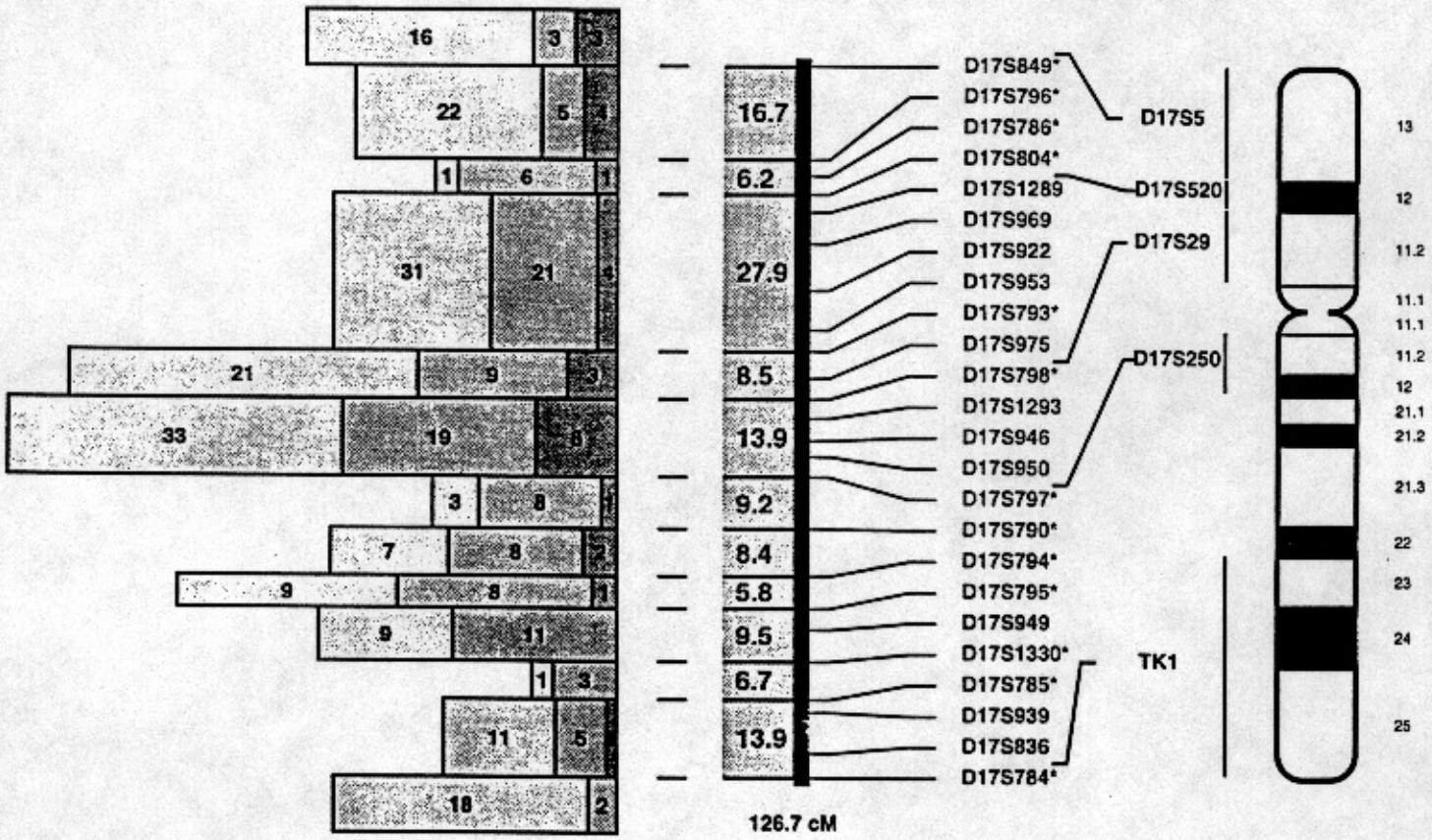
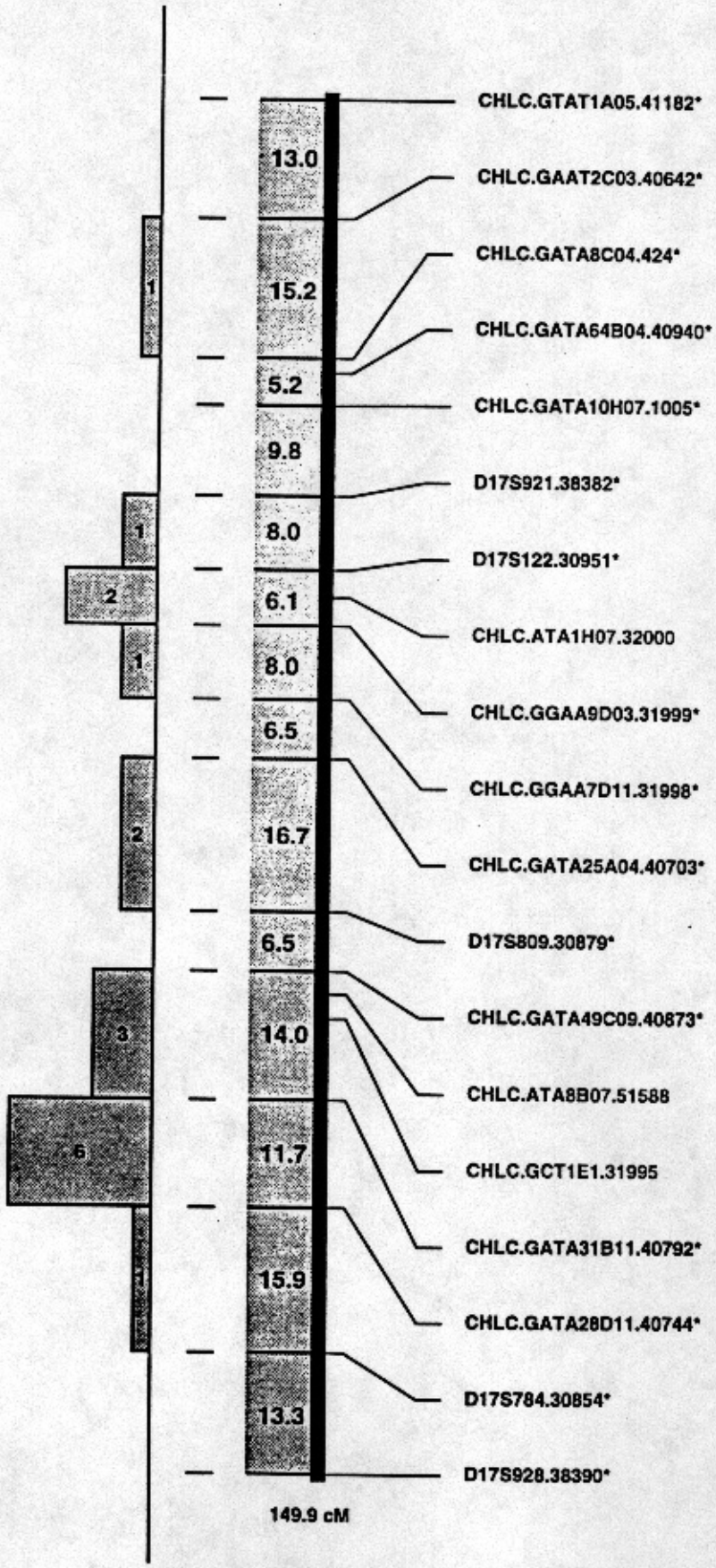
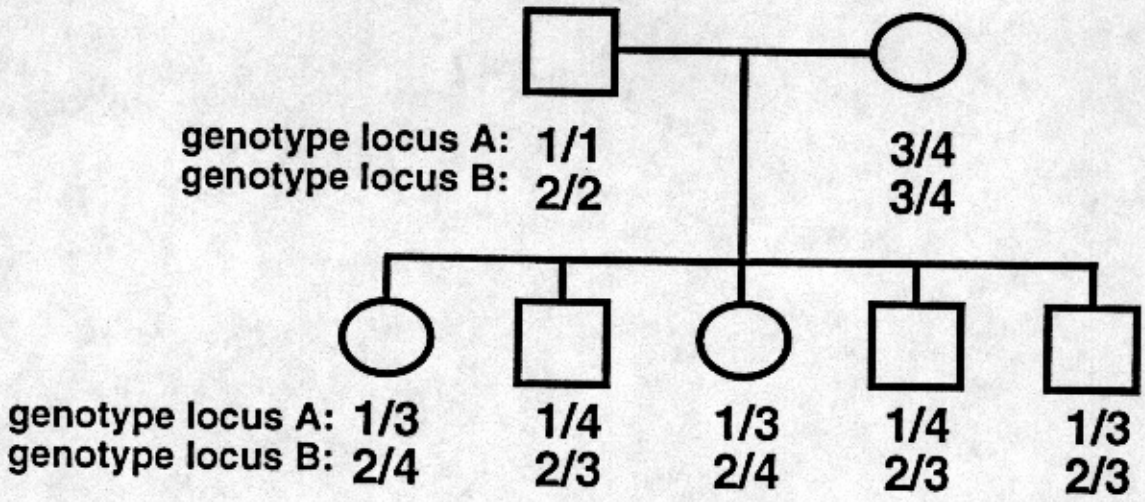

16
3
5
22
5
4
1
6
1
31
21
4
21
9
3
33
19
8
3
8
1
7
8
2
9
8
1
9
11
1
3
11
5
18
2
16.7
6.2
27.9
8.5
13.9
9.2
8.4
5.8
9.5
6.7
13.9
126.7 cM
D17S849*
D17S796*
D17S786*
D17S804*
D17S1289
D17S969
D17S922
D17S953
D17S793*
D17S975
D17S798*
D17S1293
D17S946
D17S950
D17S797*
D17S790*
D17S794*
D17S795*
D17S949
D17S1330*
D17S785*
D17S939
D17S836
D17S784*
D17S5
D17S520
D17S29
D17S250
TK1
13
12
11.2
11.1
11.1
11.2
12
21.1
21.2
21.3
22
23
24
25

CHLC Marker Distribution in V6 Screening Map of Chromosome 17




genotype locus A: 1/1
genotype locus B: 2/2
3/4
3/4
genotype locus A: 1/3
genotype locus B: 2/4
1/4
2/3
1/3
2/4
1/4
2/3
1/3
2/3

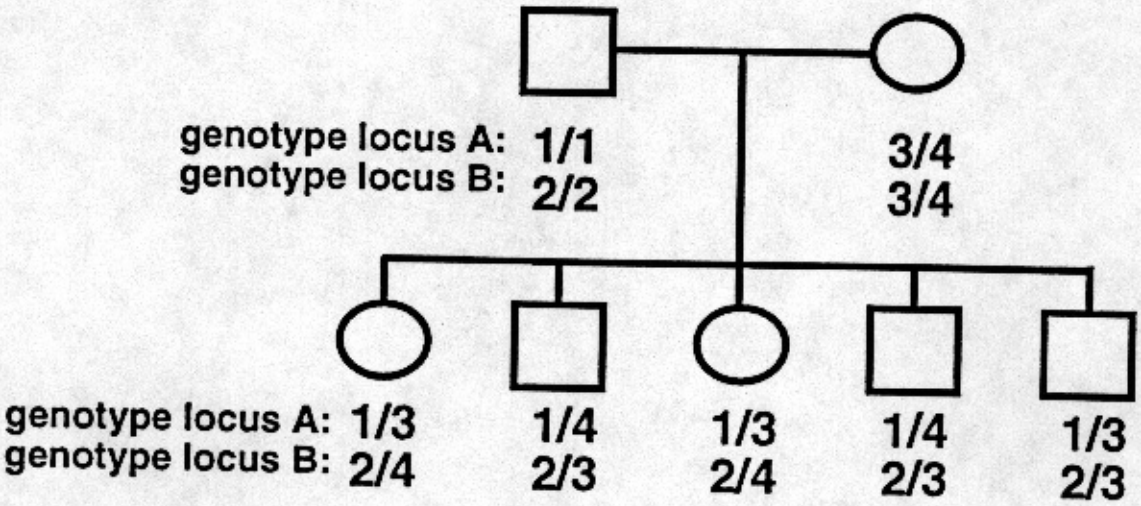
$$z(x) = \log_{10}\left[\frac{L\,(\text{pedigree given } \theta = x)}{L\,(\text{pedigree given } \theta = 0.5)}\right]$$

$$z(x) = \log_{10}\left[\frac{\theta^{R}\,(1-\theta)^{NR}}{(0.5)^{R}\,(0.5)^{NR}}\right]$$

$$z\,(0.05) = \log_{10}\left[\frac{(0.05)^{R}\,(1-0.05)^{NR}}{(0.5)^{N}}\right]$$

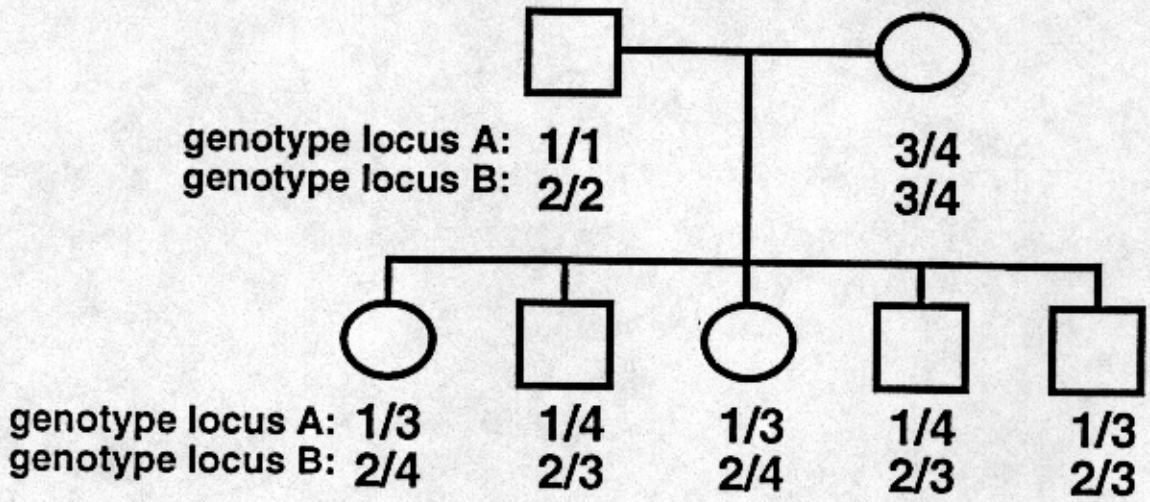
$$z\,(0.05) = \log_{10}\left[\frac{(0.05)^{1}\,(0.95)^{7}}{(0.5)^{8}}\right] = 0.9513$$

$$z\,(0.05) = 0.95.$$



probablity if linkage phase is:

		"coupling"	"repulsion"
		3|4 3|4	3|4 4|3
mother provides gamete:	3| 3|	$1/2(1-\theta)$	$1/2\ \theta$
	4| 4|	$1/2(1-\theta)$	$1/2\ \theta$
	3| 4|	$1/2\ \theta$	$1/2(1-\theta)$
	4| 3|	$1/2\ \theta$	$1/2(1-\theta)$



"coupling"	"repulsion"
3 | 4	3 | 4
3 | 4	4 | 3

$$L(\theta) = [\ (1/2\theta)4\,(1/2(1-\theta)) \quad + \quad (1/2(1-\theta))4\,(1/2\theta)\ ]\ /\ 2$$

$$= 1/64 \quad [\ \theta 4\,(1-\theta) \quad + \quad \theta\,(1-\theta)4\ ]$$

$$L\ (\theta = 0.2) = 1/64\ [\ 0.00128 \quad + \quad 0.08192\ ]$$

$$= 0.0013$$

$$Z = \log 10\ [\ L(\ \theta = 0.2\ )\ -\ L\ (\ \theta = 0.50\ )\ ]$$

$$= 0.124$$

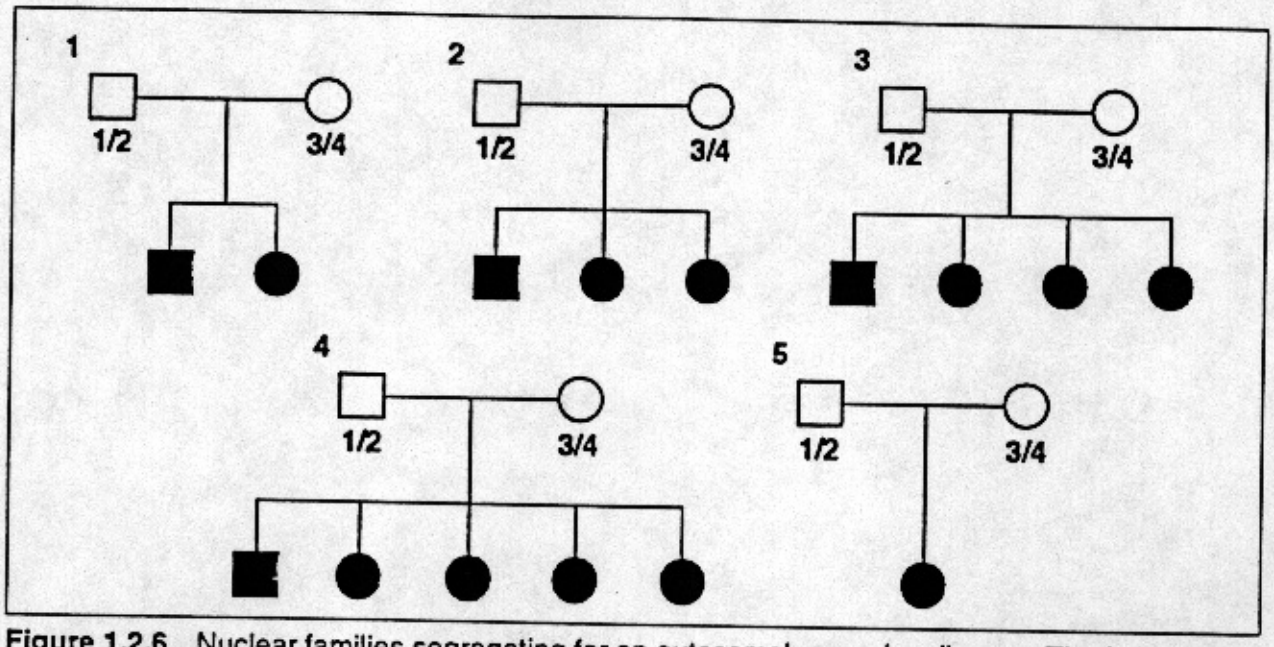


Figure 1.2.6 Nuclear families segregating for an autosomal recessive disease. The father has a 1/2 marker genotype, while the mother has a 3/4 marker genotype. Figure 1.2.7 shows the expected lod scores for each family.

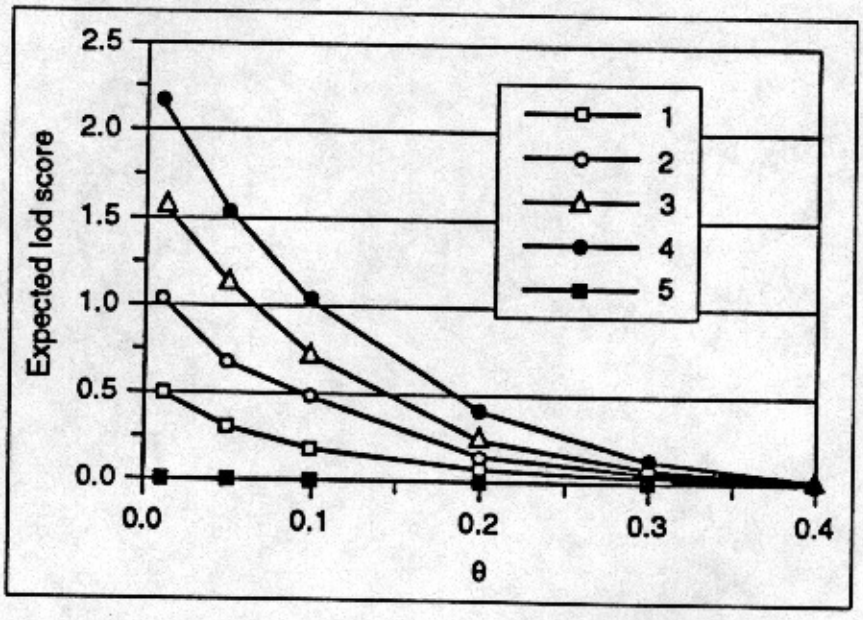
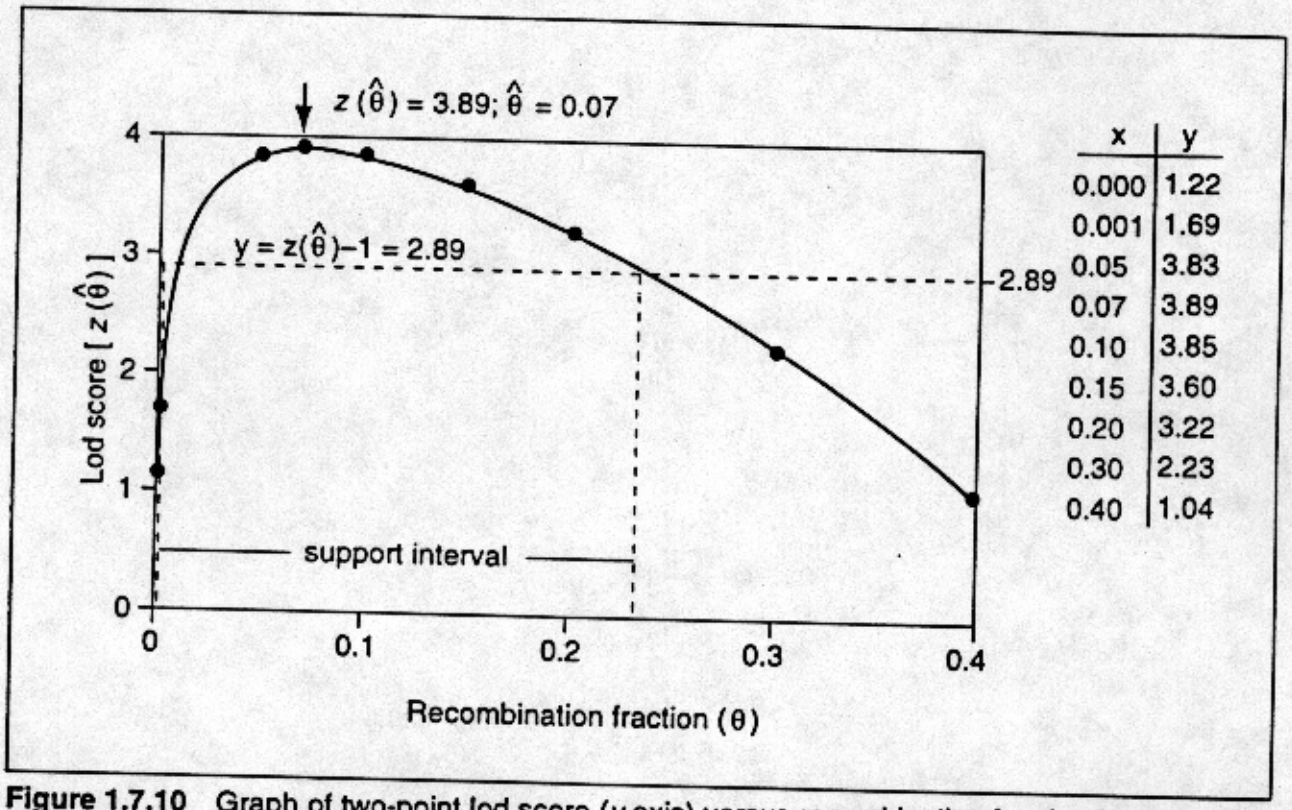


Figure 1.2.7 The expected lod score as a function of the recombination fraction (θ) and pedigree structure (shown in Fig. 1.2.6).



x	y
0.000	1.22
0.001	1.69
0.05	3.83
0.07	3.89
0.10	3.85
0.15	3.60
0.20	3.22
0.30	2.23
0.40	1.04

Figure 1.7.10 Graph of two-point lod score (*y* axis) versus recombination fraction (*x* axis) using values for marker 3 from Table 1.7.3. The horizontal line drawn across the graph is drawn at $y = z(\hat{\theta}) - 1.0$ and is used in constructing the one lod unit support interval for the maximum likelihood estimate of the recombination fraction ($\hat{\theta}$).

Table 2

VWS Linkage with 1q Markers

VWS vs.	$\theta_m = \theta_f$							Z_{max}	$\hat{\theta}$	$Z(m,f)$	$\hat{\theta}_m$	$\hat{\theta}_f$
	0	.001	.05	.10	.20	.30	.40					
REN	7.15	8.22	8.85	8.05	6.03	3.79	1.61	9.09	.02	9.28	0	.03
D1S53	−.94	1.12	3.83	3.79	3.11	2.18	1.14	3.87	.07	4.73	0	.18
CR1	−3.95	−1.47	3.14	3.43	2.87	1.83	.74	3.43	.10	3.43	.09	.10
D1S58	−.08	1.13	2.83	2.82	2.21	1.34	.49	2.88	.07	3.16	.12	0
D1S52	0.57	1.31	2.71	2.69	2.19	1.47	.66	2.74	.07	2.99	0	.11
CR2	−.22	.76	2.40	2.44	1.93	1.21	.53	2.46	.08	2.74	0	.12
DAF	−2.14	−1.86	1.35	1.92	1.81	1.18	.50	1.99	.13	2.02	.09	.15
D1S65	−.90	−.43	1.82	1.95	1.58	.10	.42	1.95	.09	1.97	.13	.06
LAMB2	−6.27	−4.10	−.36	.54	1.13	1.01	.56	1.14	.22	1.17	.26	.19

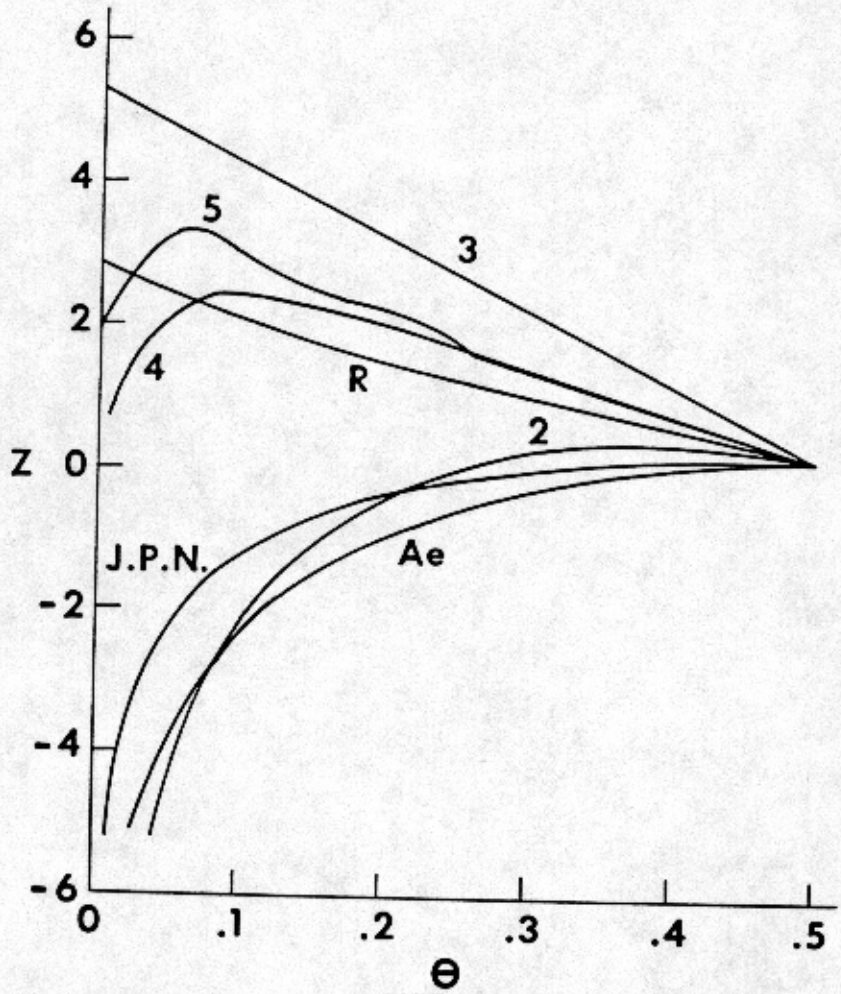


Fig. 6.2.1: Linkage of elliptocytosis with *RH* in 4 of 7 families. At a recombination value of $\theta = .05$, the lod score Z is greater than 2 in families 3,4,5, and R (linkage) and less than 2 in families 2, Ae, and JPN (no linkage).