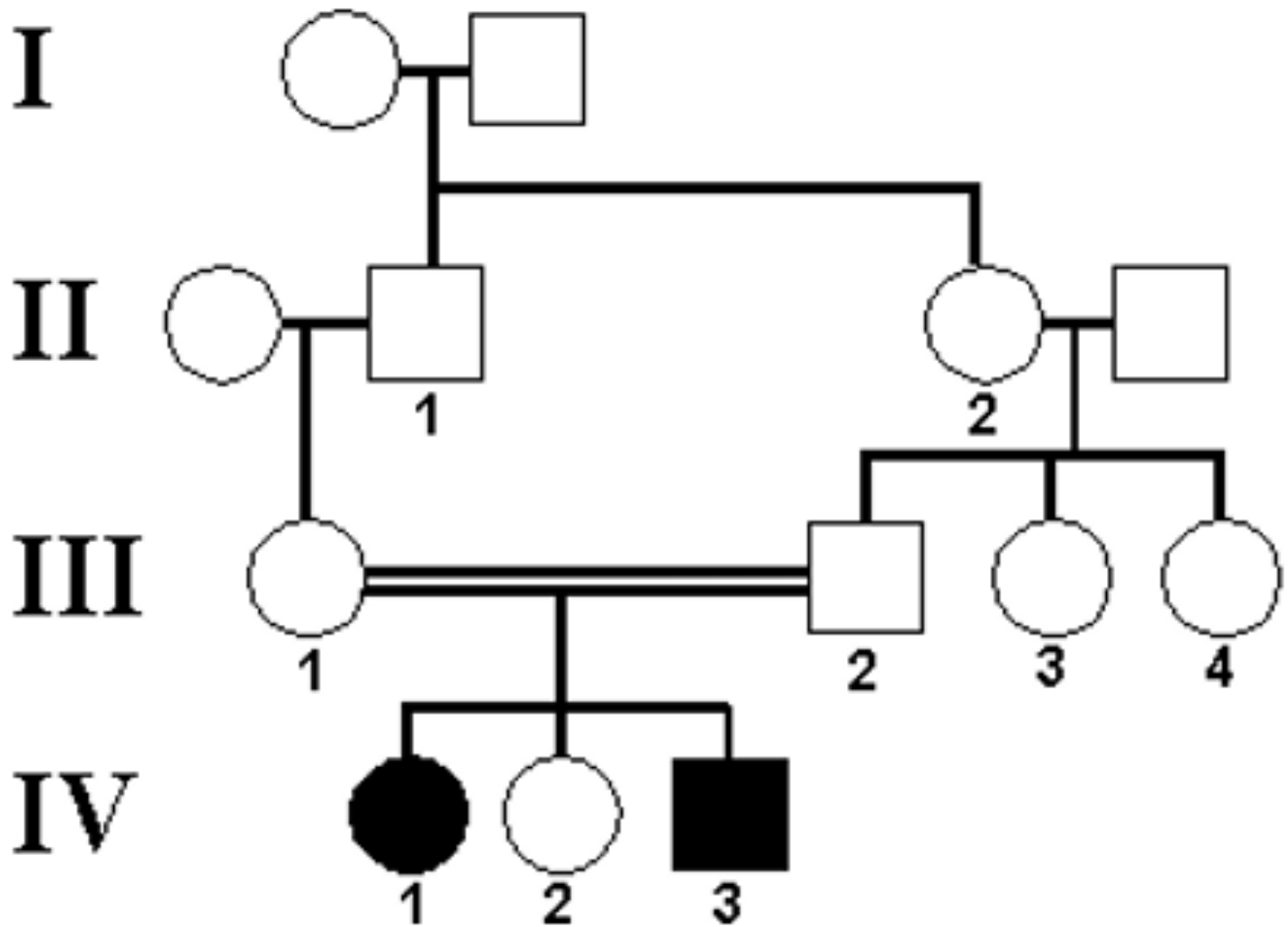

I
II
1
2
III
1
2
3
4
IV
1
2
3

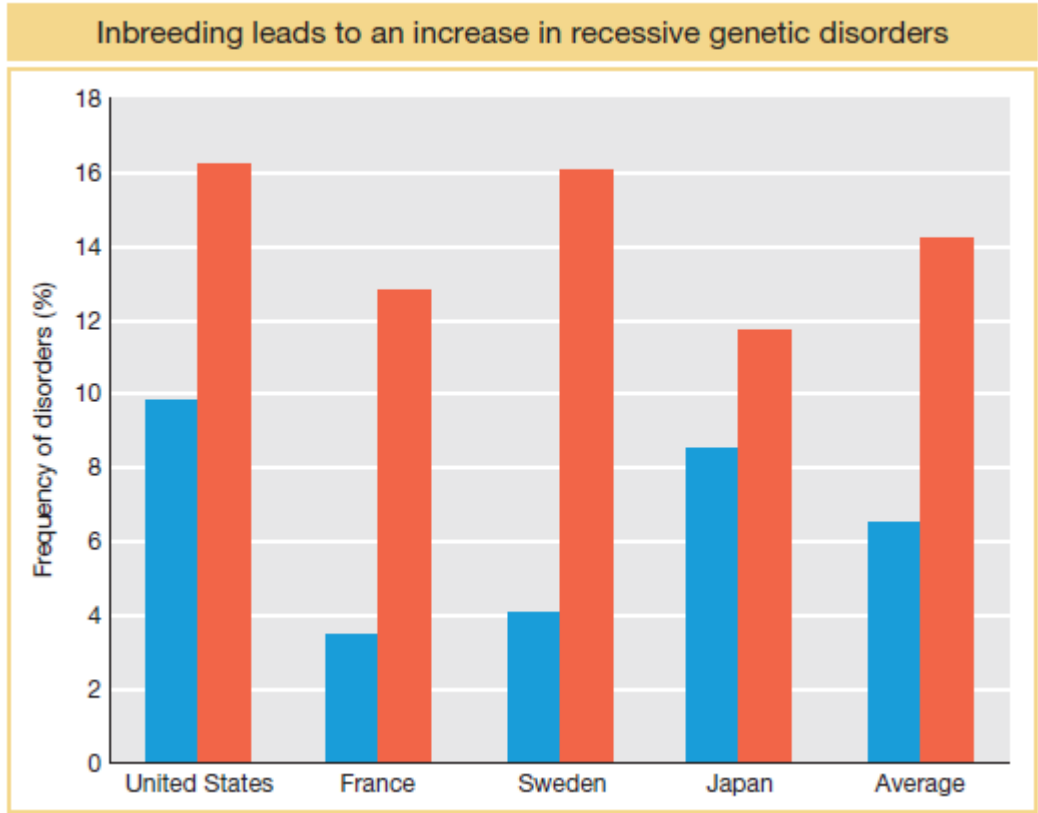
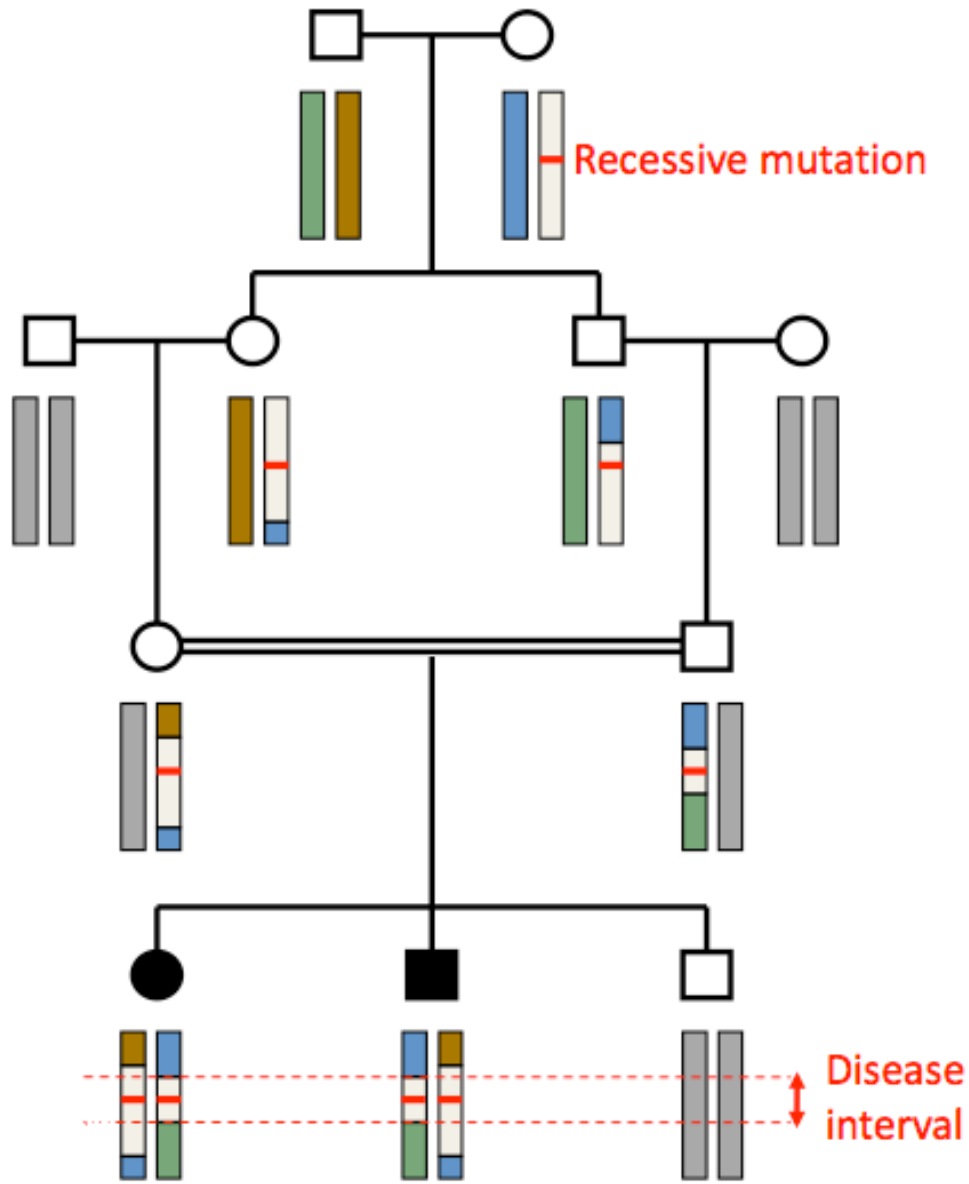
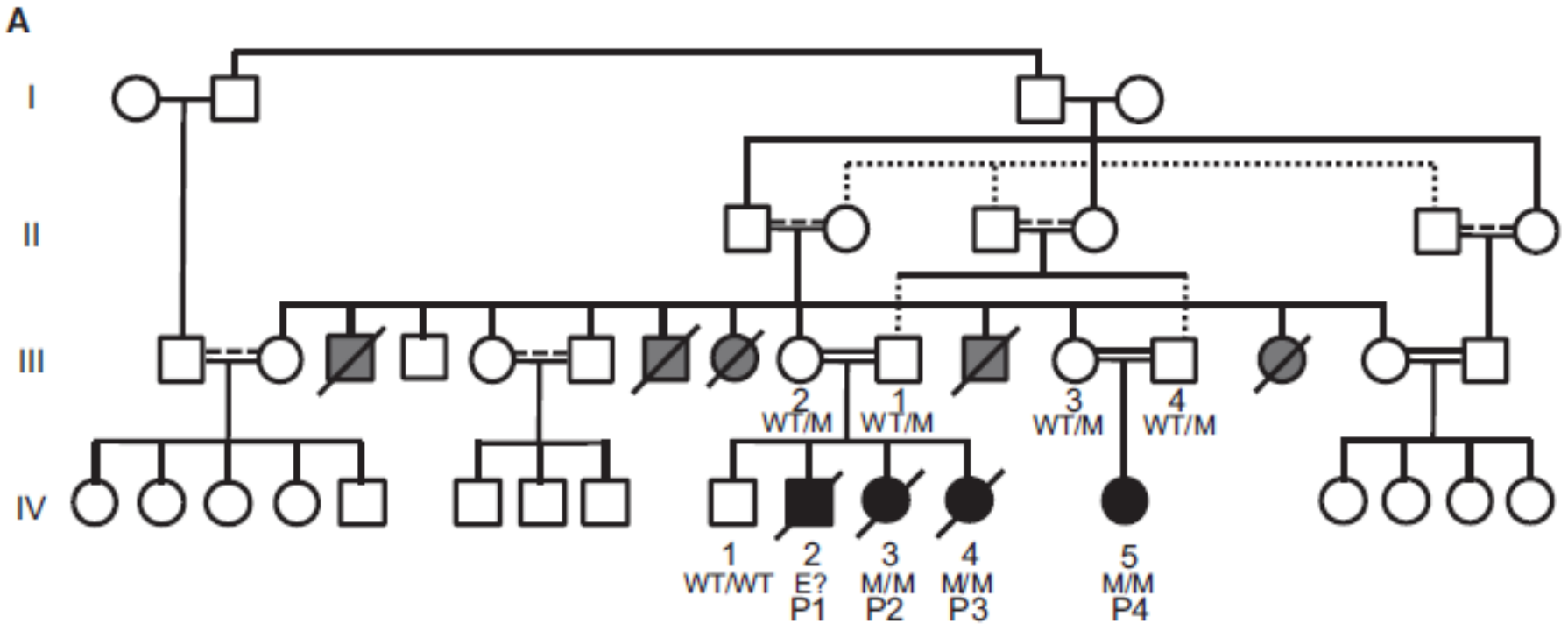


FIGURE 18-13 Frequency of genetic disorders among children of unrelated parents (blue columns) compared to that of children of parents who are first cousins (red columns). [*Data from C. Stern,* Principles of Human Genetics, *W. H. Freeman, 1973.*]


Recessive mutation
Disease
interval

Whole-Exome-Sequencing-Based Discovery of Human FADD Deficiency

Alexandre Bolze,[1] Minji Byun,[1,13] David McDonald,[2,13] Neil V. Morgan,[3,13] Avinash Abhyankar,[1,13] Lakshmanane Premkumar,[4,13] Anne Puel,[5] Chris M. Bacon,[6] Frédéric Rieux-Laucat,[7] Ki Pang,[8] Alison Britland,[9] Laurent Abel,[1,5] Andrew Cant,[2,10] Eamonn R. Maher,[3,11] Stefan J. Riedl,[4] Sophie Hambleton,[2,10] and Jean-Laurent Casanova[1,5,12,*]



Genotyped P2, P3, P4, III.1, III.2, III.3, III.4 and IV.1. Two 8 and 9 Mb regions homozygous in patients but not in healthy individuals. Exome sequencing of P3.

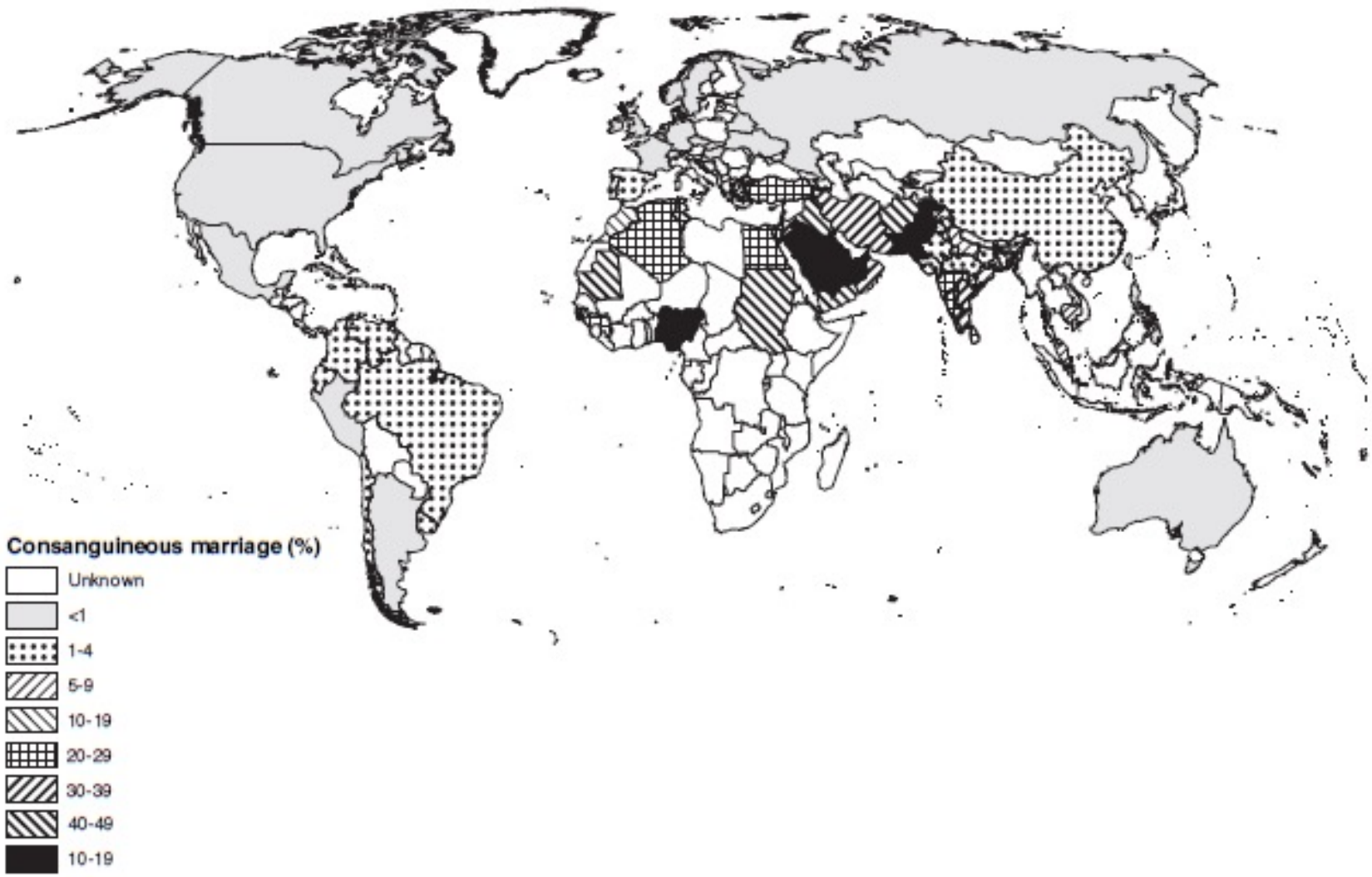


Fig. 17.2 Global distribution of consanguineous marriages

Table 17.1 Human genetic relationships

Biological relationship	Genetic relationships	Coefficient of relationship	Coefficient of inbreeding
Incest[a]	First degree	0.5	0.25
Uncle-niece Double first cousin	Second degree	0.25	0.125
First cousin	Third degree	0.125	0.0625
First cousin once removed Double second cousin	Fourth degree	0.0625	0.0313
Second cousin	Fifth degree	0.0313	0.0156
Second cousin once removed Double third cousin	Sixth degree	0.0156	0.0078
Third cousin	Seventh degree	0.0078	0.0039

[a]Incest is defined as a sexual relationship between father–daughter, mother–son or brother–sister

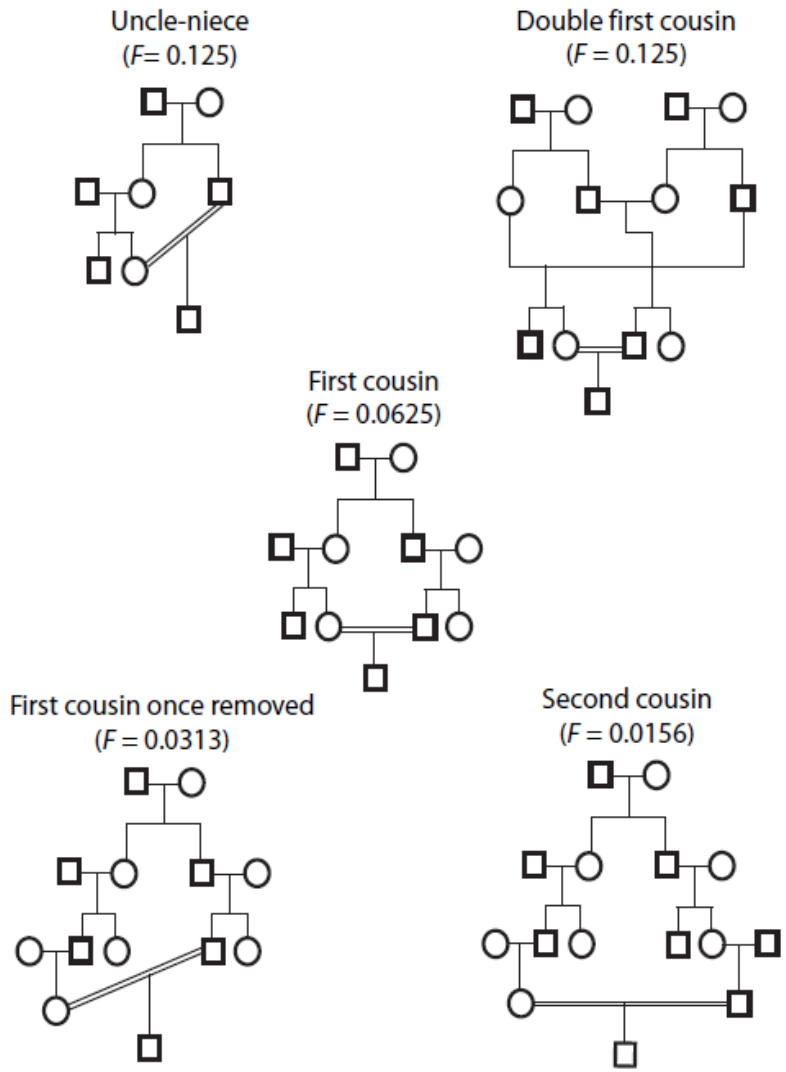


Fig. 17.1 Consanguineous pedigrees