u/2)$	$2f15^3 fo \cdot u^2/8$
15/0	16/16	15/0	$2f15 fo \cdot 2f15 fo (u/4 \cdot u/4)$	$4f15^2 fo^2 \cdot u^2/16$
15/0	0/16	15/15	$2f15 \cdot fo \cdot f15^2 (1/2 u/2 + u/4 \cdot 0)$	$2f15^3 fo \cdot u/4$
15/15	16/0	15/0	$f15^2 \cdot 2f15 fo (u/2 \cdot 1/2 + 0 \cdot u/2)$	$2f15^3 fo \cdot u/4$
15/0	16/0	15/0	$2f15 fo \cdot 2f15 fo (u/4 \cdot 1/2 + 1/2 \cdot u/4)$	$4f15^2 fo^2 \cdot u/4$
15/15	16/0	15/15	$f15^2 f15^2 (u/2 \cdot 0 + 0 \cdot u/2)$	0

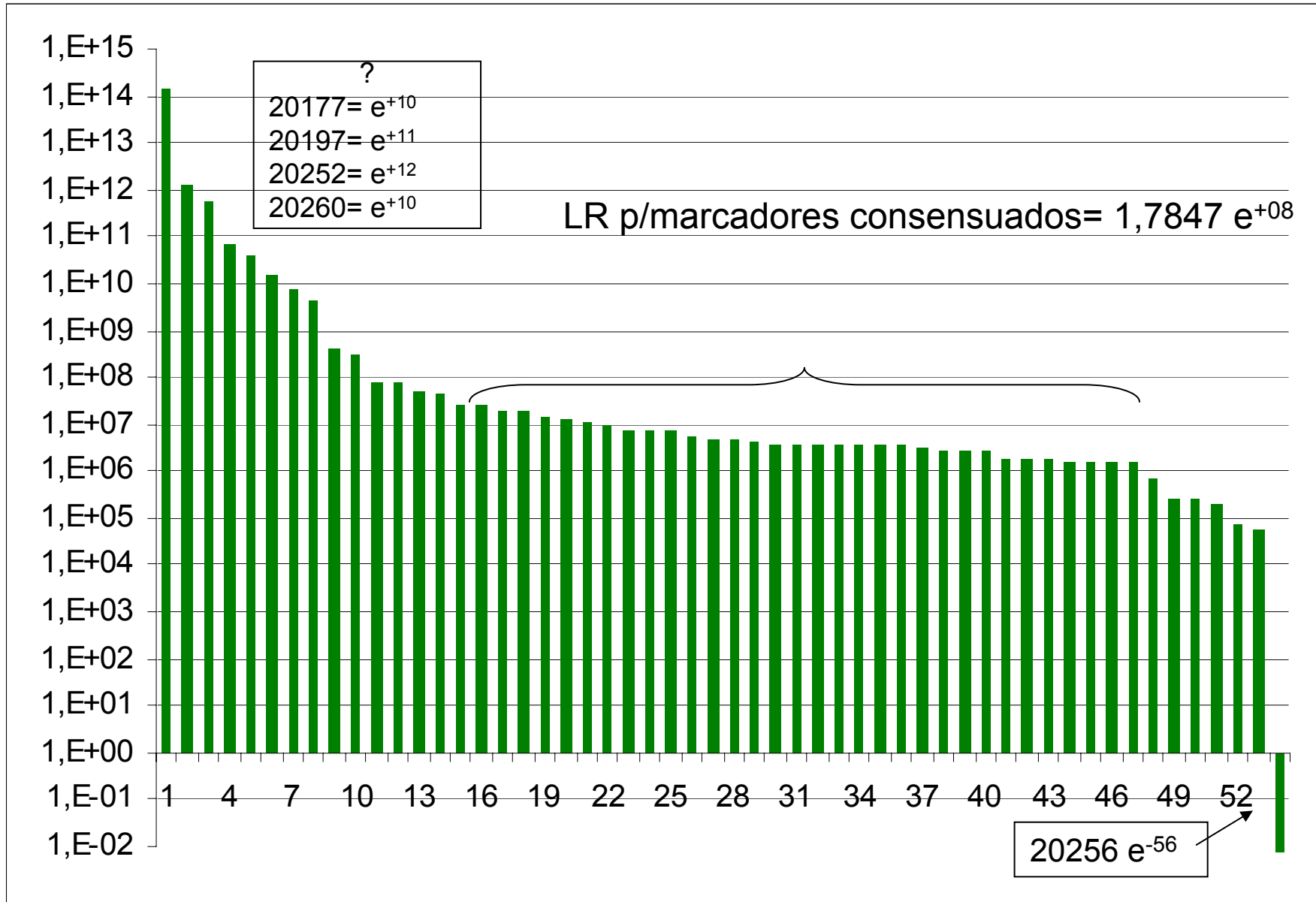
Genotipos			$Y = \sum(\text{Pr}G_{HA} \cdot \text{Pr}G_M \cdot (\text{Pr}_{HA \rightarrow 1} \cdot \text{Pr}_M \rightarrow 2))$	
$G_{PA}$	$G_H$	$G_M$		Y
15/15	16/16	15/15	$f_{15}^2 f_{15}^2 (f_{16} \cdot u/2)$	$f_{15}^4 f_{16} \cdot u/2$
15/0	16/16	15/15	$2f_{15} f_o \cdot f_{15}^2 (f_{16} \cdot u/2)$	$f_{15}^3 f_o f_{16} u$
15/15	16/16	15/0	$f_{15}^2 \cdot 2f_{15} f_o (f_{16} u/4)$	$f_{15}^3 f_o f_{16} u/2$
15/0	16/16	15/0	$2f_{15} f_o \cdot 2f_{15} f_o (f_{16} u/4)$	$f_{15}^2 f_o^2 f_{16} u$
15/0	0/16	15/15	$2f_{15} \cdot f_o \cdot f_{15}^2 (f_o \cdot u/2 + f_{16} \cdot 0)$	$f_{15}^3 f_o^2 u$
15/15	16/0	15/0	$f_{15}^2 \cdot 2f_{15} f_o (f_{16} \cdot \frac{1}{2} + f_o \cdot u/4)$	$f_{15}^3 f_o (f_{16} + f_o \cdot u/2)$
15/0	16/0	15/0	$2f_{15} f_o \cdot 2f_{15} f_o (f_{16} \cdot \frac{1}{2} + f_o \cdot u/4)$	$f_{15}^2 f_o^2 (2f_{16} + f_o \cdot u)$
15/15	16/0	15/15	$f_{15}^2 f_{15}^2 (f_{16} \cdot 0 + f_o \cdot u/2)$	$f_{15}^4 f_o \cdot u/2$

Genotipos			D3S1358	
$G_{PA}$	$G_H$	$G_M$	X	Y
15/15	16/16	15/15	$f15^4 \cdot u^2/4$	$f15^4 f16 \cdot u/2$
15/0	16/16	15/15	$f15^3 fo \cdot u^2/4$	$f15^3 fo f16 u$
15/15	16/16	15/0	$f15^3 fo \cdot u^2/4$	$f15^3 fo f16 u/2$
15/0	16/16	15/0	$f15^2 fo^2 \cdot u^2/4$	$f15^2 fo^2 f16 u$
15/0	0/16	15/15	$f15^3 fo \cdot u/2$	$f15^3 fo^2 u$
15/15	16/0	15/0	$f15^3 fo \cdot u/2$	$f15^3 fo (f16 + fo \cdot u/2)$
15/0	16/0	15/0	$f15^2 fo^2 \cdot u$	$f15^2 fo^2 (2f16 + fo \cdot u)$
15/15	16/0	15/15	0	$f15^4 fo \cdot u/2$

Genotipos			D3S1358		
$G_{PA}$	$G_H$	$G_M$	X	Y	LR = X/Y
15/15	16/16	15/15	$1.4718 e^{-9}$	$7.0413 e^{-7}$	LR= $4.049 e^{-3}$
15/0	16/16	15/15	$2.6567 e^{-11}$	$2.5420 e^{-8}$	
15/15	16/16	15/0	$2.6567 e^{-11}$	$1.2710 e^{-8}$	
15/0	16/16	15/0	$4.7955 e^{-13}$	$4.5884 e^{-10}$	
15/0	0/16	15/15	$5.3135 e^{-8}$	$5.3135 e^{-10}$	
15/15	16/0	15/0	$5.3135 e^{-8}$	$2.5420 e^{-5}$	
15/0	16/0	15/0	$1.9182 e^{-9}$	$9.1769 e^{-7}$	
15/15	16/0	15/15	0	$1.4718 e^{-8}$	
			$\Sigma X = 1.0971 e^{-7}$	$\Sigma Y = 2.7096 e^{-5}$	

NOTA: Considerando  $\mu$  en vez de  $\mu/2$  el LR=  $4.1761 e^{-3}$

# LR total combinado







# Conclusiones



- 53 laboratorios participantes
- La mayoría de marcadores tuvo consenso excepto pocos labs recurrentes
- D21S11: dos consensos
  - frec del alelo paterno obligado
  - frec mínimas
- D3S1358:
  - gran dispersión de resultados
  - la gran mayoría de labs consideró ambas posibilidades:
    - mutac
    - silente
- No hay consenso en el LR final
- Todos los labs consideraron el caso consistente con la H1.
- 31 sobre 34 labs comentaron la posible co-existencia de mutac+silente, 2 consideraron sólo mutación, 1 consideró sólo silente.



**MUCHAS GRACIAS**

