



Beaumont



Craniofacial Anomalies

Pablo Antonio Ysunza M.D. Ph. D.

Beaumont Royal Oak

40 YEARS
A PHYSICIAN



37 YEARS
TREATING
PATIENTS WITH
VPI

**The devil knows more for
being old than for being the
devil**



Basic Medical Genetics

Nomenclature

Inheritance

Congenital

Malformation



Localized error
of
morphogenesis

Deformation

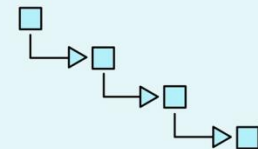


Disruption of
morphogenesis

Sequence

One
malformation
leading to
another(s)

Sequence



Syndrome (Genetics)

Group of malformations with a common etiology



Syndrome (Clinical)

Group of signs and
symptoms

Spectrum

Group of malformations of unknown or unspecified etiology



Hereditary Etiology



Linked to
Genetics

Transmitted
to
generations

Congenital

Present
from
birth



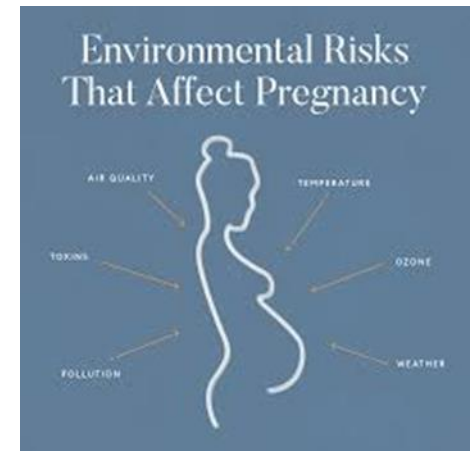
ILLUSTRATION: PEXOLOGISTUDIO/SCIENCE SOURCE

Hereditary



All hereditary anomalies are congenital

Not all congenital anomalies are hereditary
(Environment, infections, drugs, X rays)



Syndromes

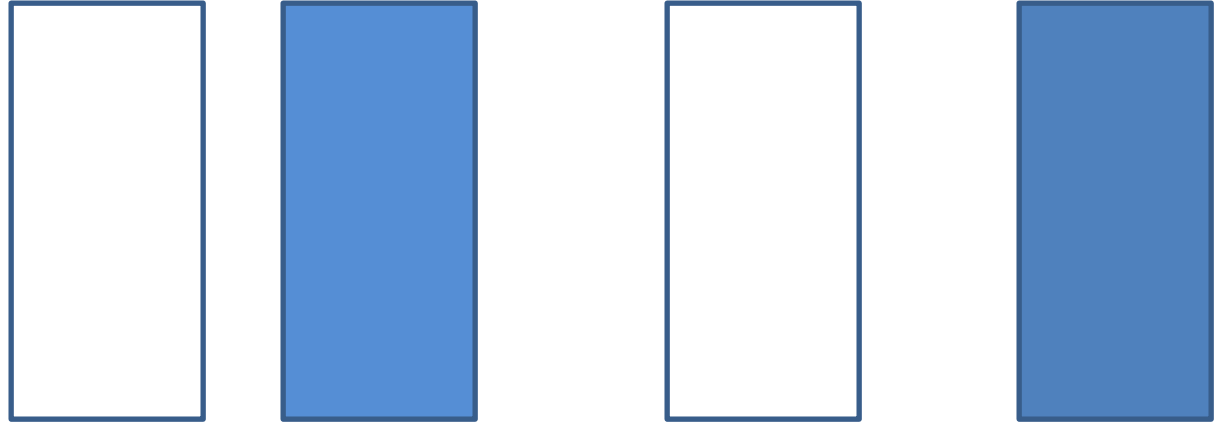


Single Gene

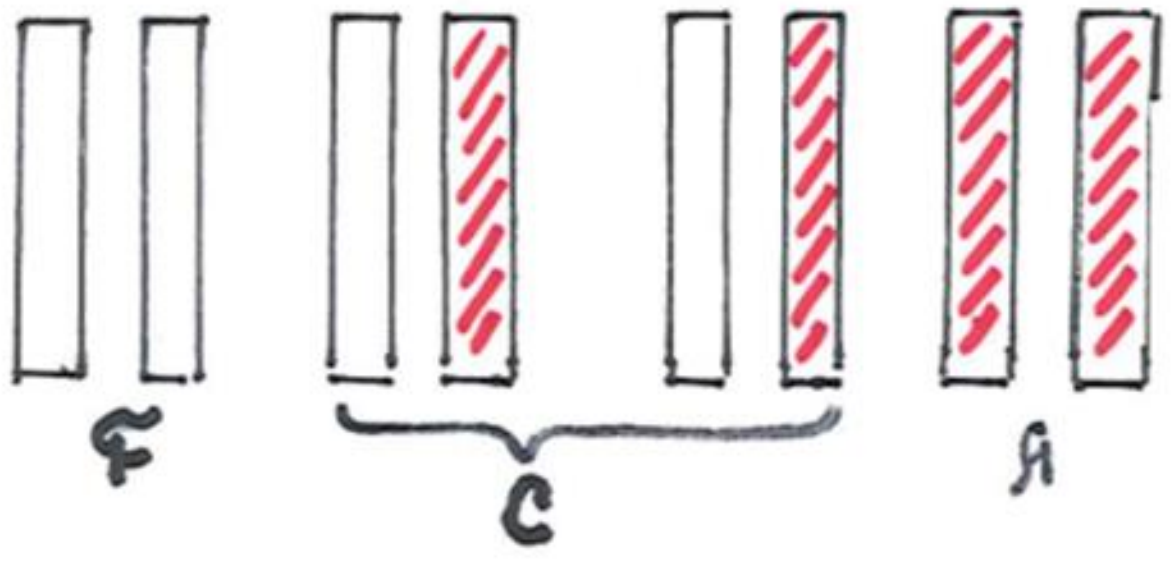
Chromosomal



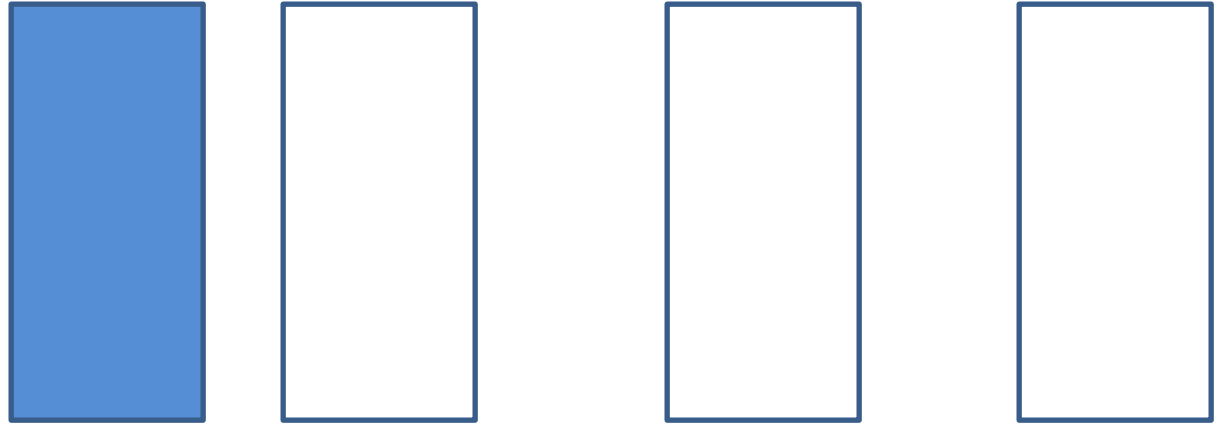
Autosomal Recessive



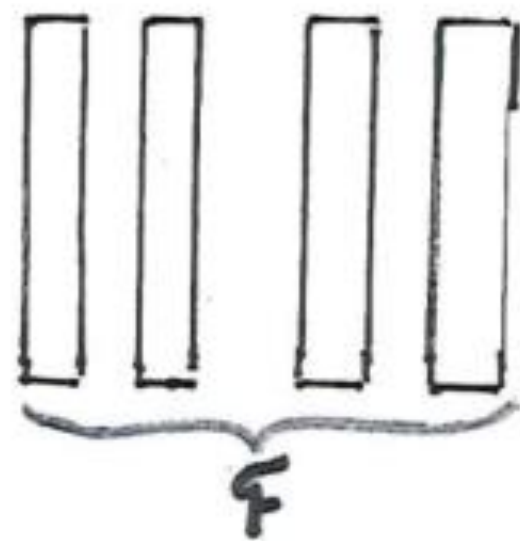
Beckwith-Wiedemann syndrome



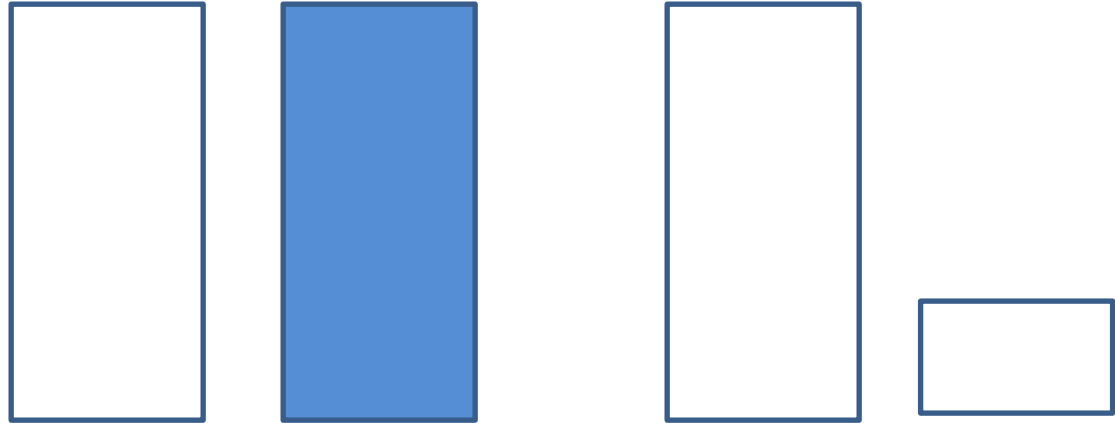
Autosomal Dominant

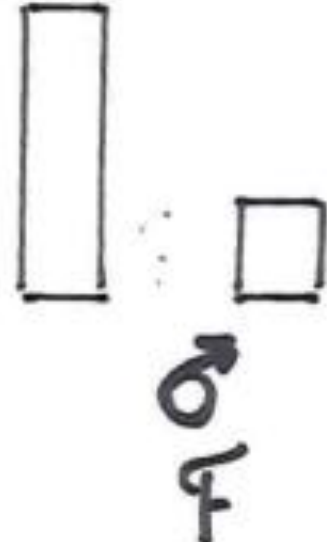
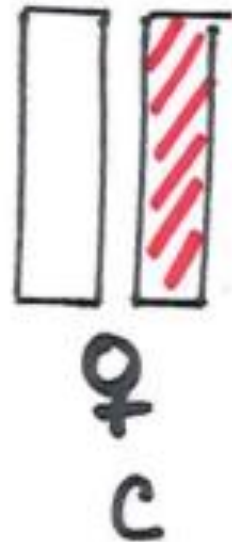
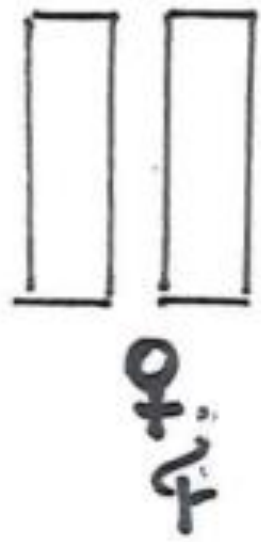


22q11.2 del.



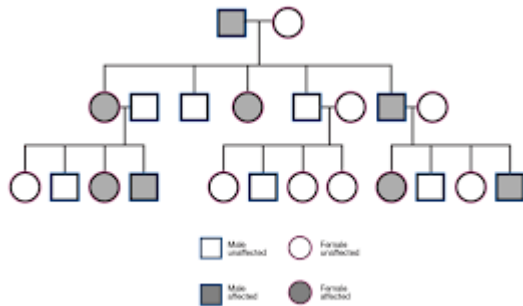
X Linked Recessive





Autosomal Dominant

Inheritance pattern



Generation to generation

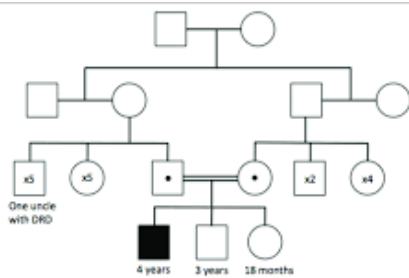
50% probability each pregnancy independent of gender

Autosomal Recessive

Inheritance pattern

Skip generations

25% probability each pregnancy
independent of gender



X Linked Recessive

Females are carriers

50% probability of males affected each pregnancy



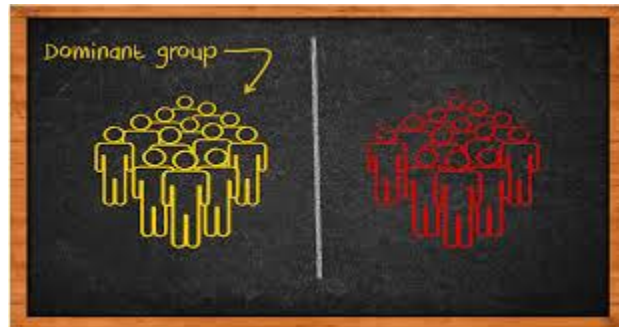
X Linked Dominant

Females and Males can be affected

Rare

50% probability of males affected each pregnancy

50% probability of females affected each pregnancy



Multifactorial Inheritance

Combined action of several low – effect genes with influence by gestational environment

Repetition risk depends on several factors.

Repetition in subsequent pregnancies increase risk.



Teratogen

Teratos



Agent causing malformations or deformations in an embryo

