Malattie Legate all'X

MALATTIE LEGATE ALL'X

I maschi hanno un solo allele e le femmine due.

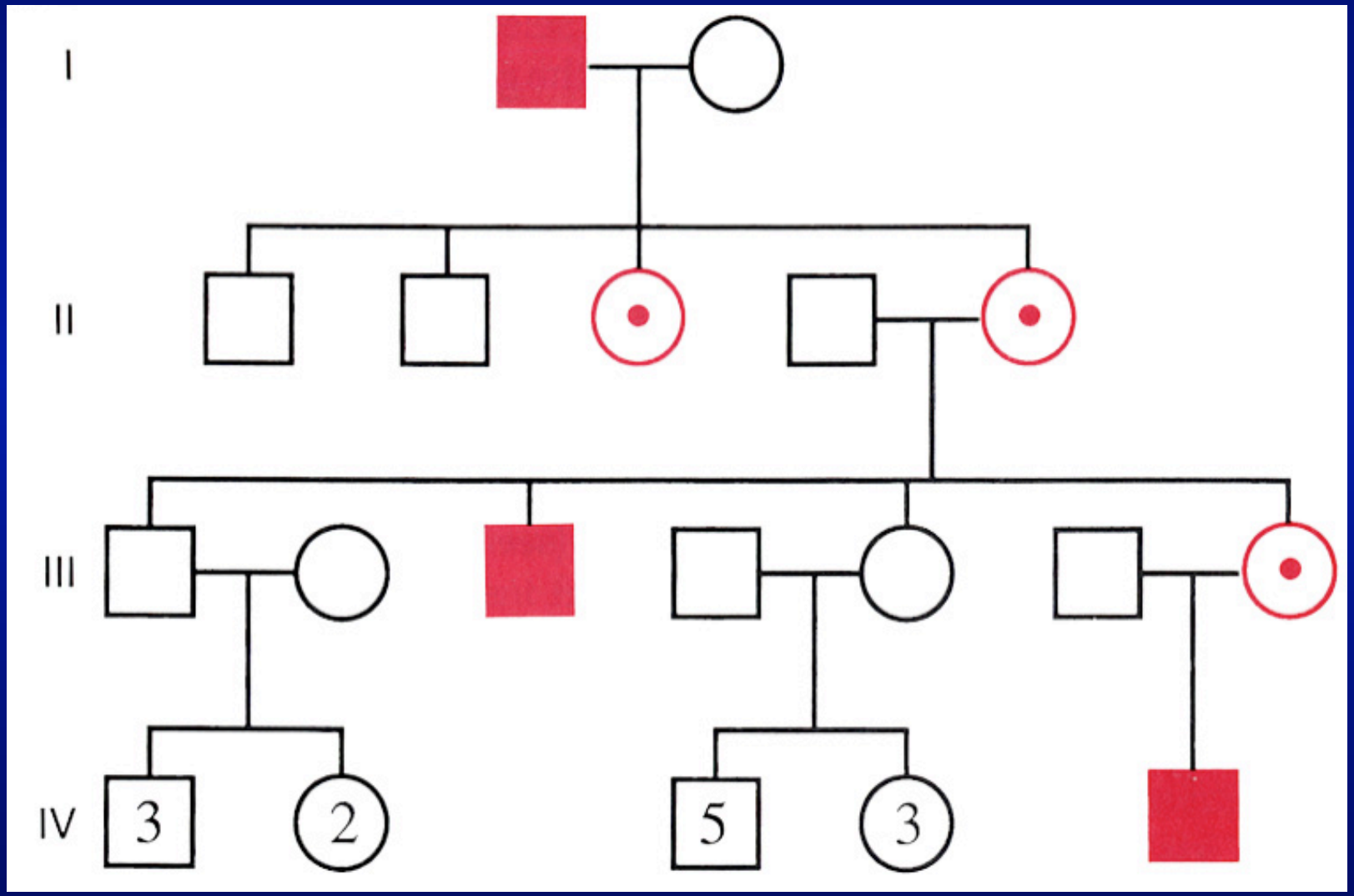
Recessive

Maschi	Femmine
A = affetto	NN = normale
N = normale	NA = normale
	AA = affetto

Dominanti

Maschi	Femmine
A = affetto	NN = normale
N = normale	NA = affetto
	AA = affetto

Malattie legate all’ X recessive



Malattie Legate All'X

RECESSIVE

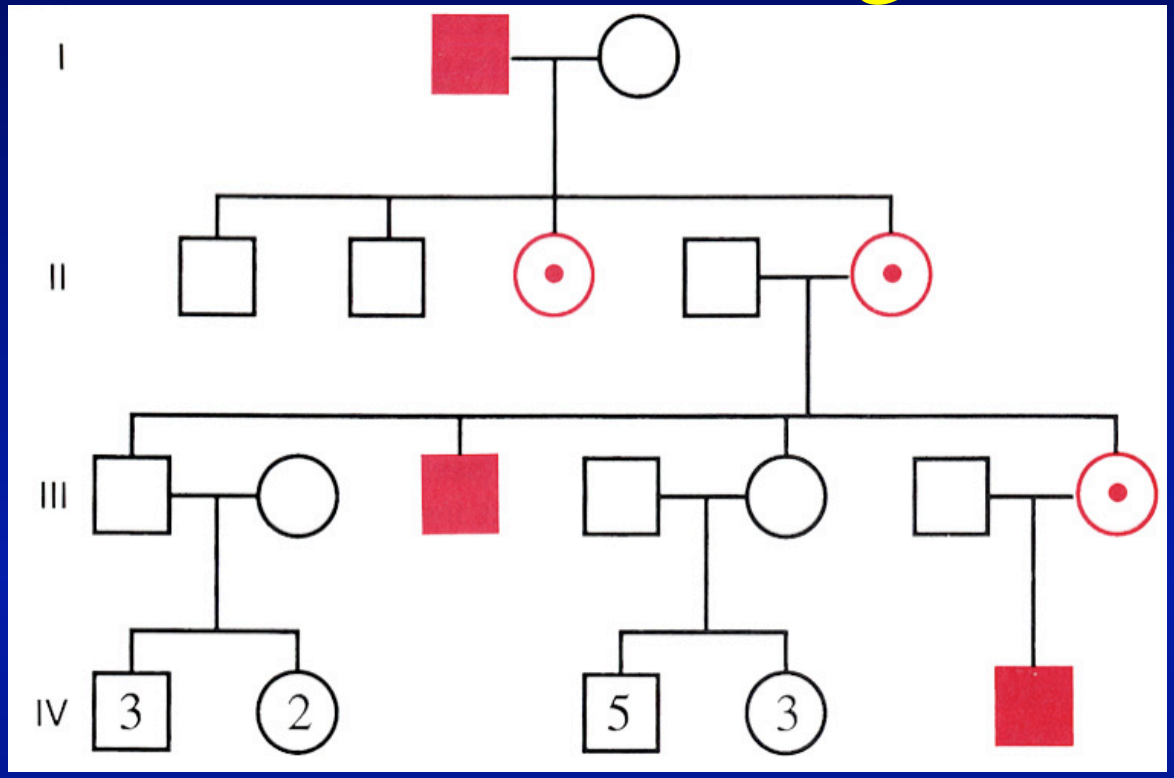
⇒ **Mai trasmissione maschio - maschio**

⇒ **Solo maschi affetti**

⇒ **Tutti i figli maschi dei maschi affetti sono sani e 100% delle figlie femmine sono portatrici**

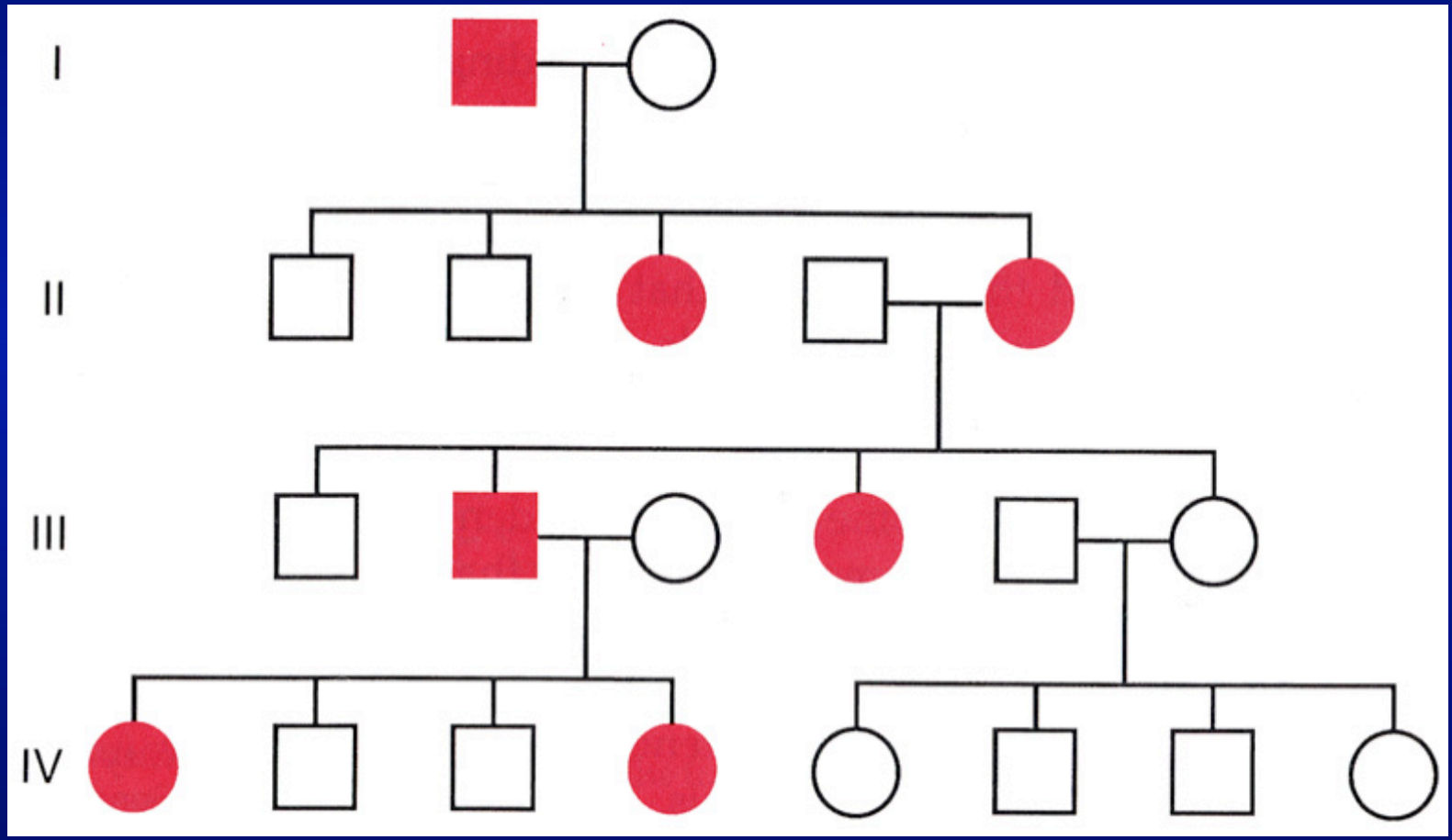
⇒ **Una femmina portatrice ha un rischio del 50% di avere figli maschi affetti e del 50% di avere figlie femmine portatrici**

Malattie Recessive Legate All' X



- ⇒ **Mai trasmissione maschio - maschio**
- ⇒ **Solo maschi affetti**
- ⇒ **Tutti i figli maschi dei maschi affetti sono sani e 100% delle figlie femmine sono portatrici**
- ⇒ **Una femmina portatrice ha un rischio del 50% di avere figli maschi affetti e del 50% di avere figlie femmine portatrici**

Malattie legate all’ X dominanti

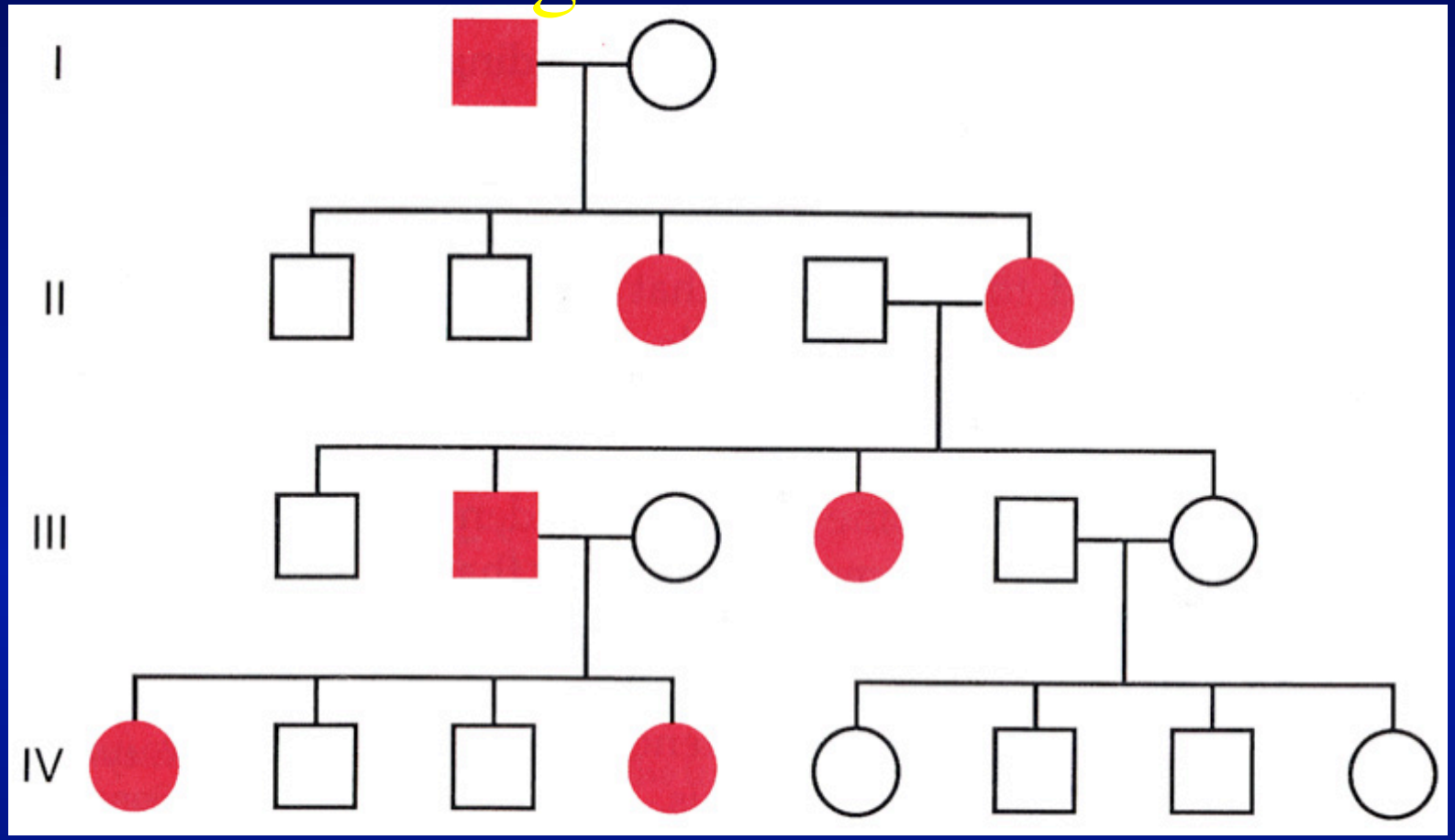


Malattie Legate All'X

DOMINANTI

- ⇒ **Mai trasmissione maschio - maschio**
- ⇒ **Affetti sia maschi che femmine**
- ⇒ **Il 100% delle figlie di un padre affetto sono affette**
- ⇒ **Una madre affetta ha un rischio del 50% di aver figli affetti (sia maschi che femmine)**

Malattie legate all'X dominanti

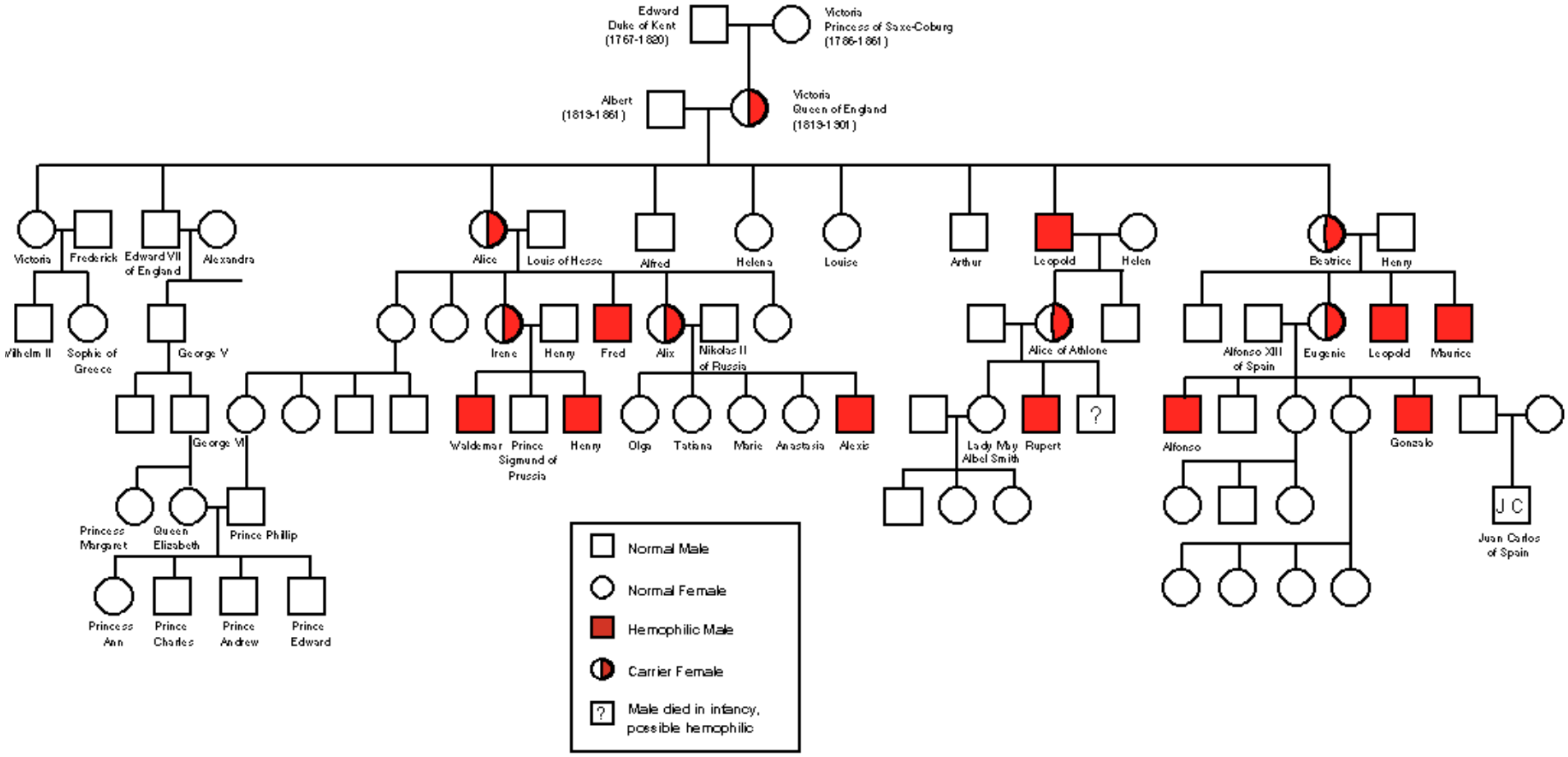


- ⇒ **Mai trasmissione maschio - maschio**
- ⇒ **Affetti sia maschi che femmine**
- ⇒ **Il 100% delle figlie di un padre affetto sono affette**
- ⇒ **Una madre affetta ha un rischio del 50% di aver figli affetti (sia maschi che femmine)**

Frequenza di alcune comuni malattie legate all'X

X-linked

Duchenne muscular dystrophy	1 in 3000 males
Hemophilia A	1 in 10,000 males
Fragile X mental retardation	See Table 1-3


Edward
Duke of Kent
(1767-1820)
Victoria
Princess of Saxe-Coburg
(1786-1861)
Albert
(1819-1861)
Victoria
Queen of England
(1819-1901)
Victoria
Frederick
Edward VII
of England
Alexandra
Alice
Louis of Hesse
Alfred
Helena
Louise
Arthur
Leopold
Helen
Beatrice
Henry
Wilhelm II
Sophie of
Greece
George V
Irene
Henry
Fred
Alix
Nikolas II
of Russia
Alice of Athlone
Alfonso XIII
of Spain
Eugenie
Leopold
Maurice
George VI
Waldemar
Prince
Sigmund of
Prussia
Henry
Olga
Tatiana
Marie
Anastasia
Alexis
Lady May
Abel Smith
Rupert
?
Alfonso
Gonzalo
J C
Juan Carlos
of Spain
Princess
Margaret
Queen
Elizabeth
Prince Phillip
Princess
Ann
Prince
Charles
Prince
Andrew
Prince
Edward
Normal Male
Normal Female
Hemophilic Male
Carrier Female
?
Male died in infancy,
possible hemophilic

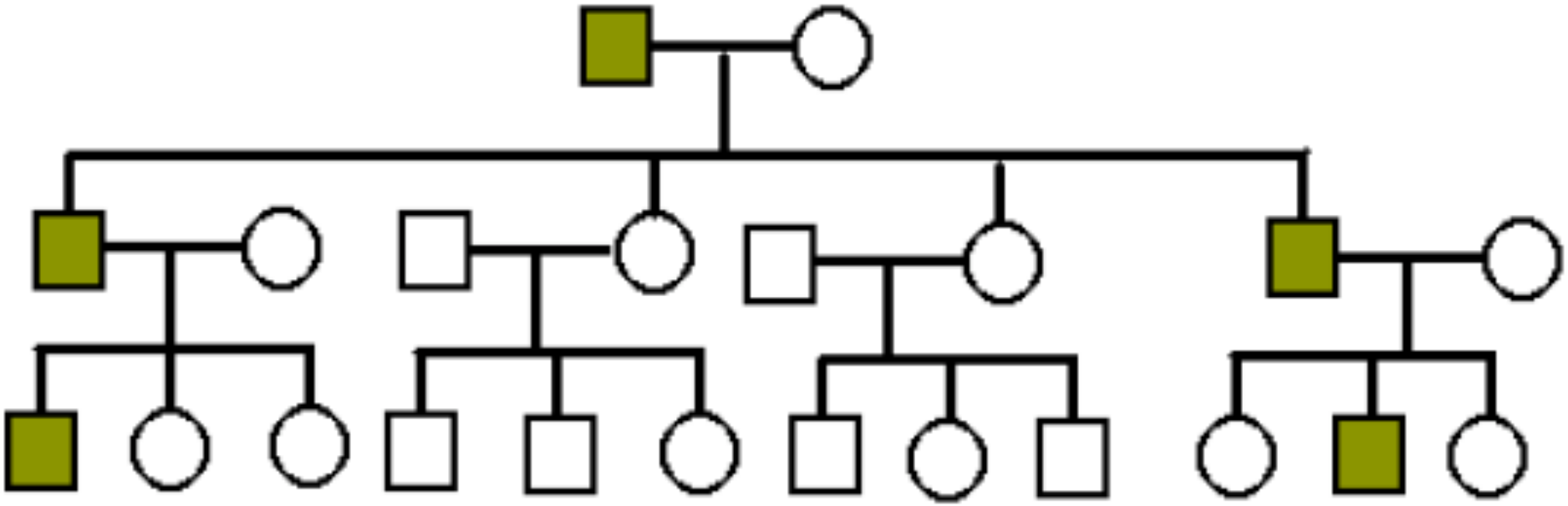
Eredità Y-Linked (rara)

- solo gli individui maschi sono affetti
- trasmissione diretta da padre a figlio
  - cromosoma Y
- i figli maschi affetti avranno sempre un padre affetto
  - a meno che sia insorta una nuova mutazione

#415000 SPERMATOGENIC FAILURE, Y-LINKED
#400042 SERTOLI CELL-ONLY SYNDROME, Y-LINKED

Sull'Y 48 geni
SRY

Y-linked inheritance



FREQUENZA DELLE MALATTIE MONOGENICHE

- La frequenza di alcune malattie varia tra I gruppi etnici (esempio anemia a cellule falciformi fibrosi cistica, malattia di Tay-Sachs, deficit di alfa 1 antitrypsina fenilchetonuria).

- Per altre malattie invece la frequenza e' meno variabile specialmente quando sono frequenti le nuove mutazioni (ad esempio acondroplasia, distrofia di Duchenne, ed Emofilia A).

http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?db=OMIM

National Center for Biotechnology Information

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Online Mendelian Inheritance in Man

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Welcome to OMIM(TM), Online Mendelian Inheritance in Man. This database is a catalog of human genes and genetic disorders authored and edited by Dr. Victor A. McKusick and his colleagues at Johns Hopkins and elsewhere, and developed for the World Wide Web by NCBI, the National Center for Biotechnology Information. The database contains textual information, pictures, and reference information. It also contains copious links to NCBI's Entrez database of MEDLINE articles and sequence information.

NEW The OMIM Morbid Map, a catalog of genetic diseases and their cytogenetic map locations arranged alphabetically by disease, is now available.

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- The OMIM Gene List

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Text: ☐ References: ☐ Clinical Synopsis: ☐
Gene Map Disorder: ☐ Contributors: ☐

See only records which have been changed in the past No Limit
Display at most 50 entries

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14 entries found, searching for "haemophilia"

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*306900 HEMOPHILIA B; HEMB
*306700 HEMOPHILIA A
*193400 VON WILLEBRAND DISEASE
*309550 FRAGILE SITE MENTAL RETARDATION 1; FMR1
134500 FACTOR VIII DEFICIENCY
134510 FACTOR VIII AND FACTOR IX, COMBINED DEFICIENCY OF
134520 FACTORS VIII, IX AND XI, COMBINED DEFICIENCY OF
*151626 LINE RETROTRANSPOSABLE ELEMENT 1; LRE1
#176270 PRADER-WILLI SYNDROME; PWS
#277480 VON WILLEBRAND DISEASE, RECESSIVE FORM
*303800 COLORBLINDNESS, PARTIAL, DEUTAN SERIES; CBD
*303630 COLLAGEN, TYPE IV, ALPHA-5; COL4A5
*311030 MCF.2 CELL LINE DERIVED TRANSFORMING SEQUENCE; MCF2
*604210 CRUMBS, DROSOPHILA, HOMOLOG OF, 1; CRB1

*306700 HEMOPHILIA A

Alternative titles; symbols

HEMOPHILIA, CLASSIC; HEMA
COAGULATION FACTOR VIIIC, PROCOAGULANT COMPONENT, INCLUDED; F8C, INCLUDED
COAGULATION FACTOR VIII, INCLUDED; F8, INCLUDED

TABLE OF CONTENTS

DESCRIPTION

Hemophilia A is an X-linked, recessive, bleeding disorder caused by a deficiency in the activity of coagulation factor VIII. Affected individuals develop a variable phenotype of hemorrhage into joints and muscles, easy bruising, and prolonged bleeding from wounds. The disorder is caused by heterogeneous mutations in the factor VIII gene which maps to Xq28. Despite the heterogeneity in factor VIII mutations, carrier detection and prenatal diagnosis can be done by direct detection of selected mutations (especially the inversions), as well as indirectly by linkage analysis. Replacement of factor VIII is done using a variety of preparations derived from human plasma or recombinant techniques. While replacement therapy is effective in most cases, 10 to 15% of treated individuals develop neutralizing antibodies that decrease its effectiveness.

NOMENCLATURE

The term hemophilia is used in reference to hemophilia A (factor VIII deficiency); hemophilia B or Christmas disease (factor IX deficiency) and von Willebrand disease (von Willebrand factor deficiency). While hemophilia A and B are X-linked disorders, von Willebrand disease has an autosomal dominant, or in some cases, an autosomal recessive mode of inheritance.

PHENOTYPE

Affected individuals develop a variable phenotype of hemorrhage into joints and muscles, easy bruising, and prolonged bleeding from wounds. A partial deficiency in heterozygous carriers was demonstrated by Rapaport et al. (1960). Hemophilia A and B are clinically similar and can only be distinguished by assays of factor VIII and IX activity. In contrast von Willebrand disease more often presents with mucocutaneous or gastrointestinal hemorrhage or menorrhagia. Tests used in its diagnosis include bleeding time, platelet aggregation, and factor VIII and von Willebrand factor assays.

CLINICAL FEATURES

The severity and frequency of bleeding in hemophilia A is inversely related to the amount of residual factor VIII (<1%, severe; 2-5%, moderate; and 5-30%, mild). The proportion of cases that are severe, moderate, and mild are about 50, 10, and 40%,