

# Dr. Laird Jackson

Hosted By:

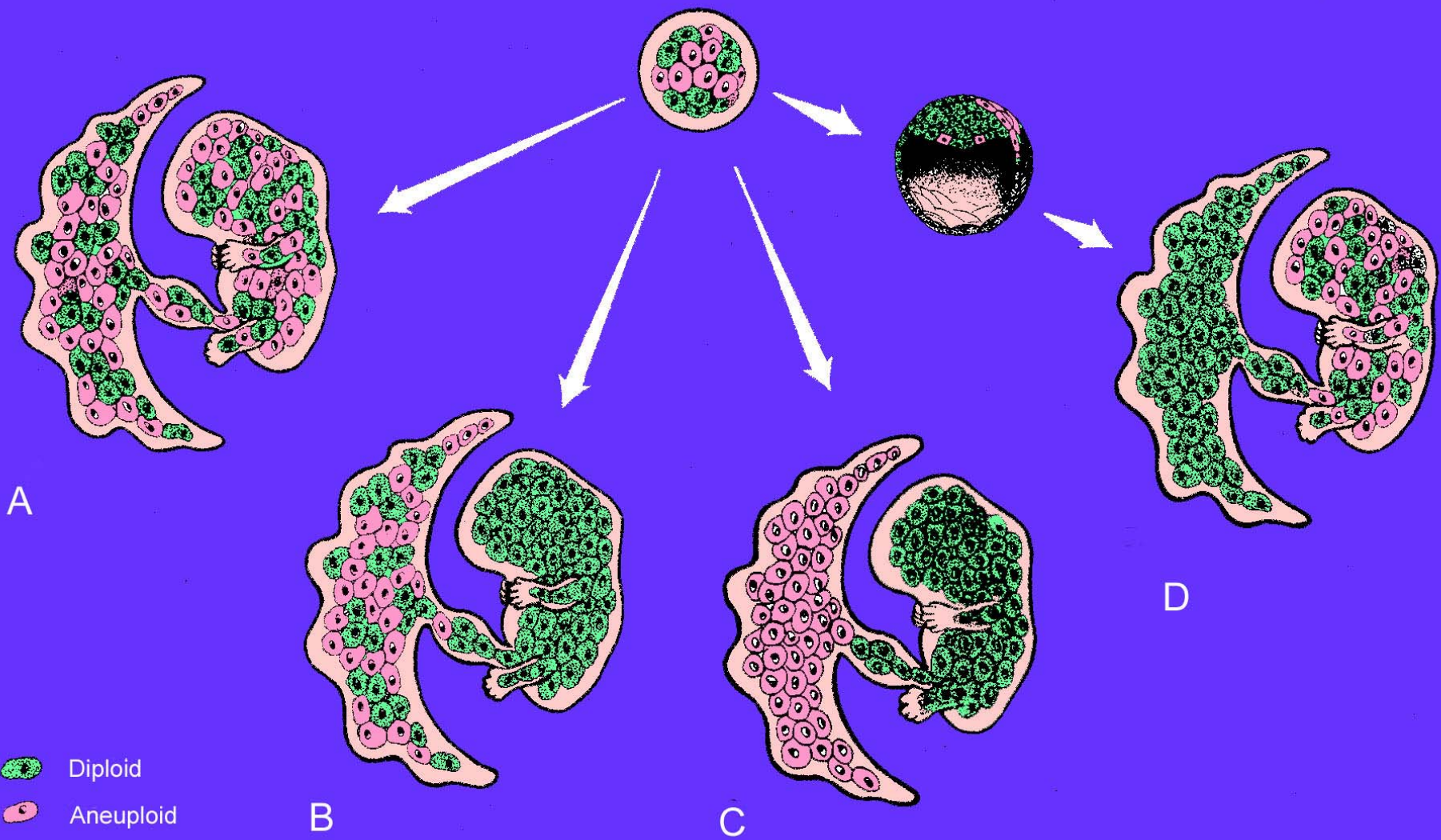
U.S. Department of Health and Human Services  
National Institutes of Health

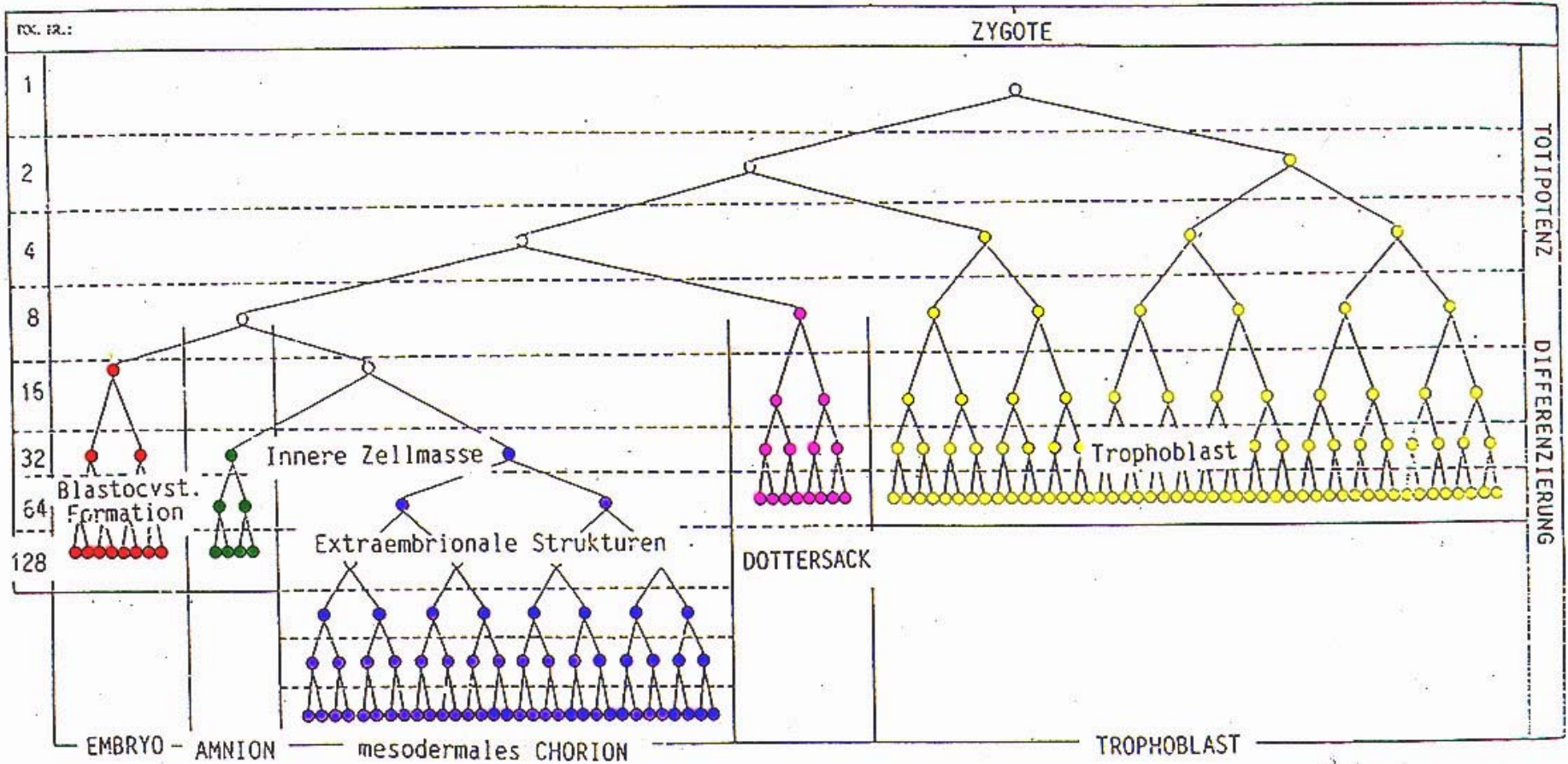
# Confined Placental Mosaicism CPM

Kalousek and Dill – Science – 1983

Studied the cytogenetics of placentas of IUFD infants

Found cytogenetic abnormalities not present in the infant's constitutional karyotype





# Confined Placental Mosaicism CPM

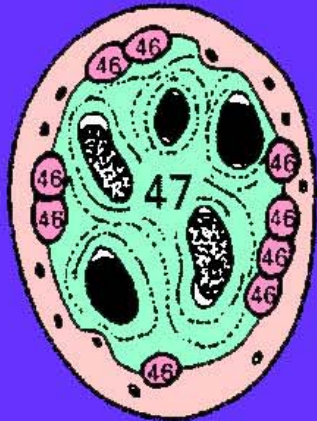
Kalousek and Vekemans – J Med Genet–1996

Post-zygotic cell division event

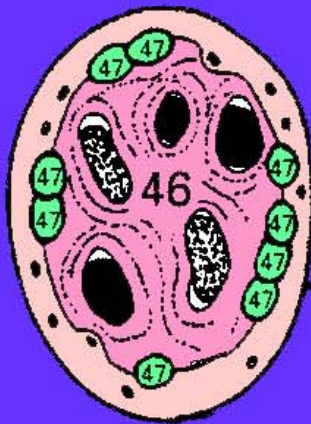
Position of mutant cell in the morula determines the impact on embryo. Unequal distribution of cell lines and small number of cells committed to the inner cell mass increases the probability that mosaic will be confined to the trophoblast



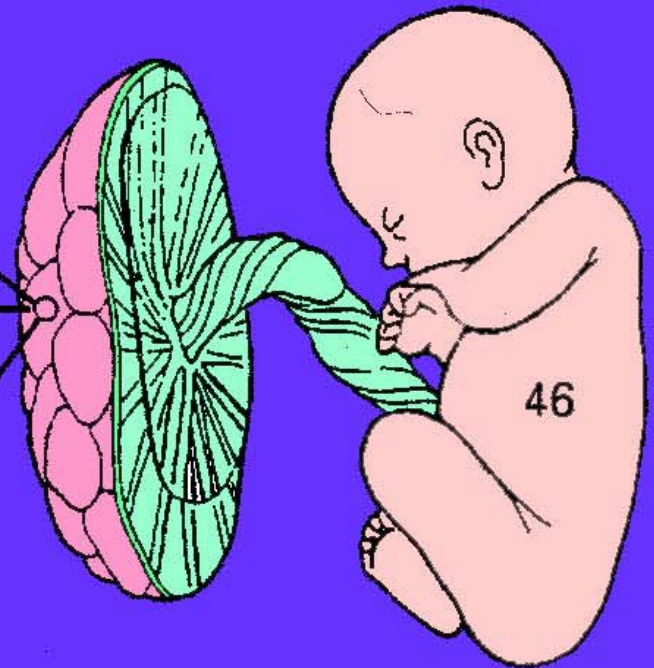
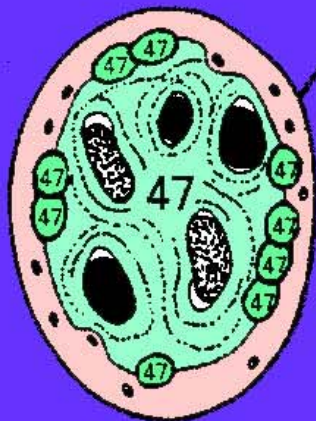
Type II



Type I



Type III



# Confined Placental Mosaicism CPM

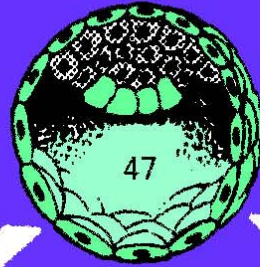
Kalousek and Vekemans – J Med Genet–1996

Post-zygotic cell division event

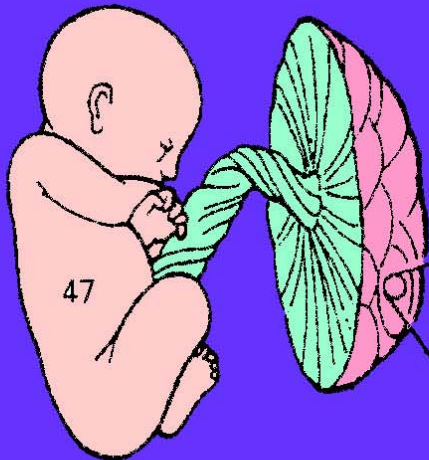
Mitotic non-disjunction in the mosaic tissue

Rescue of meiotic non-disjunction in normal tissue by loss of 1 of 3 trisomic chromosomes

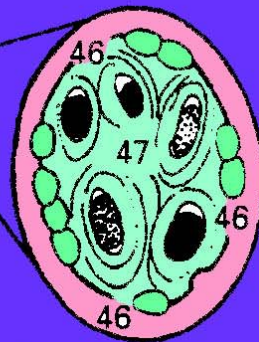
Trisomic zygote



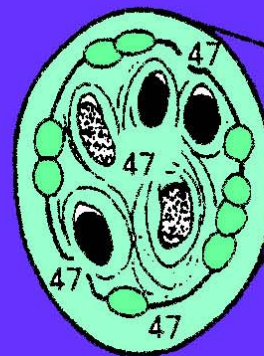
Postzygotic  
chromosome  
loss



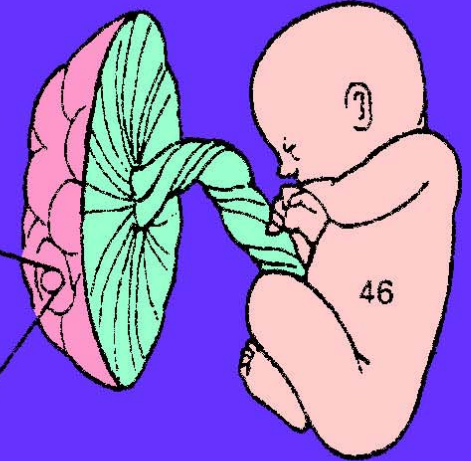
Non-mosaic  
trisomic  
fetus



Mosaic placenta  
with confinement  
of diploidy to  
trophoblast



Trisomic placenta

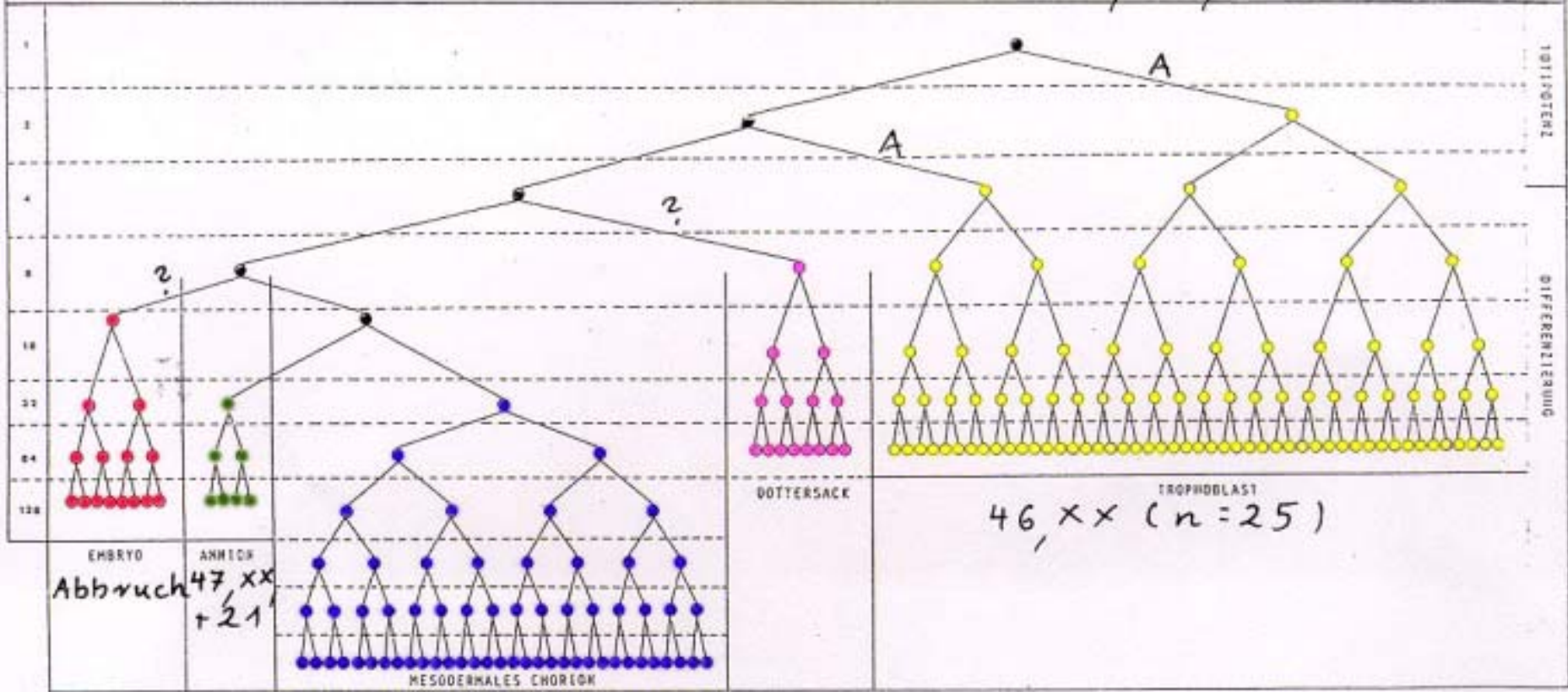


Non-mosaic  
diploid  
fetus



EXC. NR.: 00420

TRGOTIC: 47, XX, +21



EMBRYO  
Abbruch 47, XX  
+21

MESODERMALES CHORION

47, XX, +21 (n=50)

DOTTERSACK

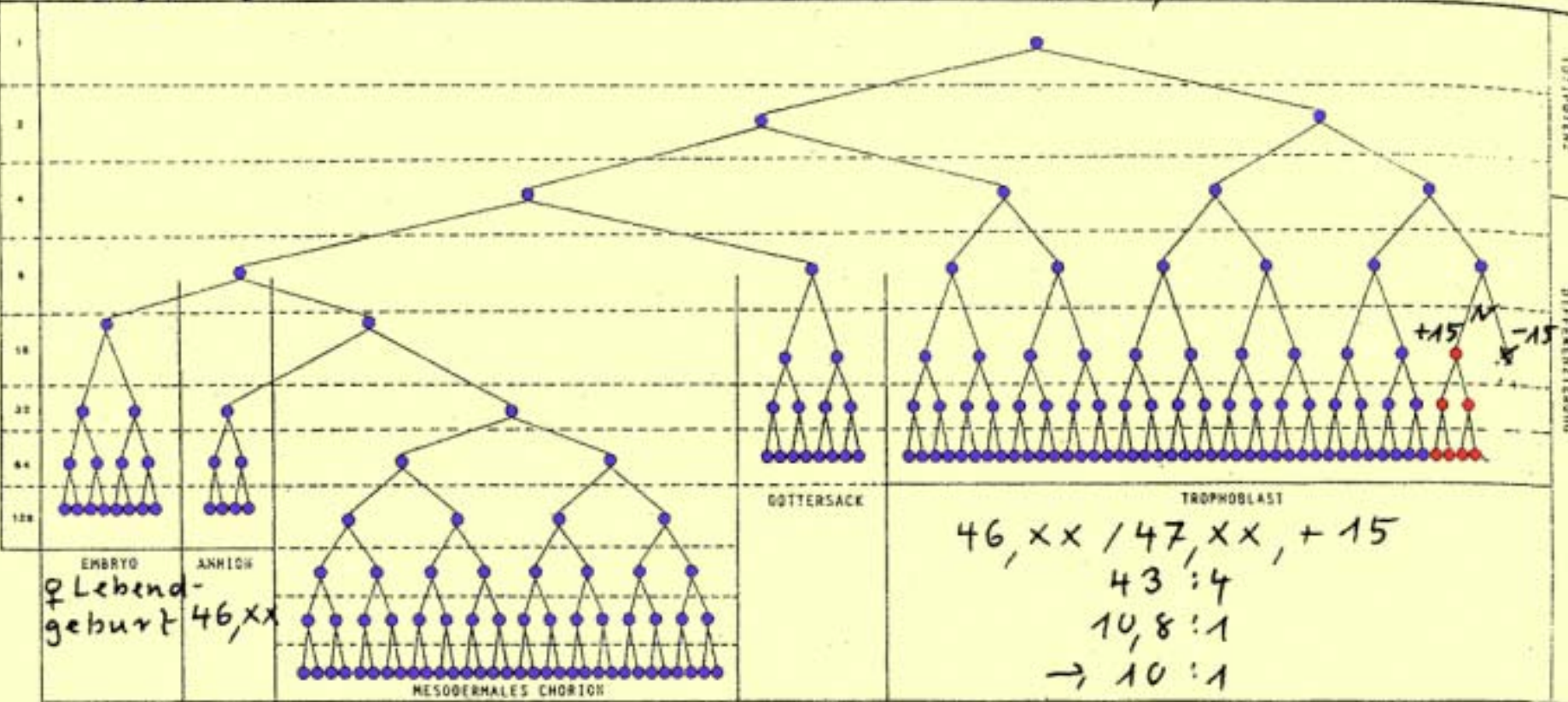
TROPHOBLAST  
46, XX (n=25)

1911 POTENZ

DIFFERENZIERUNG

REC. NR.: 08343

ZYGOTE: 46, XX



EMBRYO  
♀ Lebend-  
geburt 46, XX

ANHION  
46, XX

MESODERMALES CHORION

GOTTERSACK

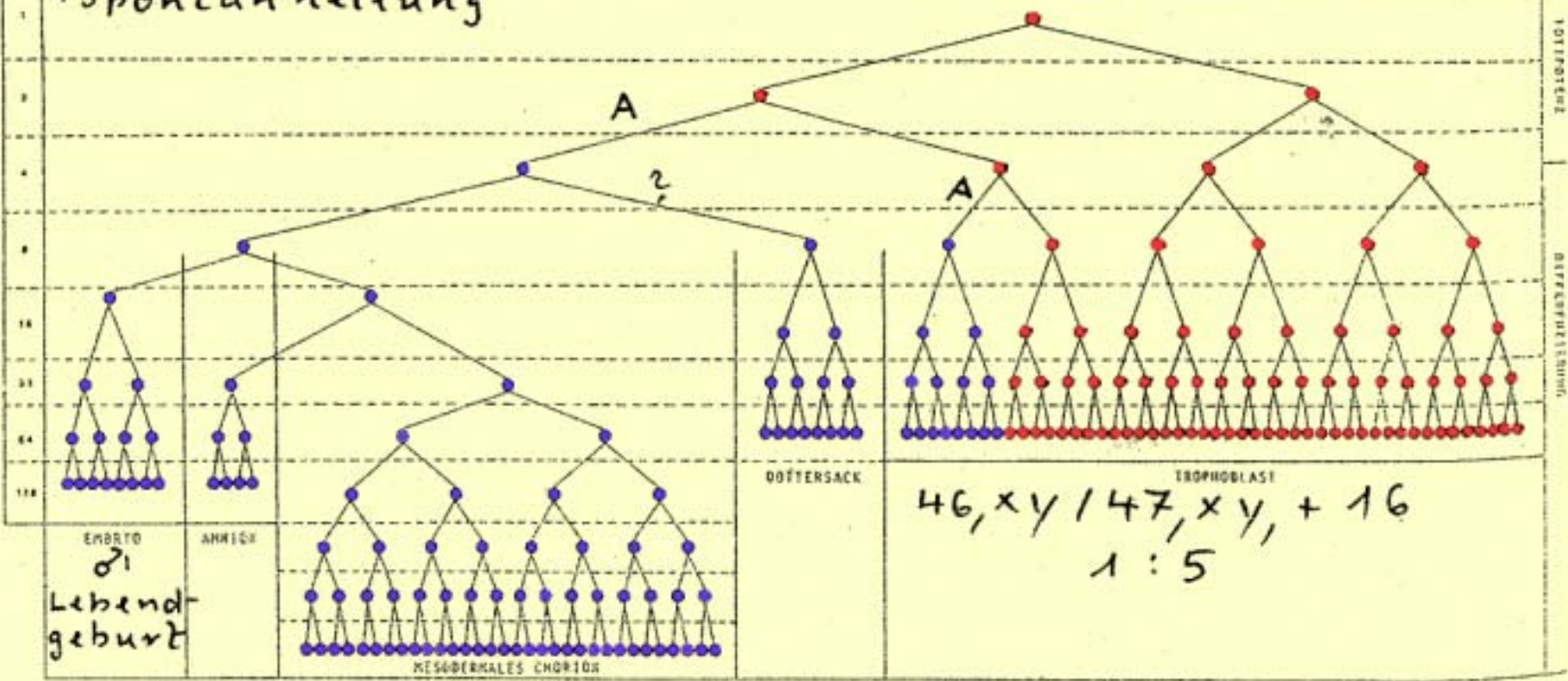
TROPHOBLAST

46, XX (n = 75)

Doc. Nr.: 00539

ZYGOTIC:  $47, xy, +16$

"Spontanheilung"



EMBRYO  
♂  
Lebendgeburt

ANNEX

MESODERMALES CHORION

DOTTERSACK

TROPHOBLAST

$46, xy / 47, xy, +16$   
1:5

TOTIPOTENZ  
DIFFERENZIERUNG



# Confined Placental Mosaicism

## CPM

Trisomy 7

IUFD is not a prominent feature

UPD present in 1/9

1/6 meiotic

5/6 mitotic

Kalousek et al 1996

# Confined Placental Mosaicism

## CPM

Trisomy 2 – 9 cases

IUFD is associated with high cell prevalence but only in cases ascertained for IUFD – growth catching up in those

UPD not found

Shaffer et al 1996



# Confined Placental Mosaicism CPM

Trisomy 16

IUFD is associated with high cell prevalence

UPD not as significant in effect on IUFD

Kalousek et al 1993

# Confined Placental Mosaicism CPM

101 cases of CPM – 88 with amnio followup

Normal distribution of birth weights – 12.3% below the 10<sup>th</sup> percentile

8 pregnancies with 13, 16, or 22 – 2 SAB; 75% birth weight below 10<sup>th</sup> percentile

Leschot et al 1996

# Confined Placental Mosaicism CPM

21,232 CVS 1983-1992 – Jefferson

0.8-2.0% CPM involving variety of chromosome

Compromised perinatal outcome increased

Correlation hampered by lack of placental study

# Confined Placental Mosaicism CPM

13,827 cases – 256 (1.9%) CPM – Germany

126,465 cases – 700 (5.5%) CPM-Eucromic

No increase in congenital malformations

11.1 vs 5.6% (control) low birth weight

Most catch up in growth

# Confined Placental Mosaicism

## CPM

Although CPM occurs at a significant level, the large number of individual chromosomes involved and the need for longitudinal study of the degree of CPM throughout placental development – suggest that a collaborative registry and protocol directed study will be needed to develop practical guidelines