

Vorlesung „Allgemeine Biologie für Informatiker und Mathematiker 1“

Teil Genetik 1

Wintersemester 2002/2003

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1. Vererbung und Variabilität

- 1.1 Umweltbedingte Variabilität**
- 1.2 Genetisch bedingte Variabilität**
- 1.3 Phänokopien**

2. Vererbung als biologisches Grundphänomen

- 2.1 Die Mendelschen Regeln**
- 2.2 Mendel aus heutiger Sicht**
- 2.3 Ergänzungen zu den Mendelschen Regeln**

3. Die Chromosomentheorie der Vererbung

- 3.1 Die eukaryotische Zelle**
- 3.2 Der Zellzyklus – Mitose**
- 3.3 Der Zellzyklus – Meiose**
 - 3.3.1 Meiose I**
 - 3.3.2 Meiose II**
- 3.4 Lebenszyklen von Eukaryoten**
- 3.5 Das eukaryotische Chromosom – Crossing over und Kartierung**
- 3.6 Der Karyotyp**
- 3.7 Geschlechtschromosomen**
- 3.8 Polytäne Chromosomen (Riesenchromosomen)**
- 3.9 Extrachromosomale Gene**

4 Grundlagen menschlicher Vererbung

4.3 Stammbaumforschung

4.4 Erbkrankheiten – geschlechtsgebundene Gene

4.5 Menschliche Erbkrankheiten – Autosomale Gene

4.5.2 Rezessive Allele

4.5.3 Dominante Allele

4.6 Genetische, meist nichterbliche Krankheiten

4.6.2 Autosomale Aberrationen

4.6.3 Geschlechtschromosomenaberrationen

4.7 Genetische Familienberatung

4.8 Früherkennung von Erbkrankheiten

4.8.2 Cytologische Untersuchungen

4.8.3 Biochemische Methoden

4.9 Dosiskompensation – die Lyon-Hypothese

4.10 Genomische Prägung (Imprinting)

4.11 Genomprojekte

5. Molekulare Grundlagen der Vererbung

5.1 DNA als Träger der Erbinformation

5.2 Die Verdopplung des Erbmaterials (Replikation)

5.3 DNA-Schäden und Reparatur

5.3.1 Die Photoreaktivierung

5.3.2 Die Exzisionsreparatur

5.3.3 Die Nachreplikationsreparatur

5.3.4 Die SOS-Reparatur

5.4 Defekte im Reparatursystem

5.5 Mutationen

5.6 Mutagenitätstests

5.7 Veränderung des genetischen Materials durch Rekombination

5.7.1 Durch allgemeine Rekombination entsteht Crossing-over zwischen Chromosomen

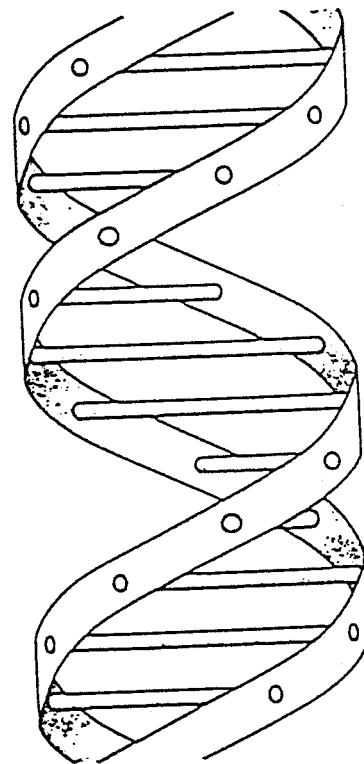
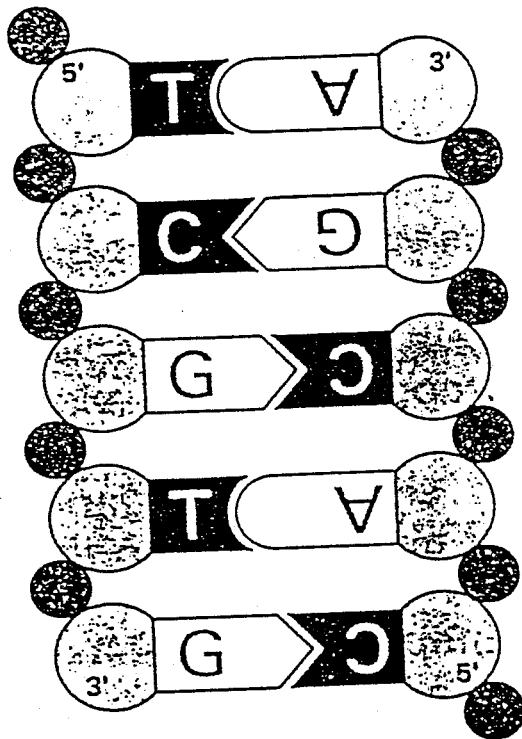
5.7.2 Sequenzspezifische Rekombination zwischen ringförmigen DNA-Molekülen

5.7.3 Transposition

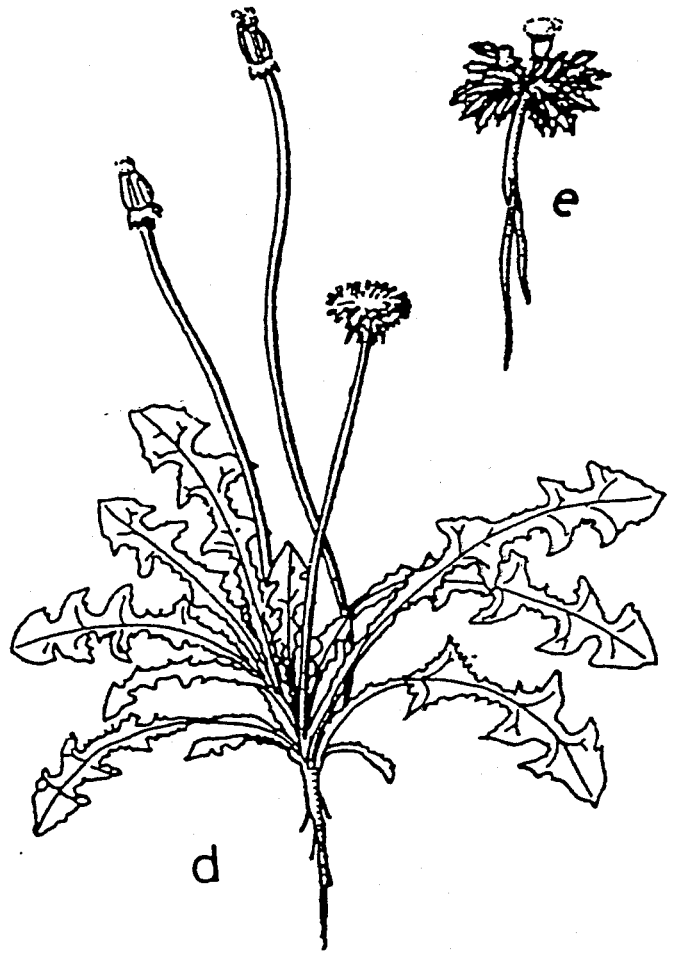
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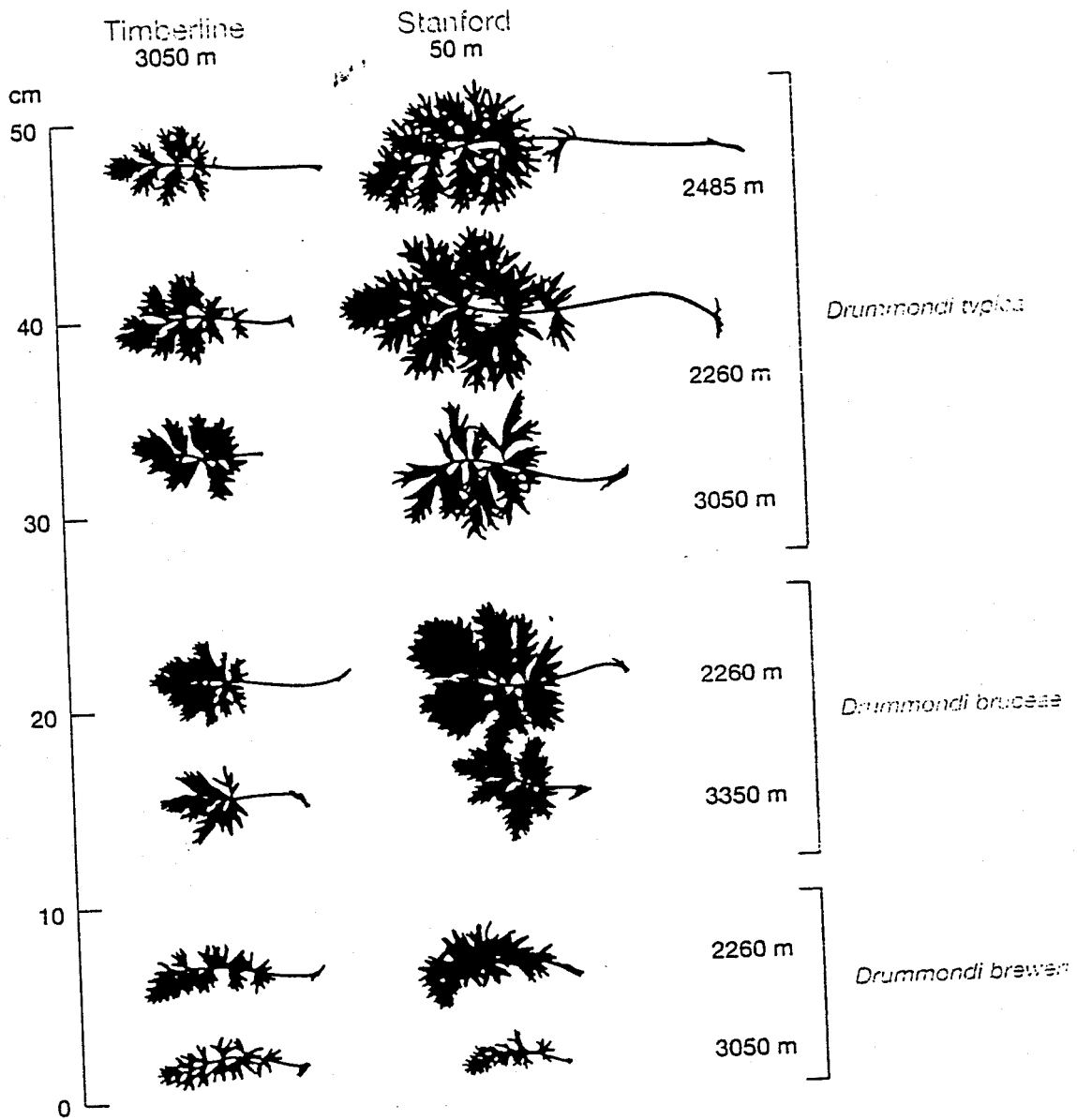






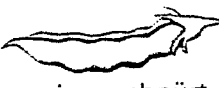



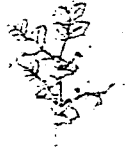
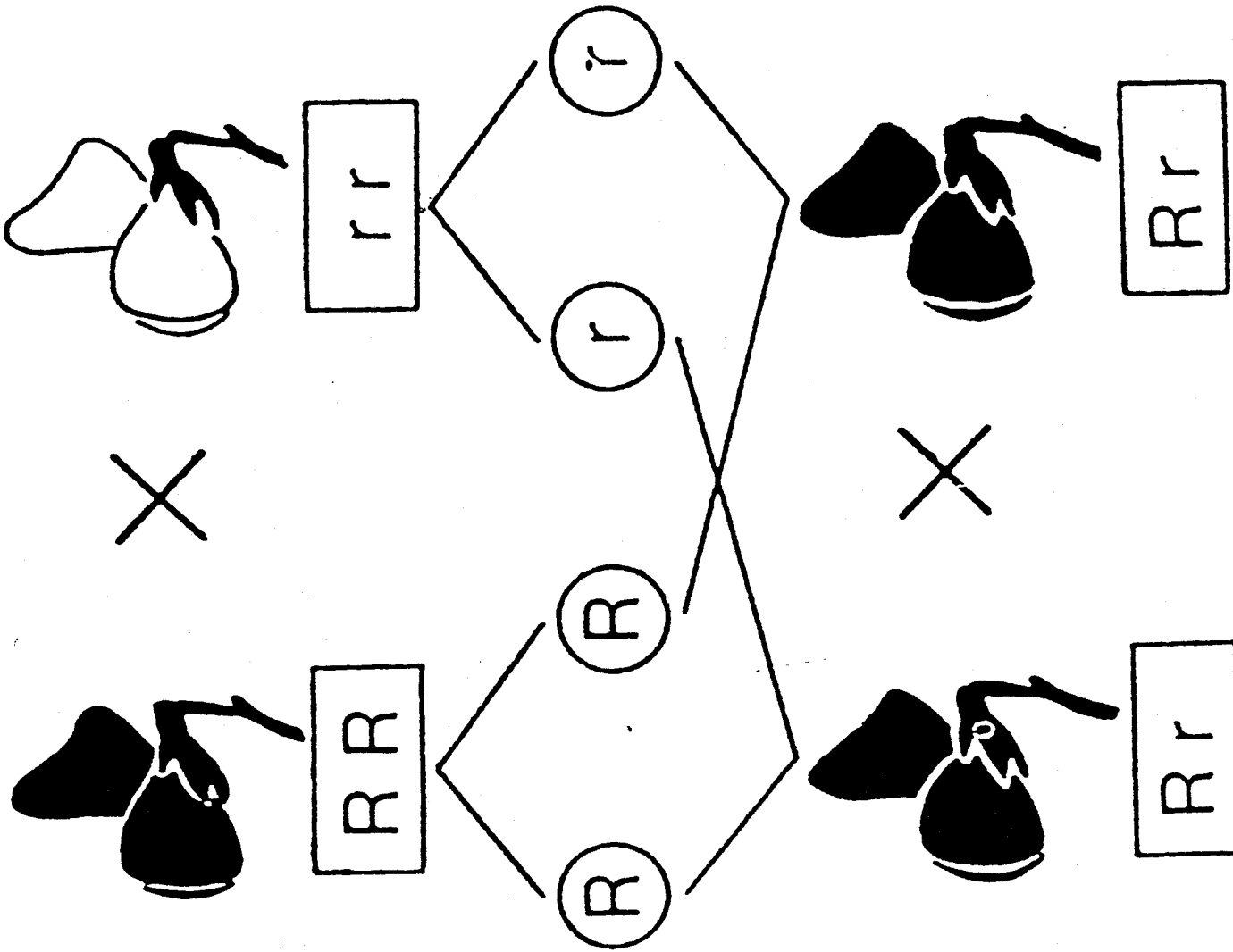


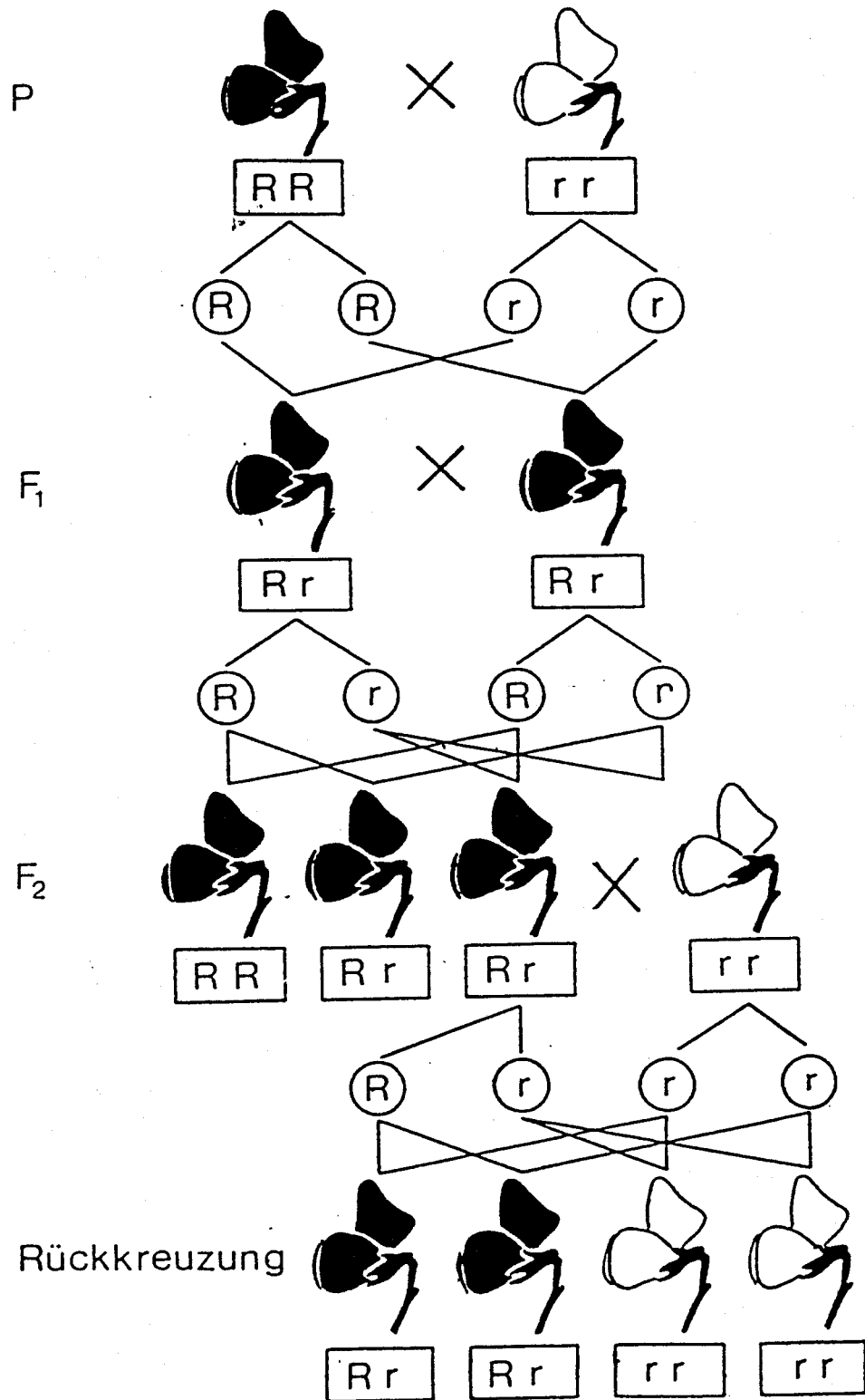
Tabelle 13.1: Die Ergebnisse der F₁-Kreuzungen Mendels mit sieben Merkmalen der Gartenerbse

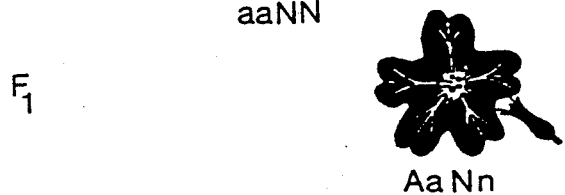
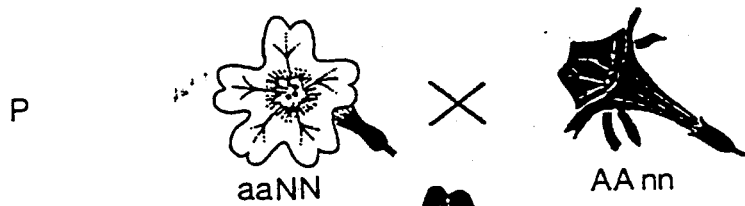
Merkmal	dominantes Allel	rezessives Allel	F ₂ -Generation dominant : rezessiv	Verhältnis
Blütenfarbe	 pupurfarben	 weiß	705 : 224	3,15 : 1
Blütenstellung	 axial	 terminal	651 : 207	3,14 : 1
Samenfarbe	gelb	 grün	6022 : 2001	3,01 : 1
Samenform	rund	runzlig	5474 : 1850	2,96 : 1
Hülsenform	 aufgebläht	 eingeschnürt	882 : 299	2,95 : 1
Hülsenfarbe	 grün	 gelb	428 : 152	2,82 : 1
Stengellänge			787 : 277	2,84 : 1

P



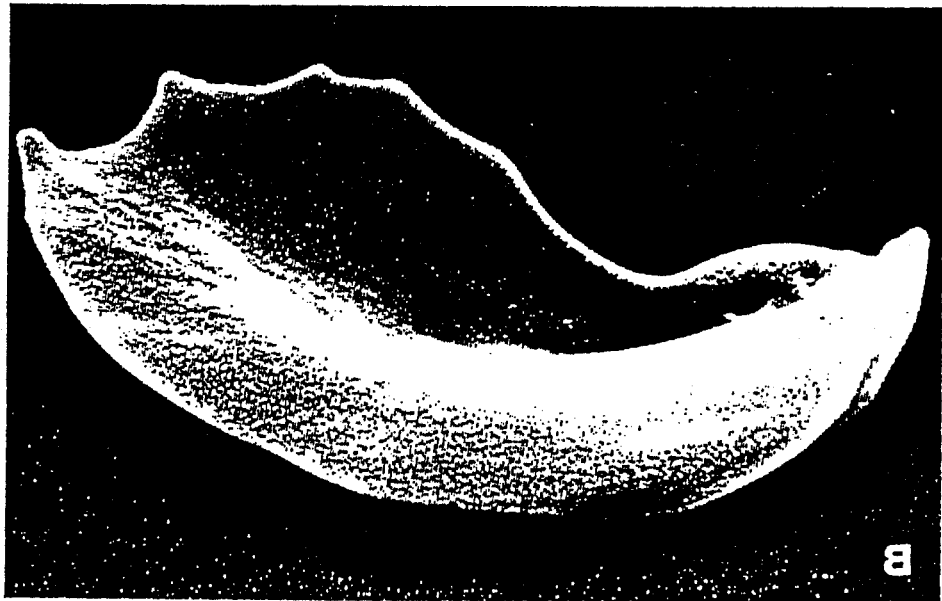
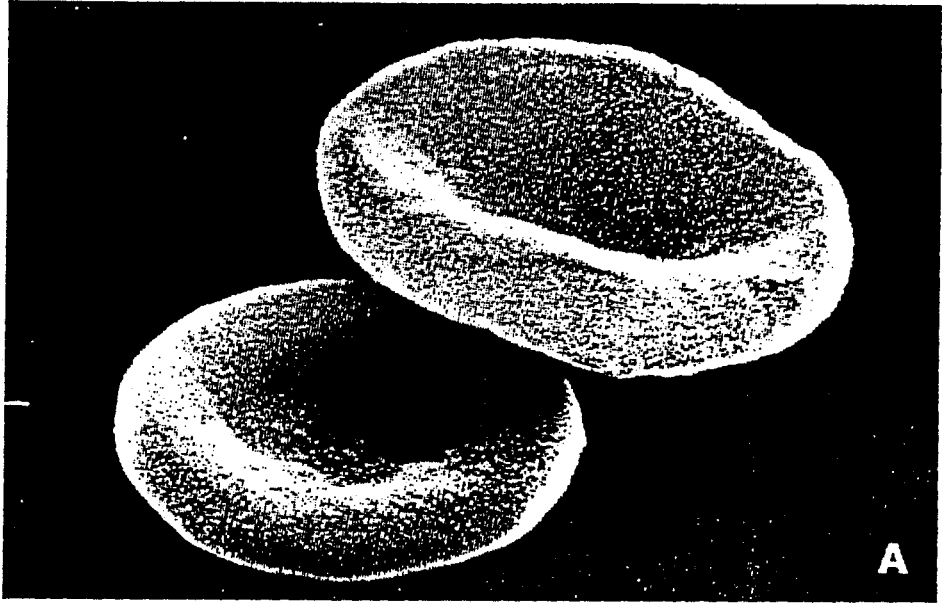
F1

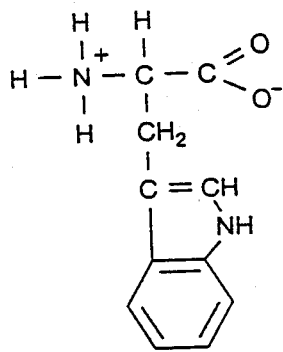




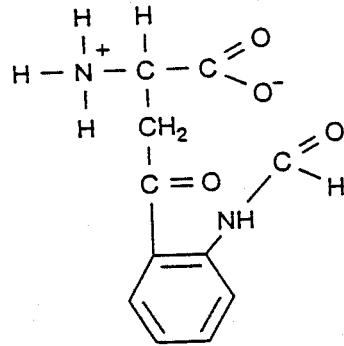
F ₂	AN	aN	An	an
AN	 AA NN	 Aa NN	 AA Nn	 Aa Nn
aN	 Aa NN	 aa NN	 Aa Nn	 aa Nn
An	 AA Nn	 Aa NN	 AA nn	 Aa nn
an	 Aa Nn	 aa Nn	 Aa nn	 aa nn

Genotyp	Oberflächen- Antigen	Antikörper- bildung
I^A/I^A oder I^A/I^0	A	Anti-B
I^B/I^B oder I^B/I^0	B	Anti-A
I^A/I^B	A und B	keine
I^0/I^0	keine	Anti-A und Anti-B

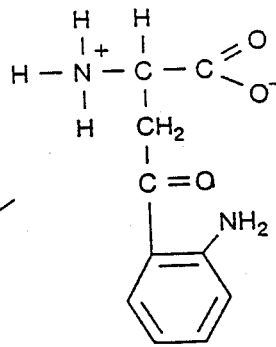




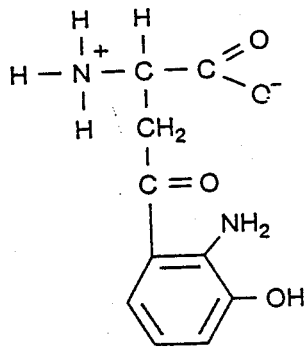
L-Tryptophan



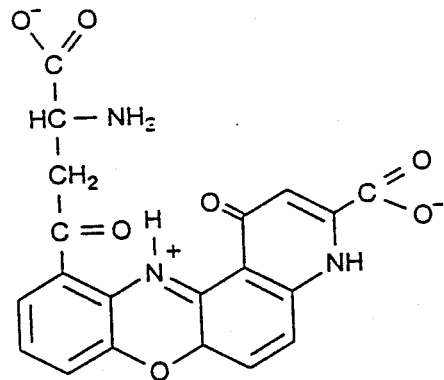
N-Formylkynurenin



Kynurenin

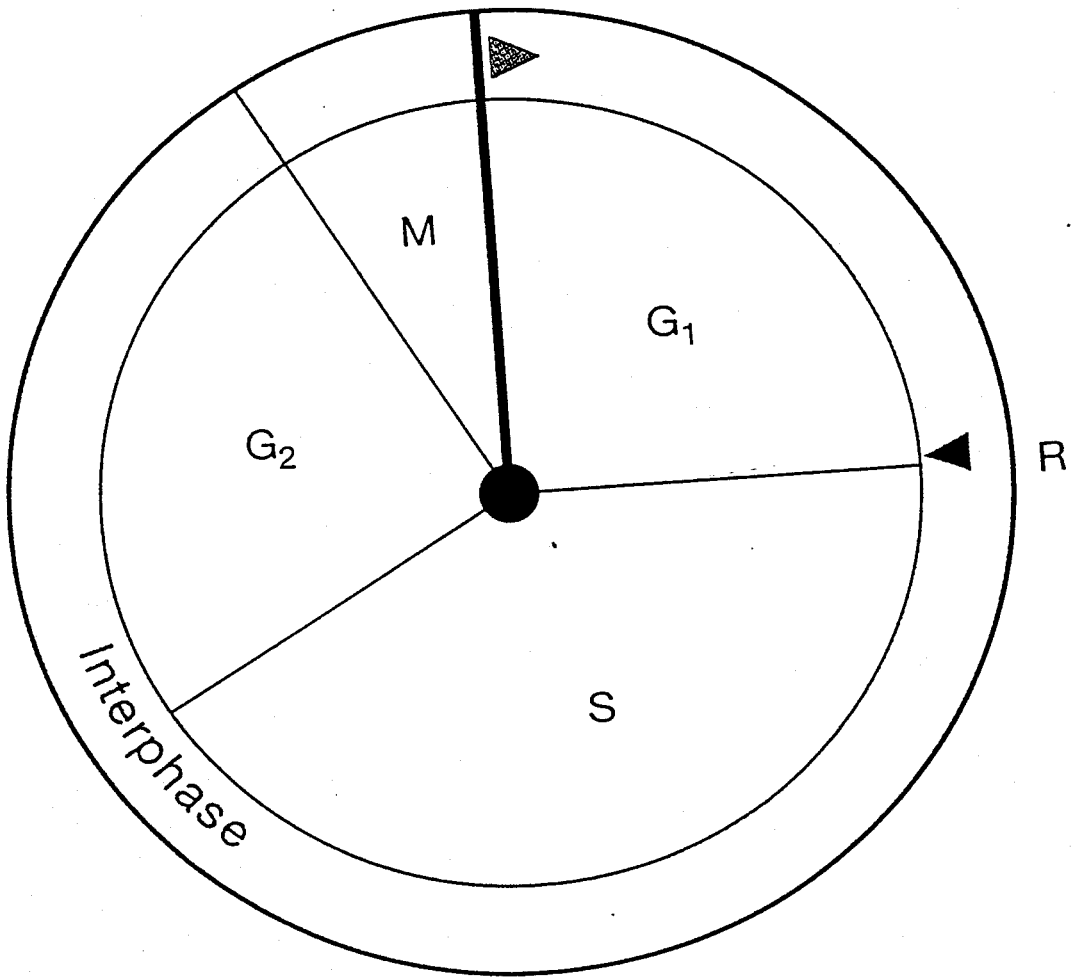


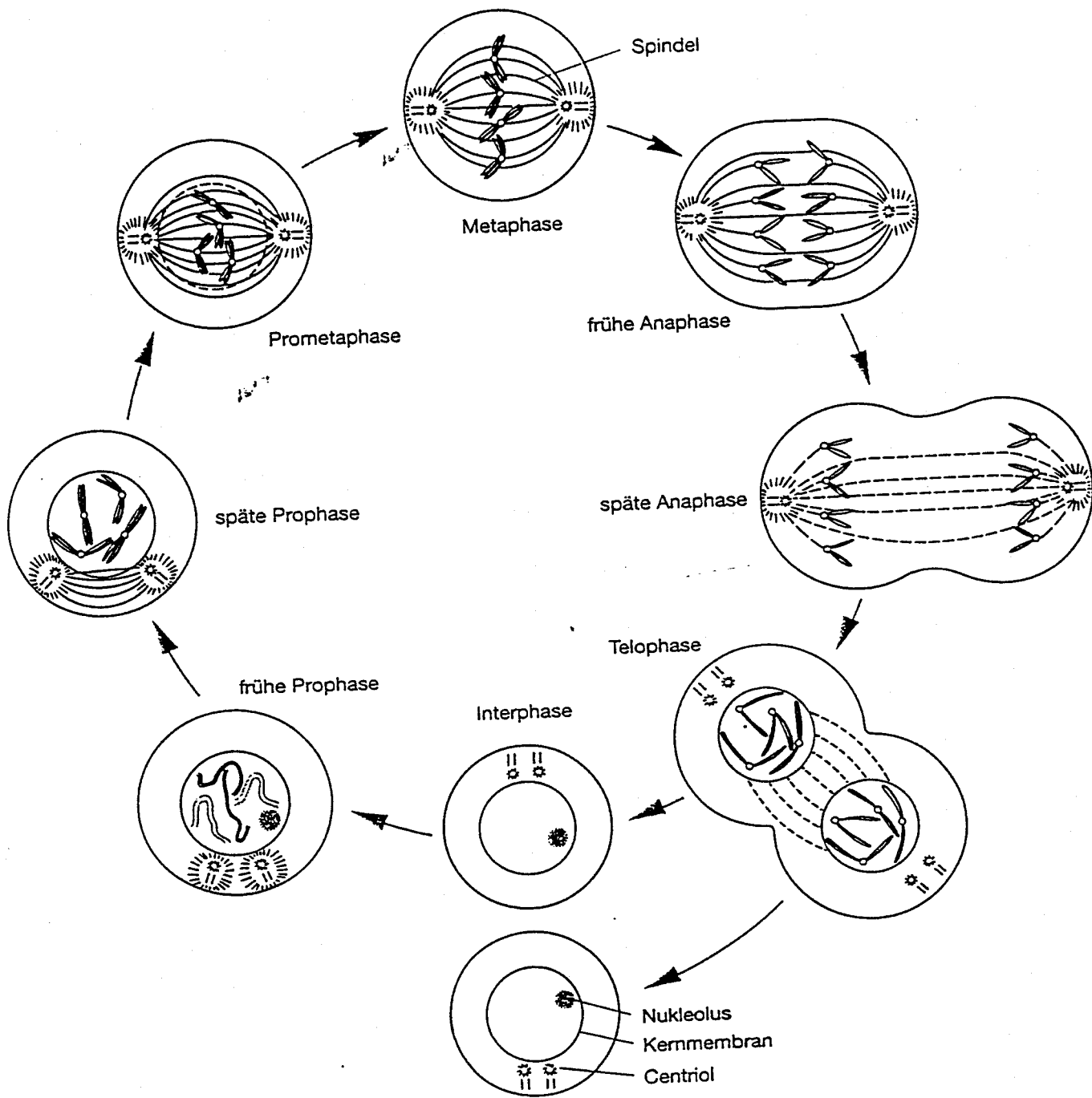
3-Hydroxykynurenin

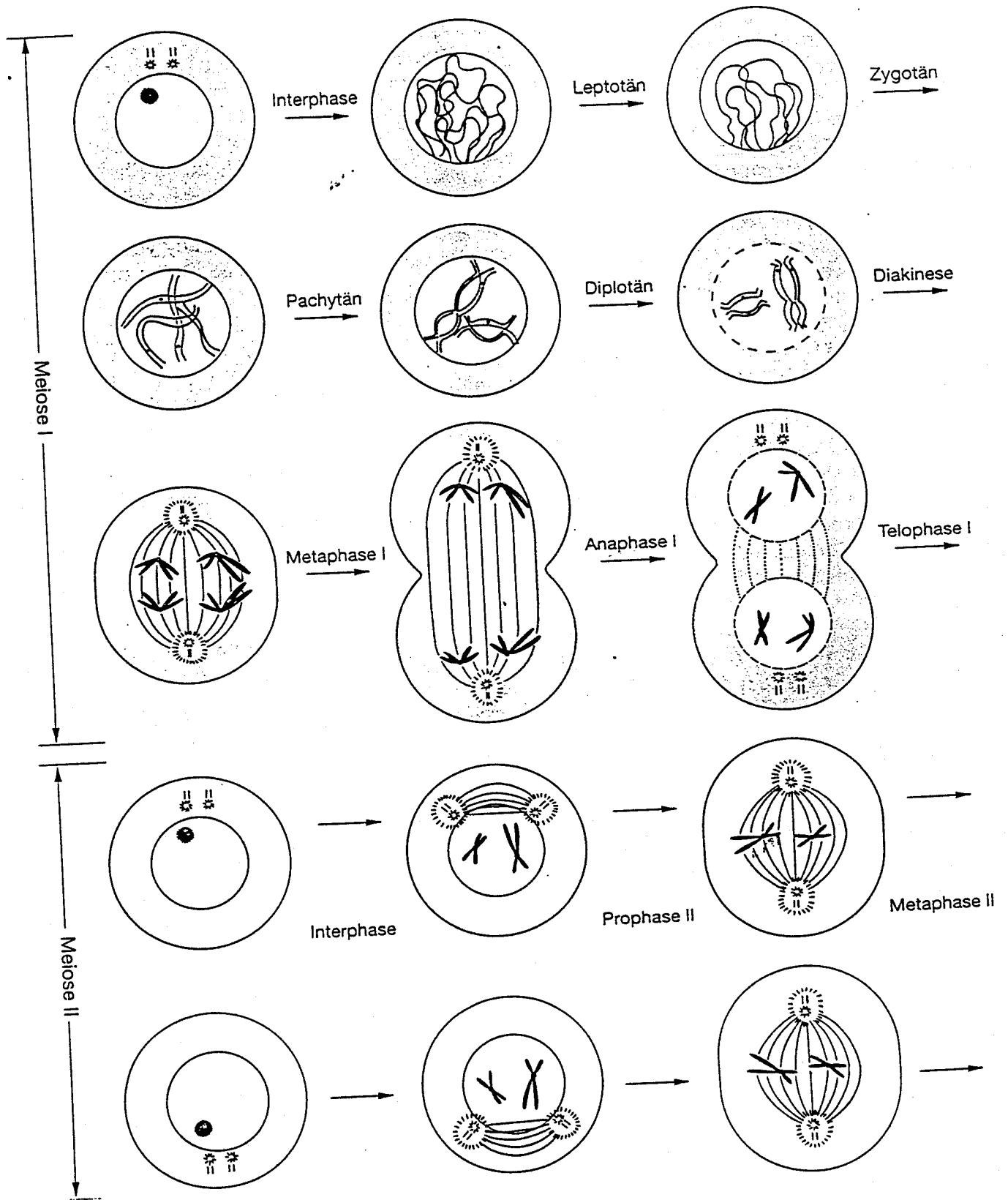


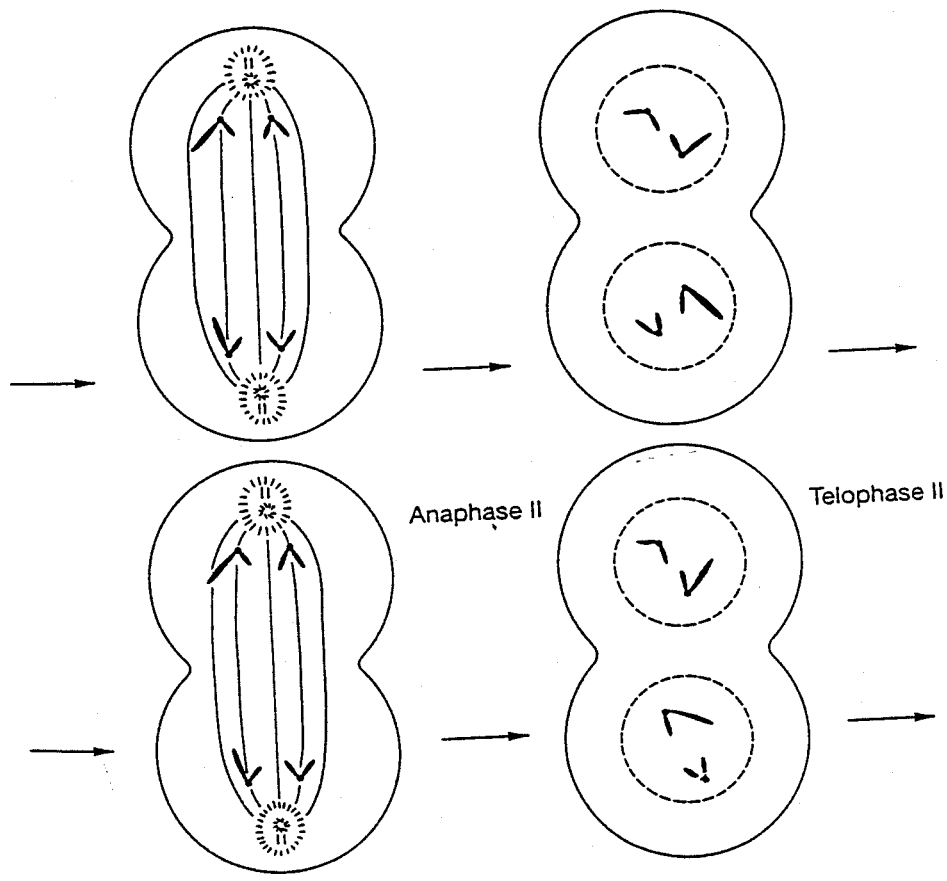
Xanthommatin

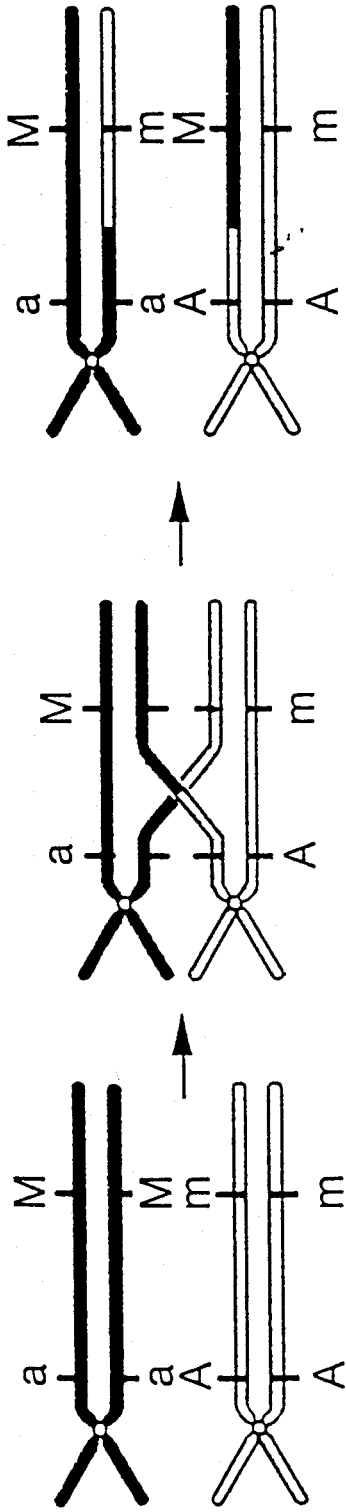




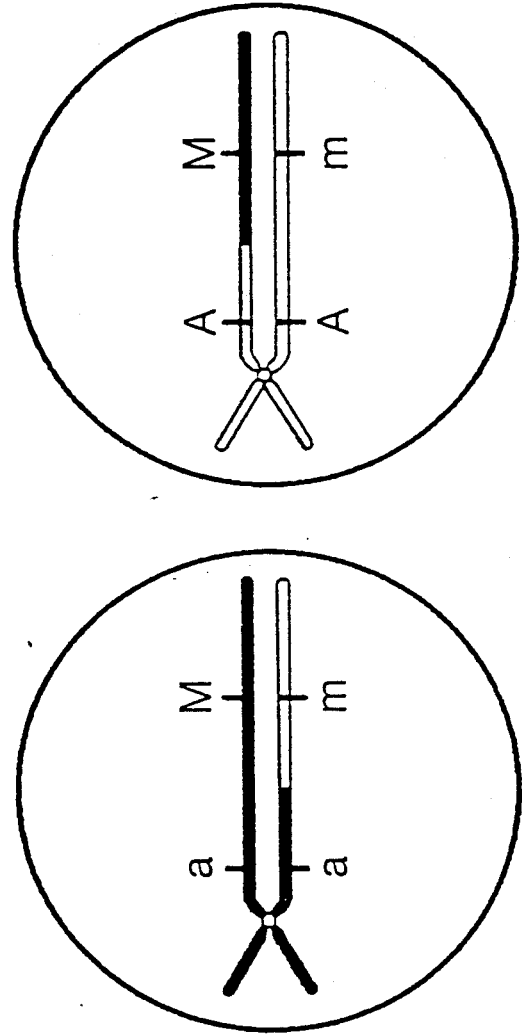




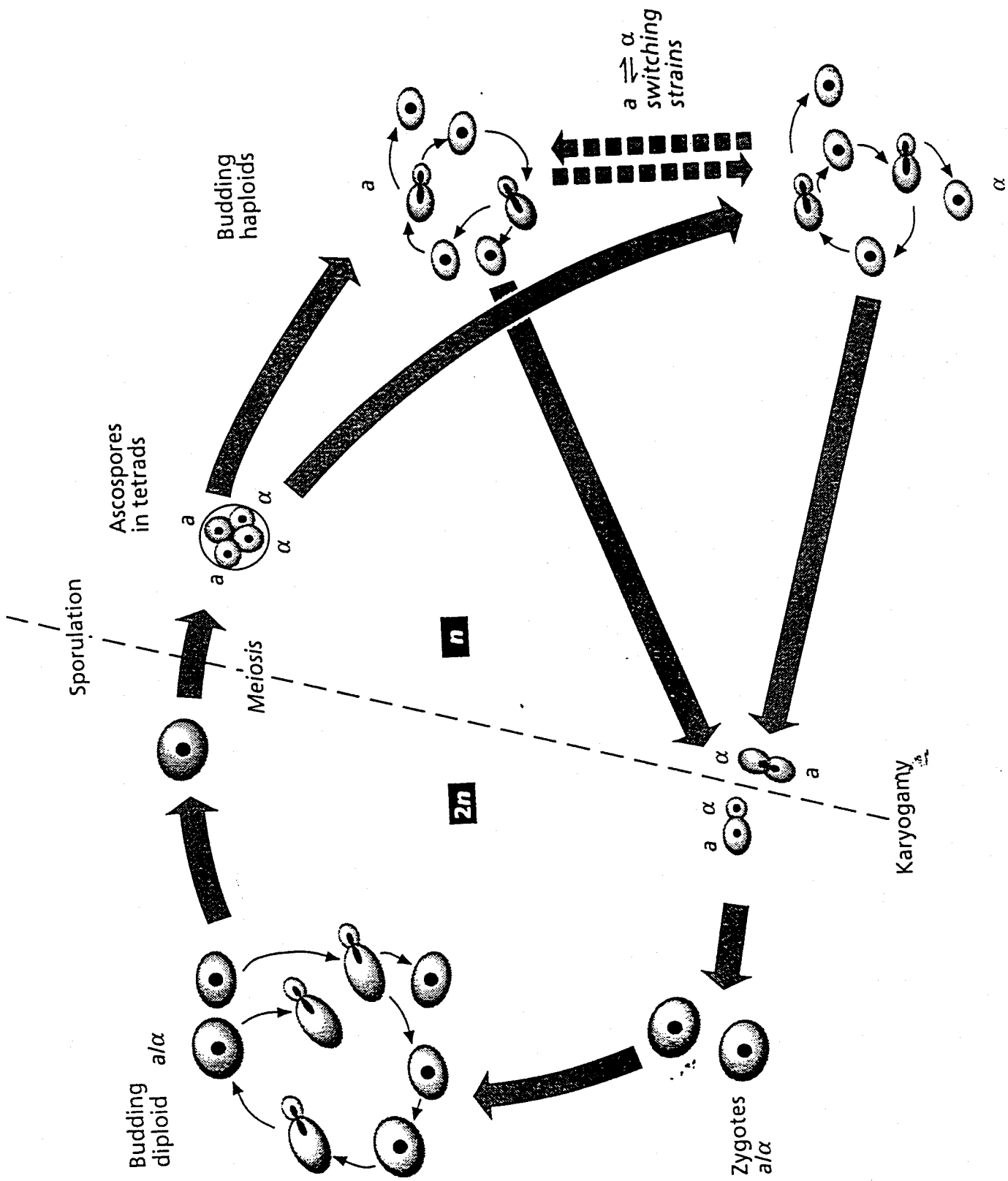


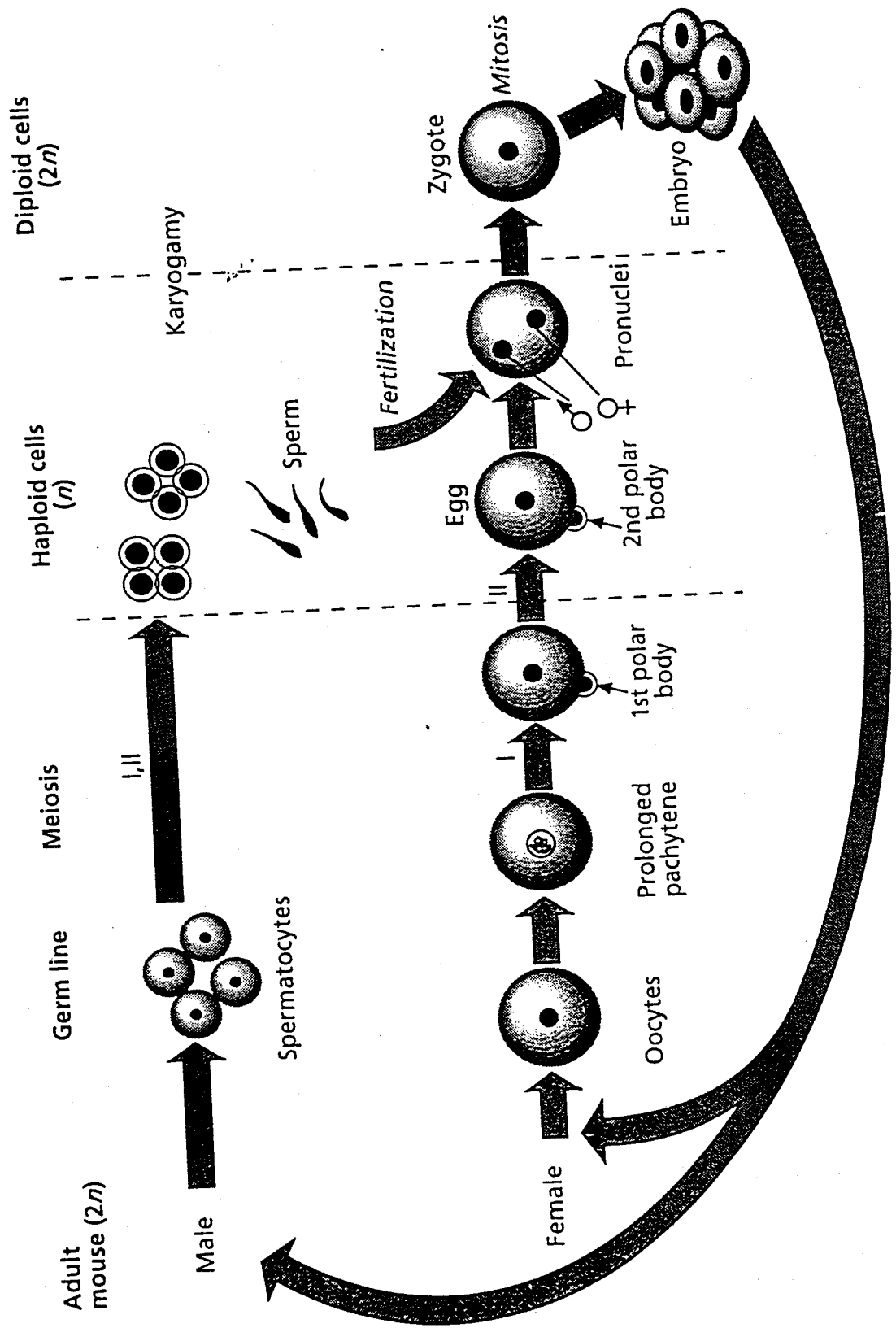


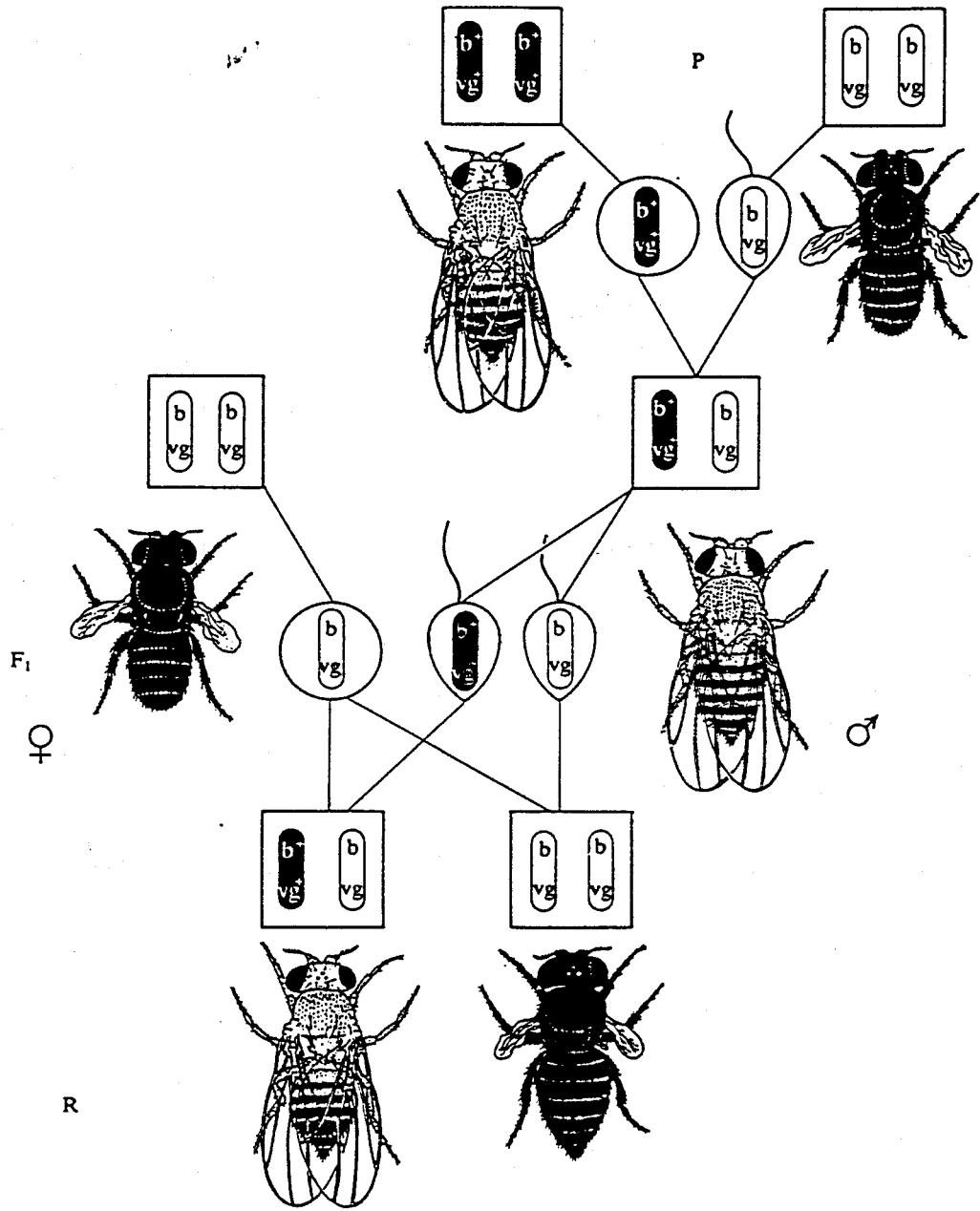
meiotische Prophase I

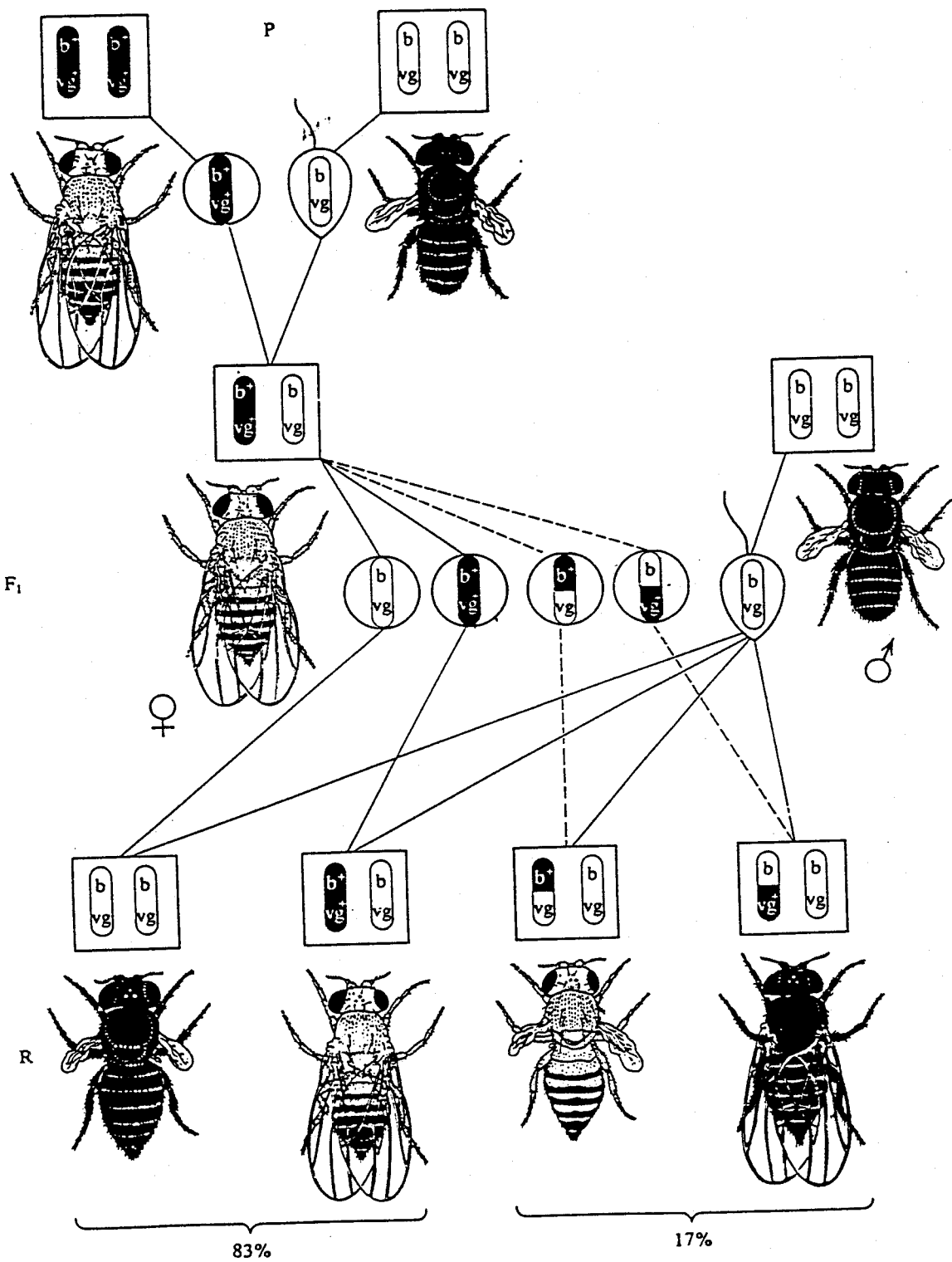


sekundäre Meiozyten









Mutter	w^+	m^+	fw
	w	m	fw^+

Nach Rückkreuzung
 gefundene
 Gametentypen

142 = 68,27%
 Parentaltypen ohne Austausch

2. w	m	$+$	fw
3. $+$	m	$+$	fw
4. w	$+$	fw	
5. $+$	$+$	$+$	fw
6. w	m	fw	
7. w	$+$	$+$	fw
8. $+$	m	fw	

59 = 28,37%
 Rekombinantenklasse I:
 (einfacher) Austausch von w

4 = 1,92%
 Rekombinantenklasse II:
 (einfacher) Austausch von fw

3 = 1,44%
 Rekombinantenklasse III:
 doppelter Austausch

Daraus Austauschhäufigkeiten:

w/m (Typen 3 + 4 + 7 + 8)	= 29,81%
m/fw (Typen 5 + 6 + 7 + 8)	= 3,36%
w/fw (durch Addition)	= 33,17%
w/fw (unmittelbar 3 + 4 + 5 + 6)	= 30,29%
Differenz	2,88%

Rh-null ?
Neuroblastom
Porphyrie

Leukämie-1

N-ras

PK-Defizienz
Lupus erythromat.
Faktor V

Faktor XIII B
Faktor H

1

Muskeldystrophie
GM2-Gangliosidosis
Xeroderma pigm. C
Faktor VII
c-fms

5

Albinismus
Leukämie, ak. lymphobl.
Galactosämie

Leukämie-2
c-abl
Xeroderma pig., A
Porphyrie

9

Duchenn. Muskeld.
Retinoblastom

Faktor VII
Faktor X

13

c-erbA
Galactosidase-Def.

17

Alzheimer
Schizophrenie

21

c-myc

Colon Cancer

Xeroderma pigm. B

Albinismus

Immunoglobulin L-Kette

2

Faktor XIII A

c-myb

6

Severe comb. Immunod.

Hexokinase-Defizienz
Cockayne Syndrom

Hexokinase-Defizienz

Leukämie, ak. T-Zell
Porphyrie, cong, erythr.

10

Immunoglobulin H-Kette

14

Protoporphyrin

18

Immunoglobulin L-Kette
c-bcl
c-sis

22

c-raf

GM1-Gangliosid

Schizophrenie ?

3

c-erbB

Cystische Fibrose

7

β -Globingene
c-H-ras
Wilms Tumor
Leukämie, ak. T-Zell

Albinismus

11

Albinismus
Prader-Willi-Syindr.
Marfan Syndrom
Tay-Sachs-Syndrom
GM2-Gangliosid
Tyrosinase

15

Severe comb. Immunod.

19

Y

Huntington
PKU DHPR

Leukämie, ak. lym.

Faktor XI

4

c-mos
c-myc

8

c-K-ras

Phenylketonurie

12

α -Globin

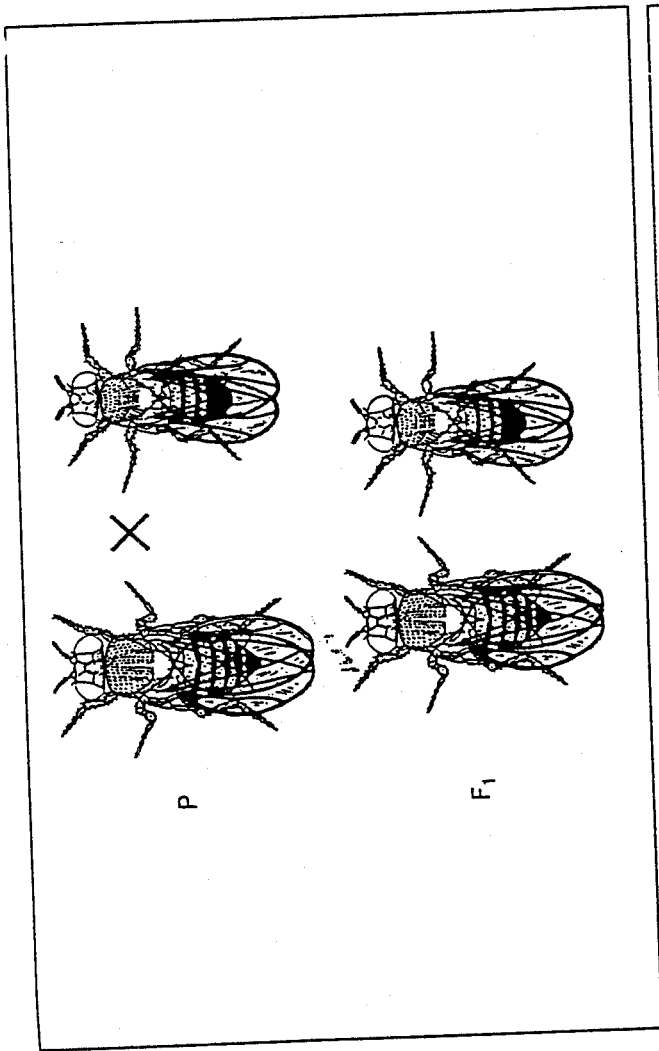
c-fes

16

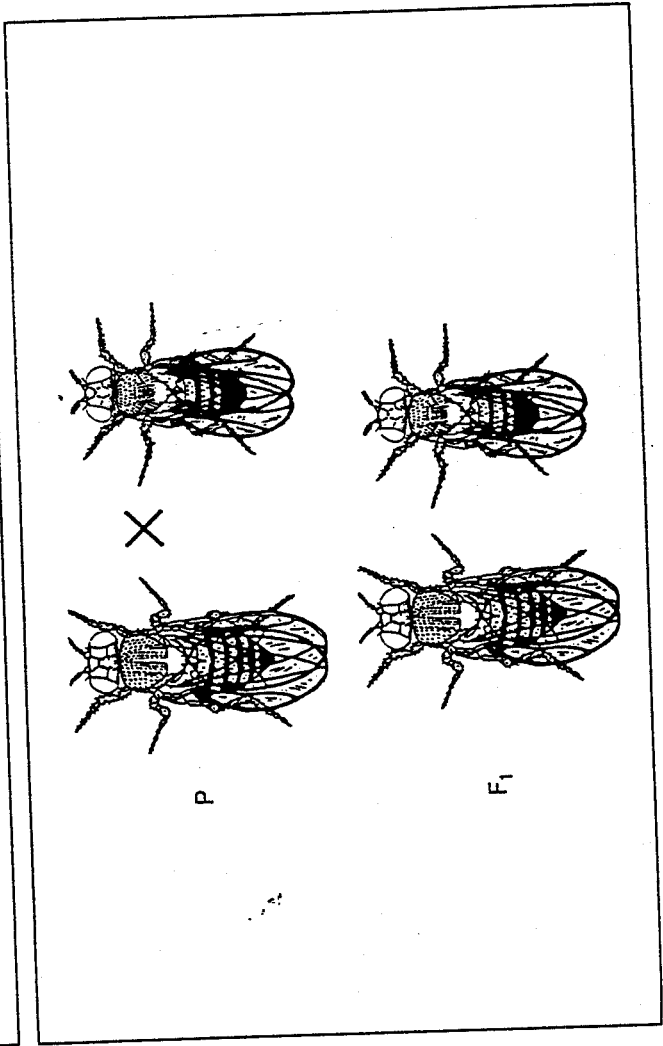
c-src

20

X



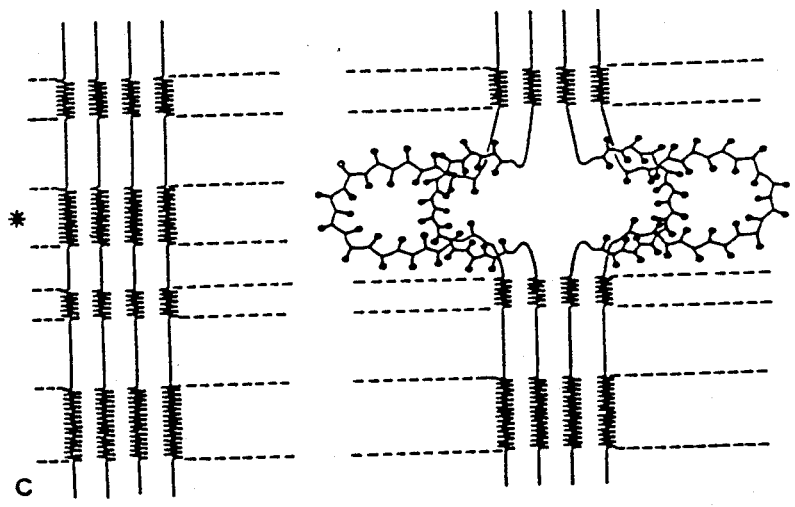
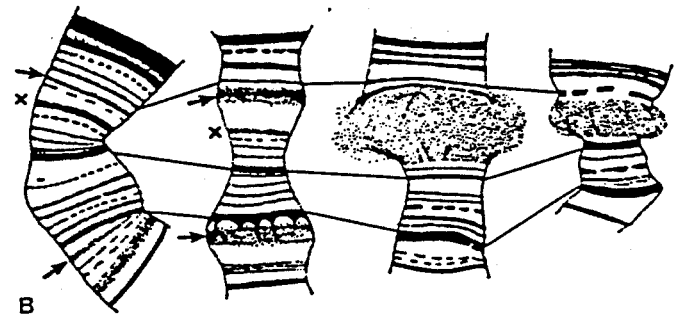
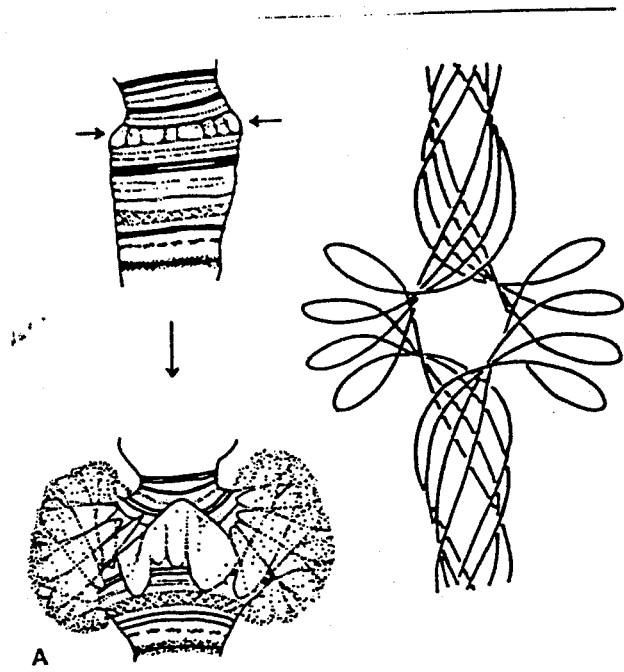
	W	Y
w ⁺	w ⁺ W	w ⁺ Y
w ⁺	w ⁺ W	w ⁺ Y



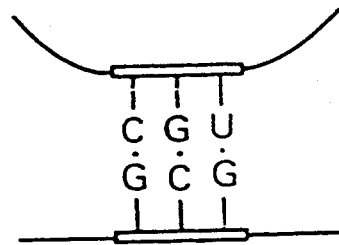
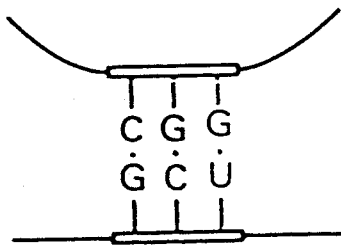
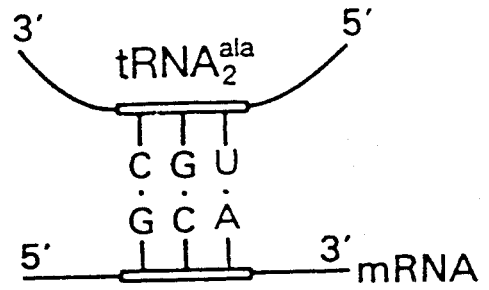
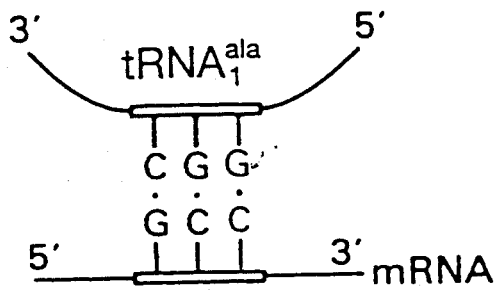
	w ⁺	Y
w	ww ⁺	wY
w	ww ⁺	wY

B

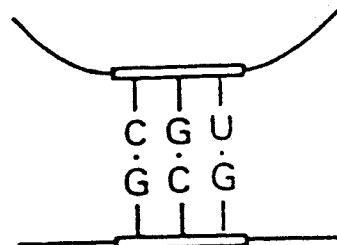
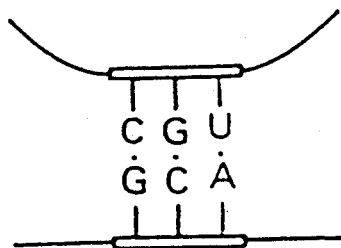
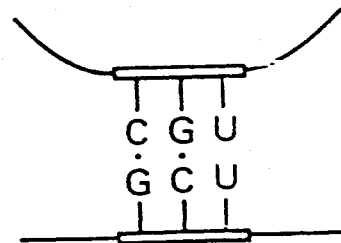
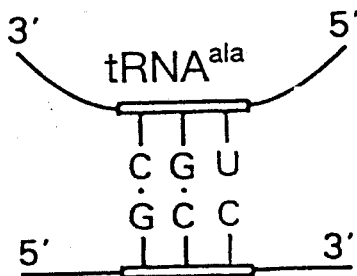
A

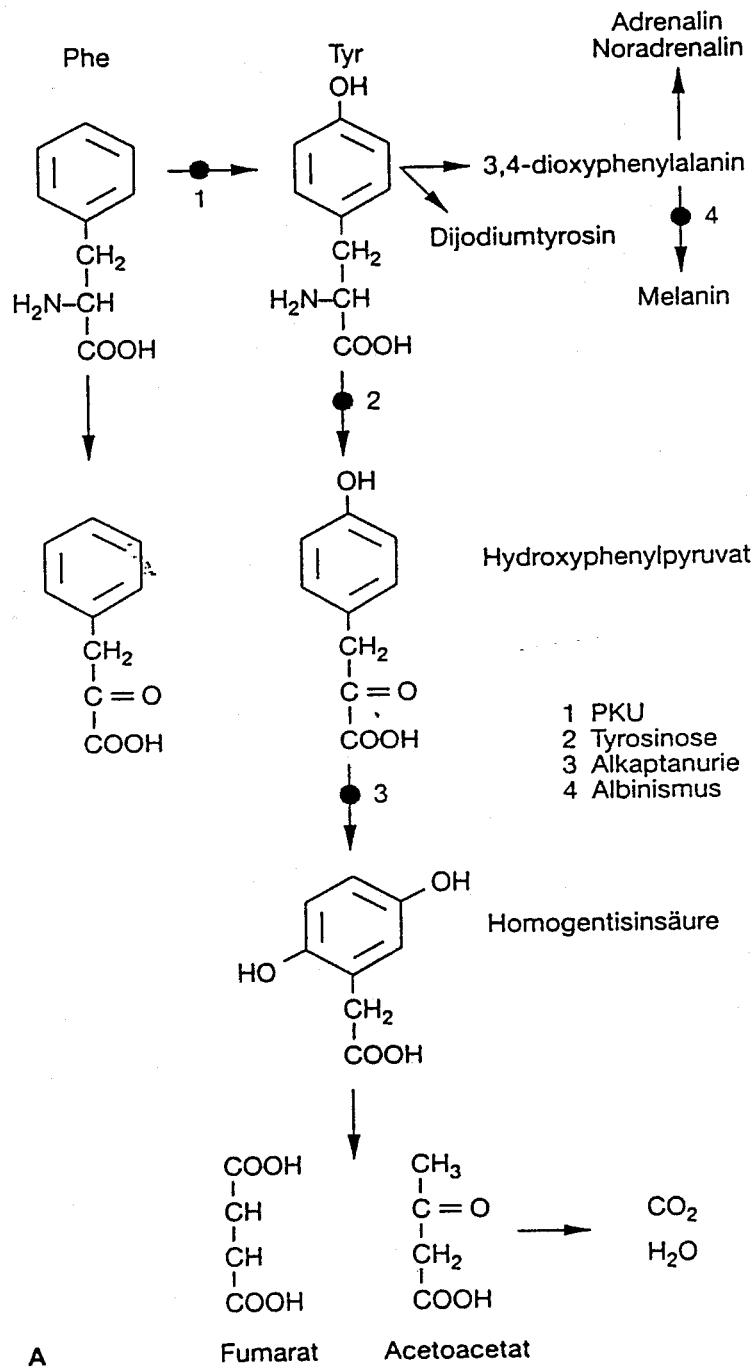


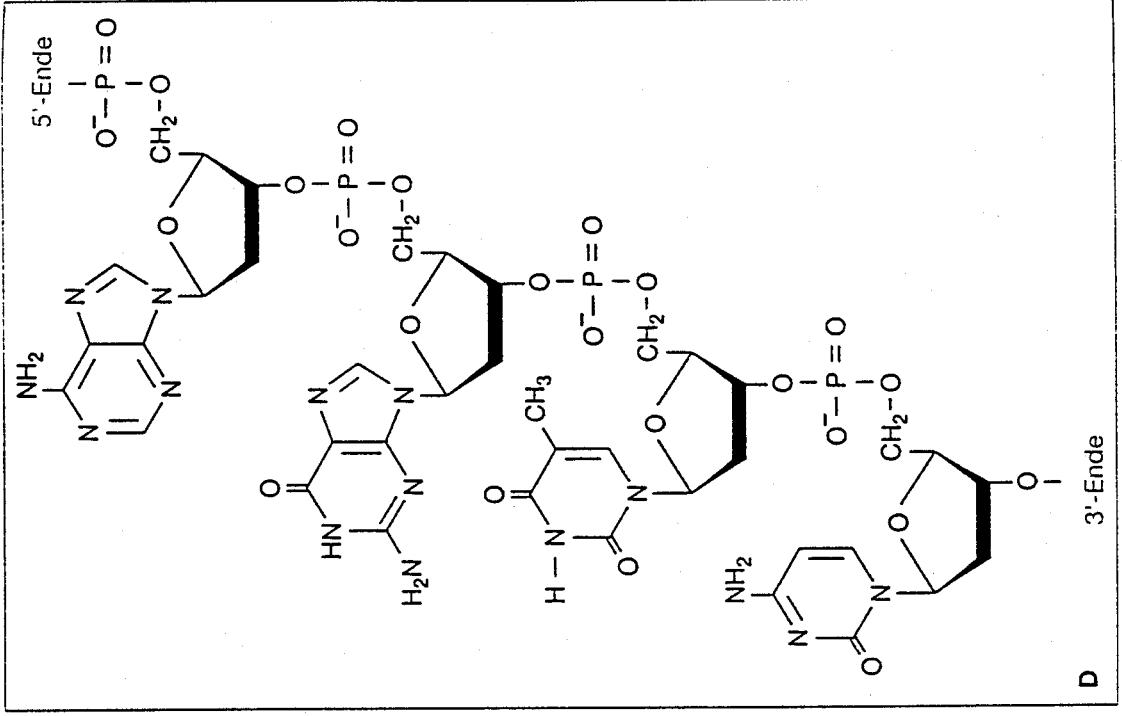
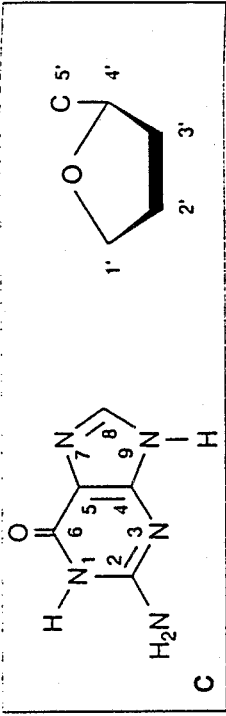
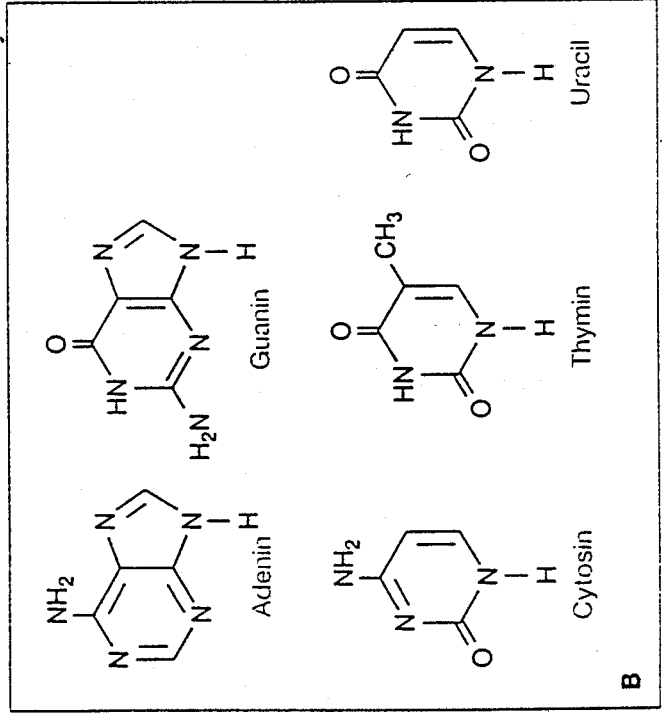
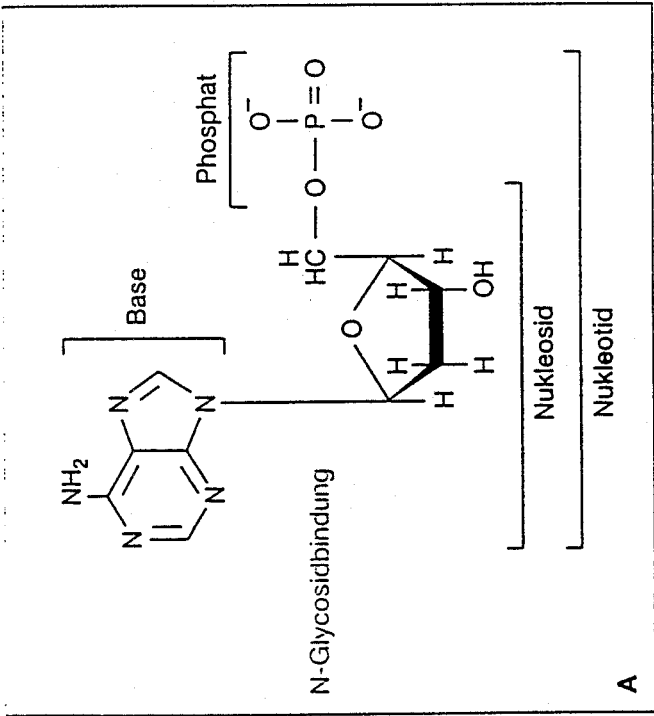
a) Standard-„Wobble“-Regeln

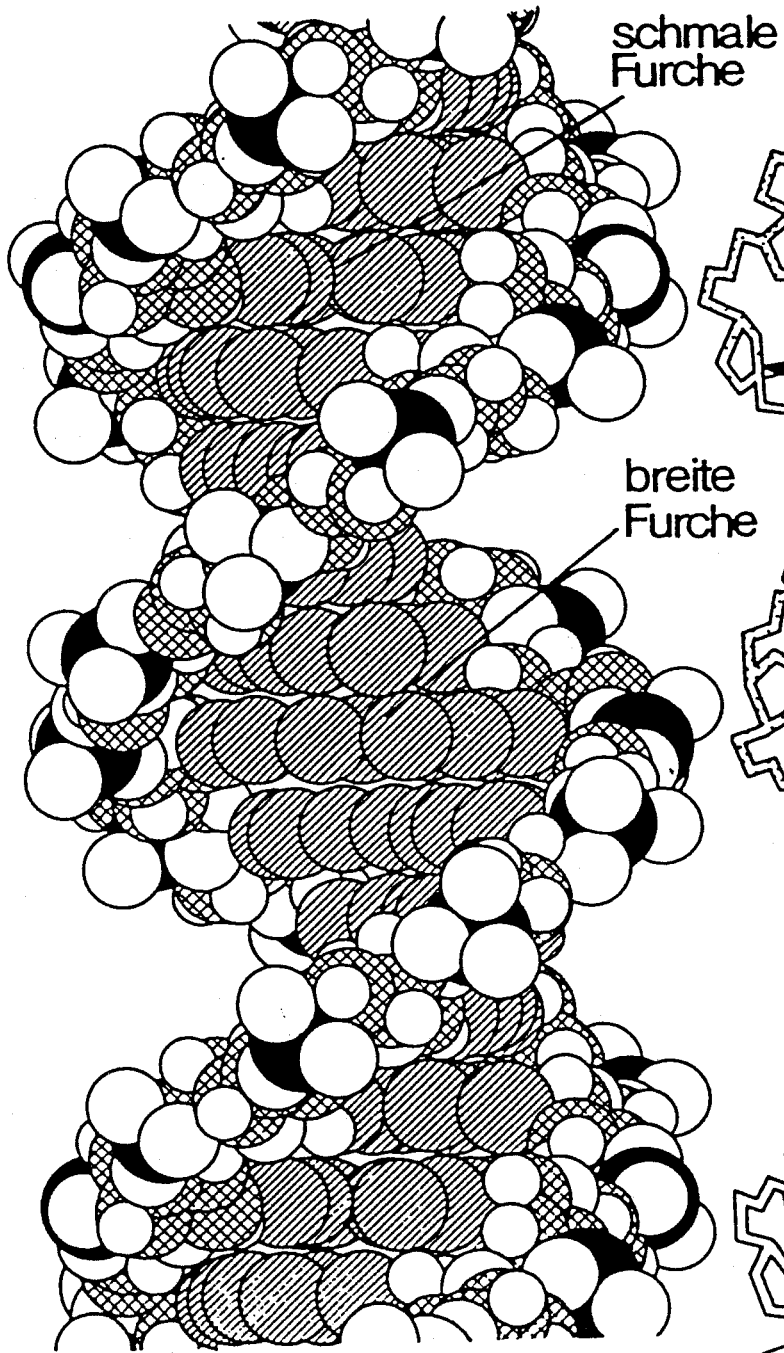


b) „Superwobble“ in menschlichen Mitochondrien

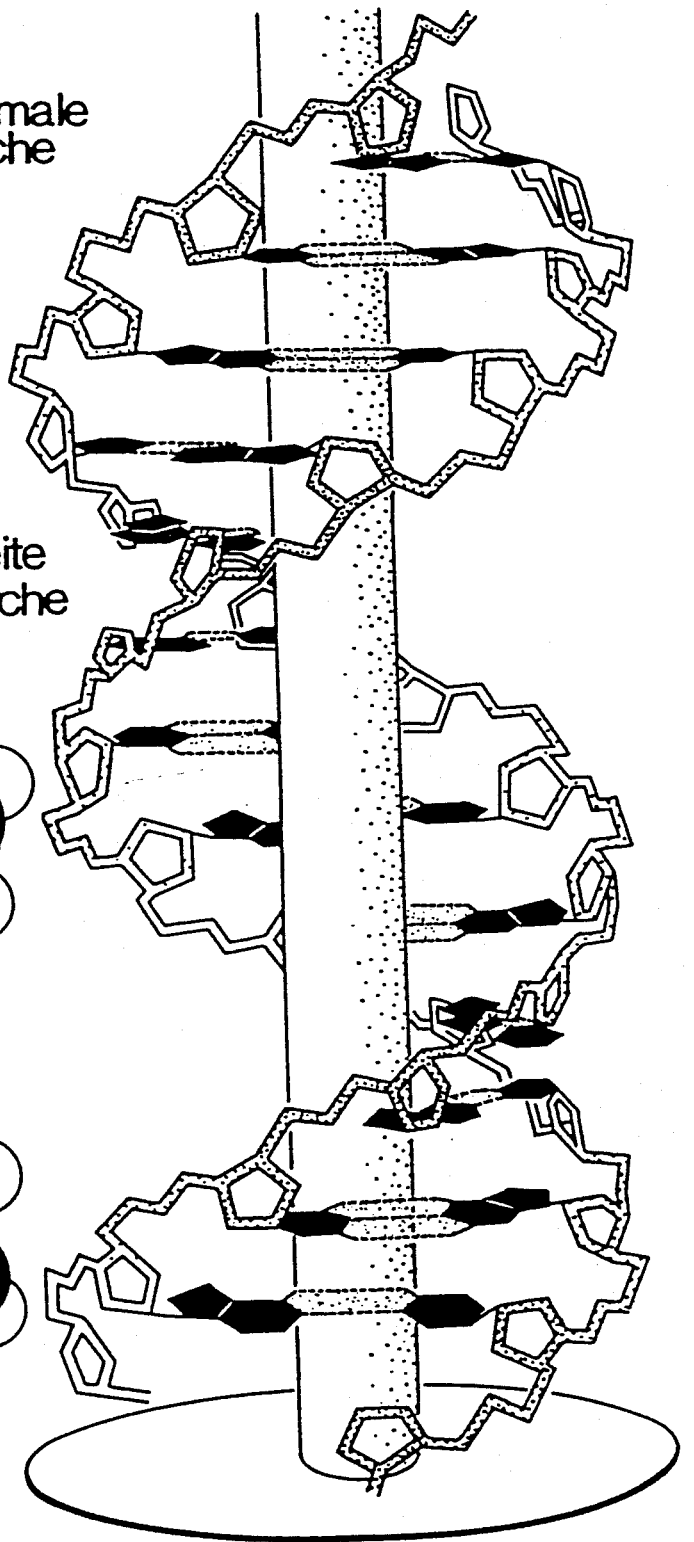




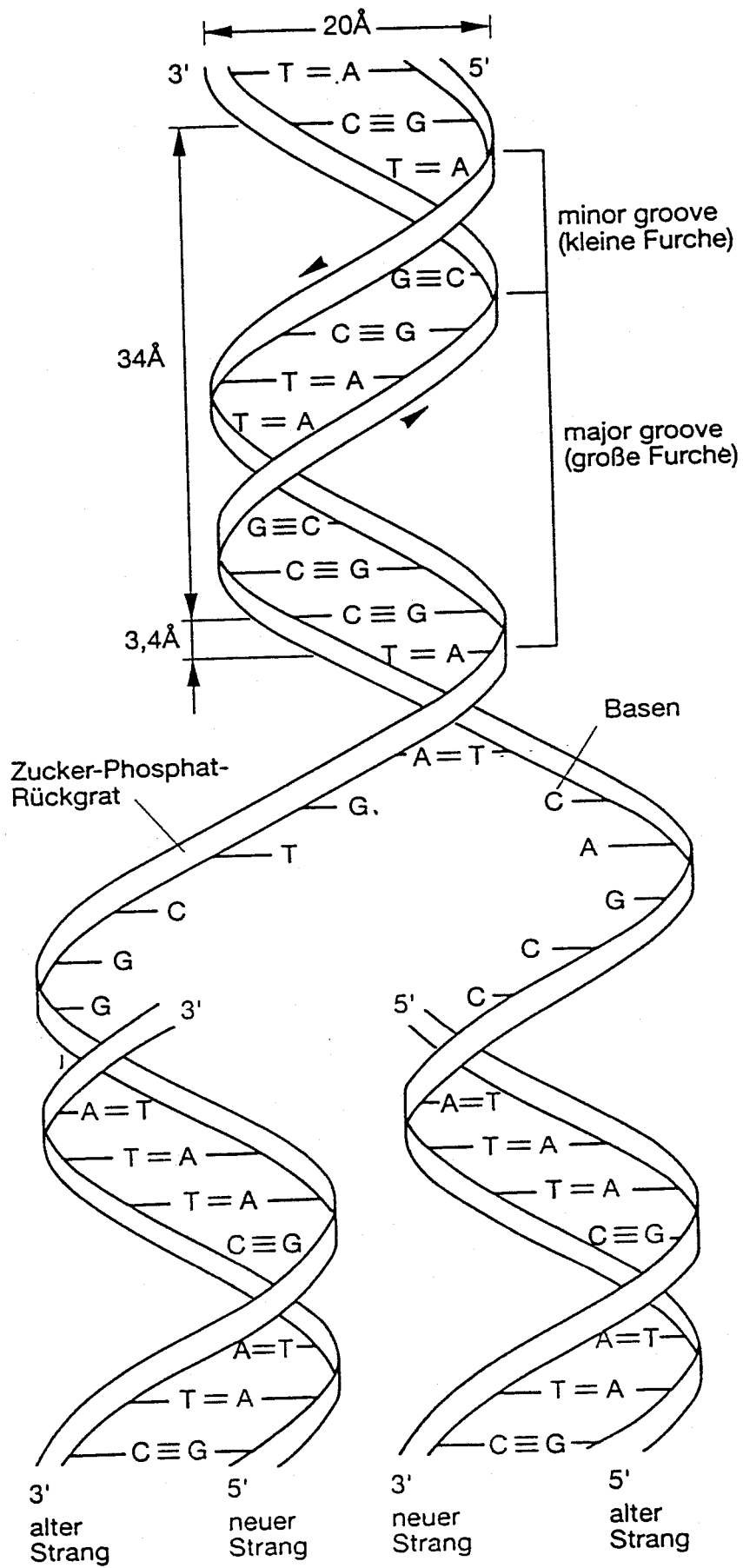


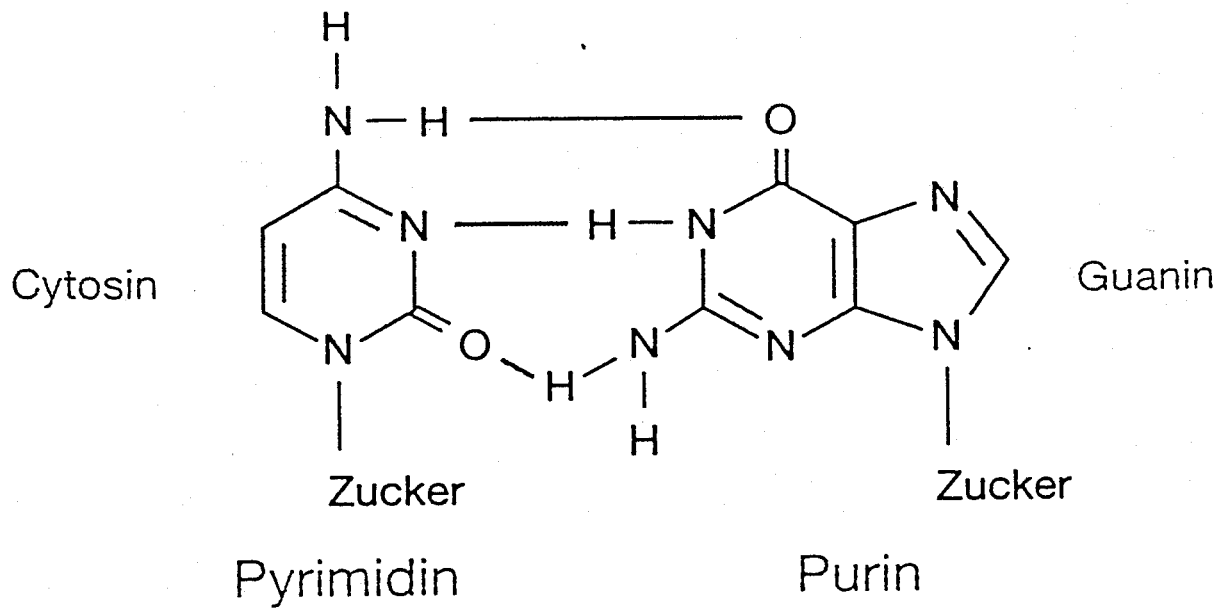
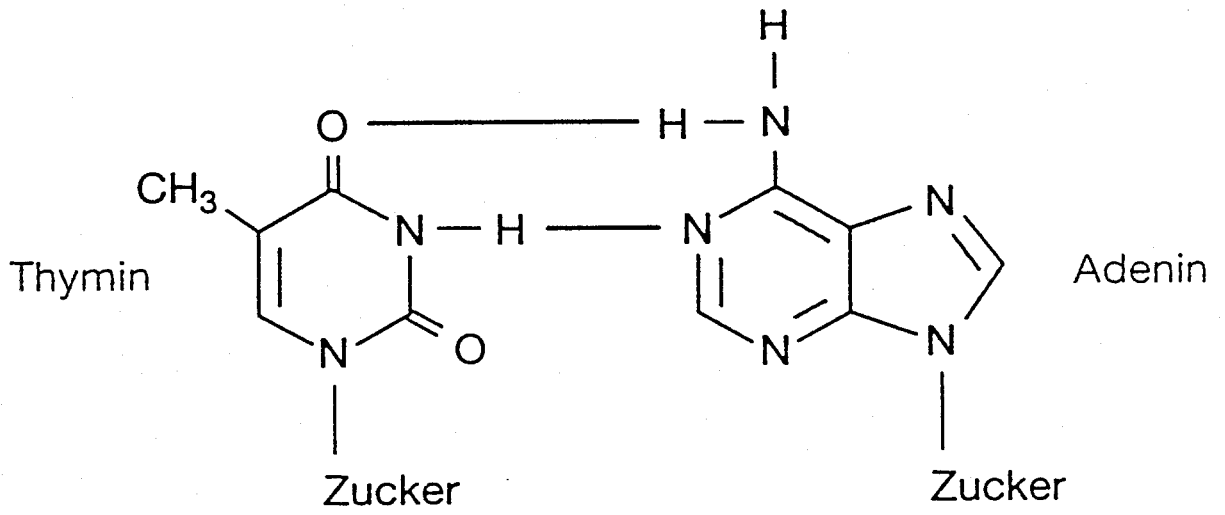


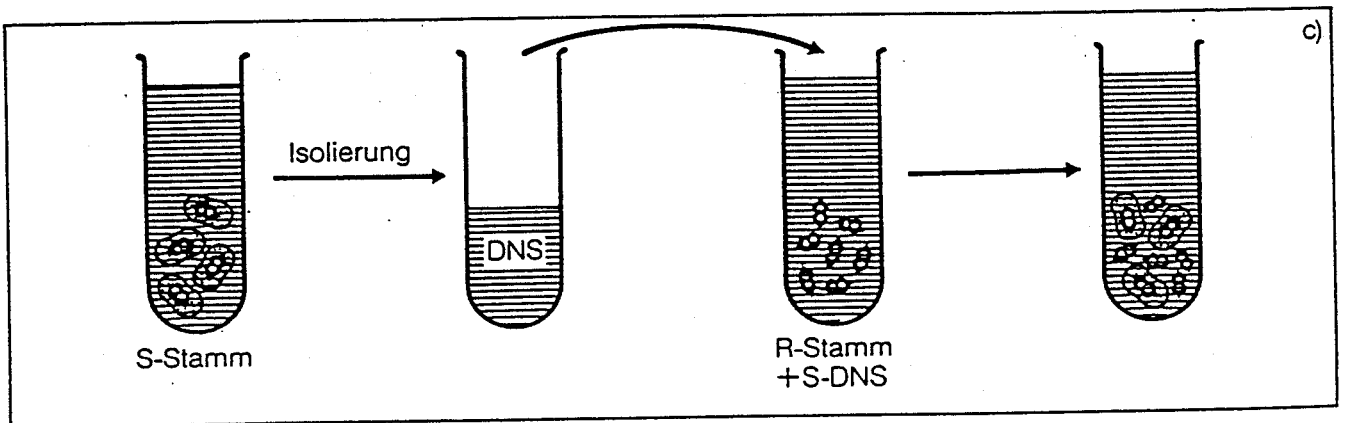
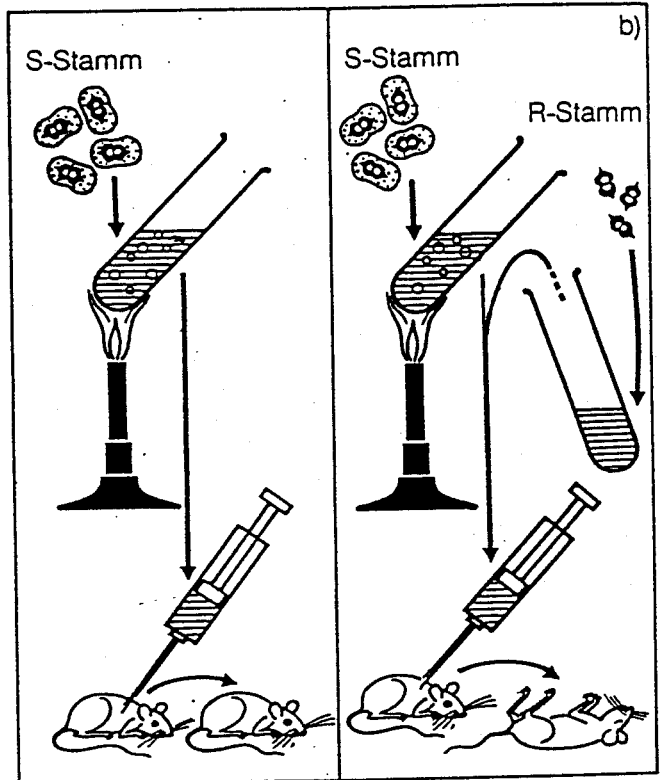
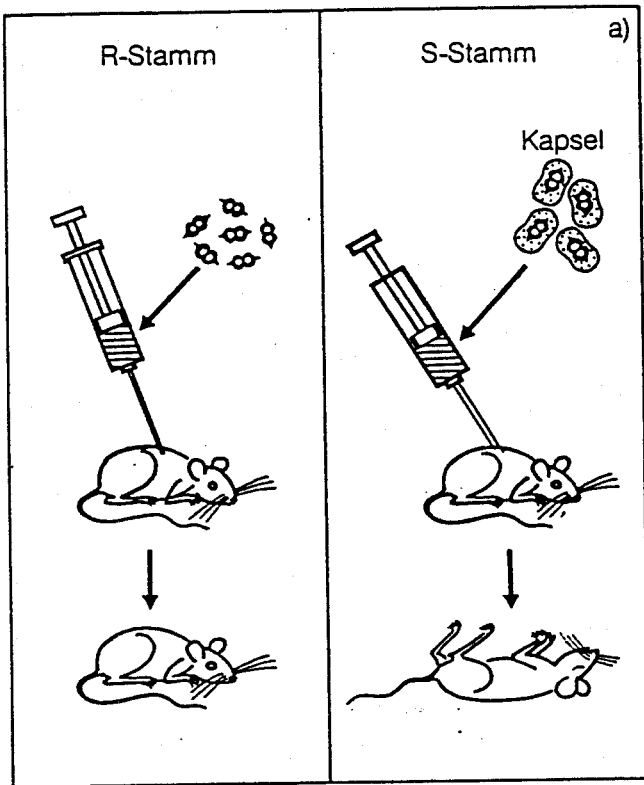
a



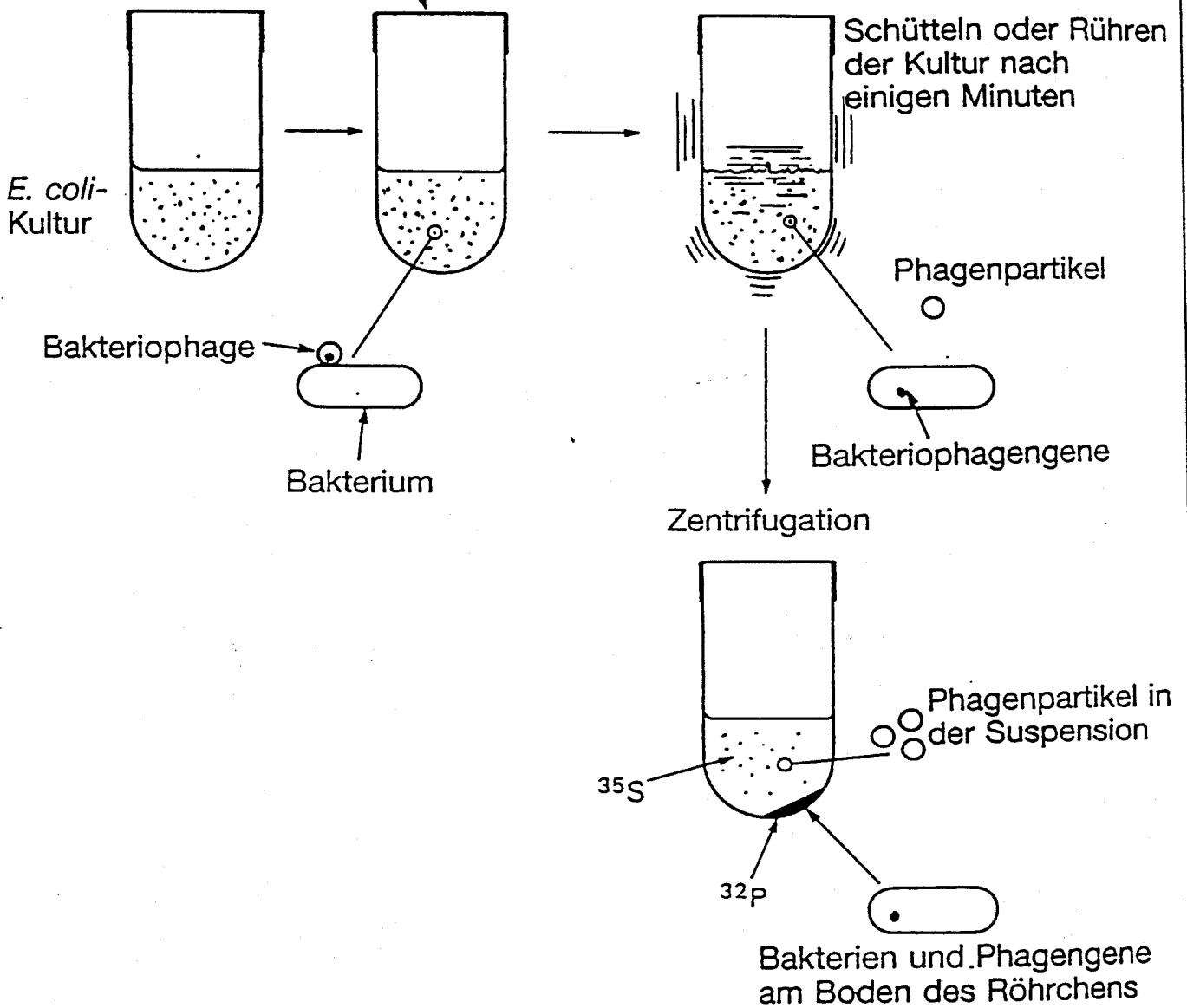
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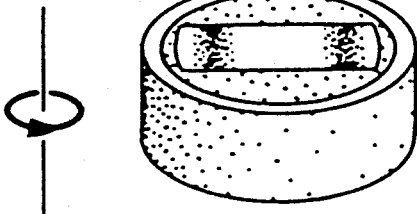




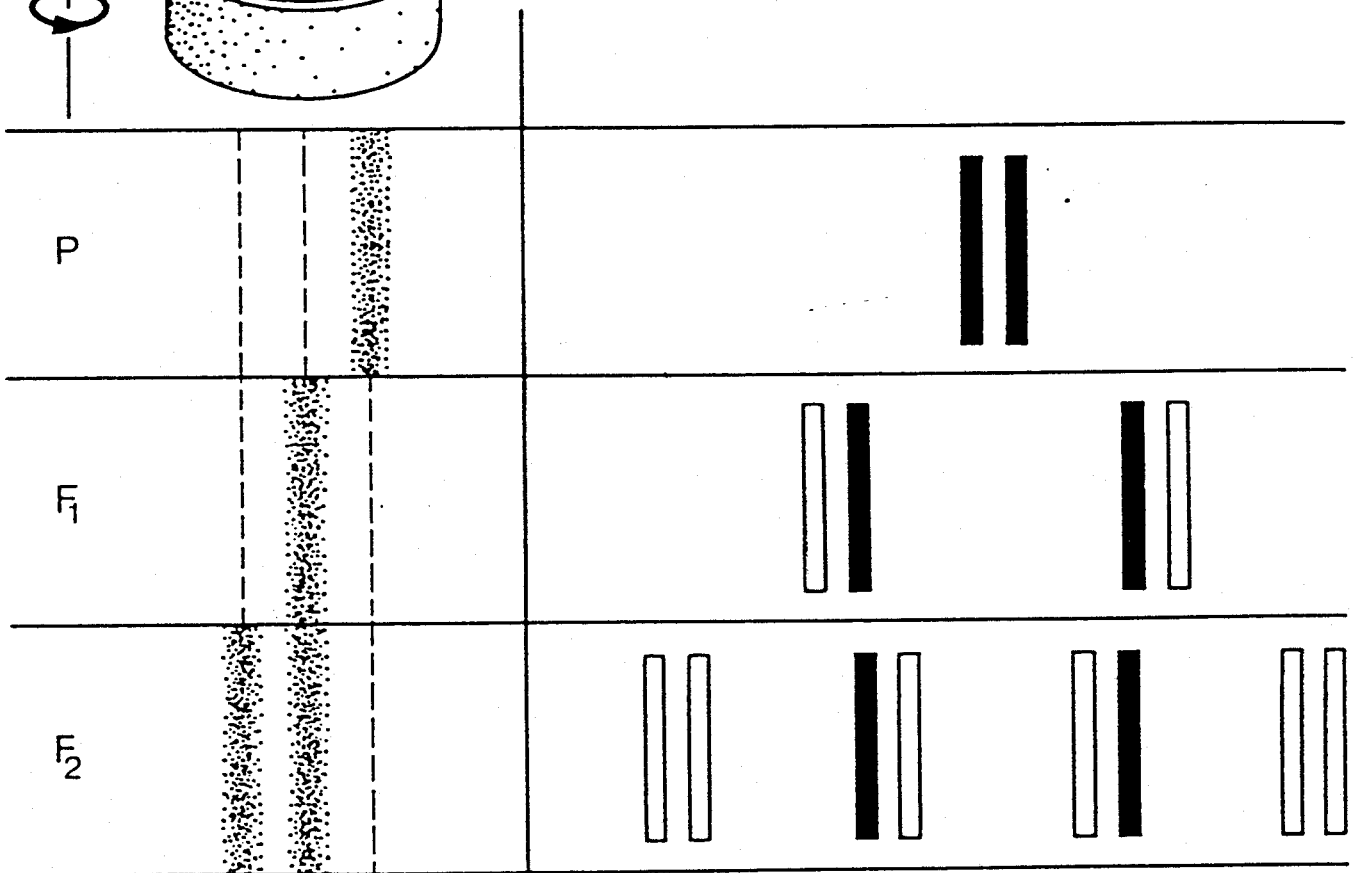
Zufügen von ^{32}P - und ^{35}S -markierten
Bakteriophagen



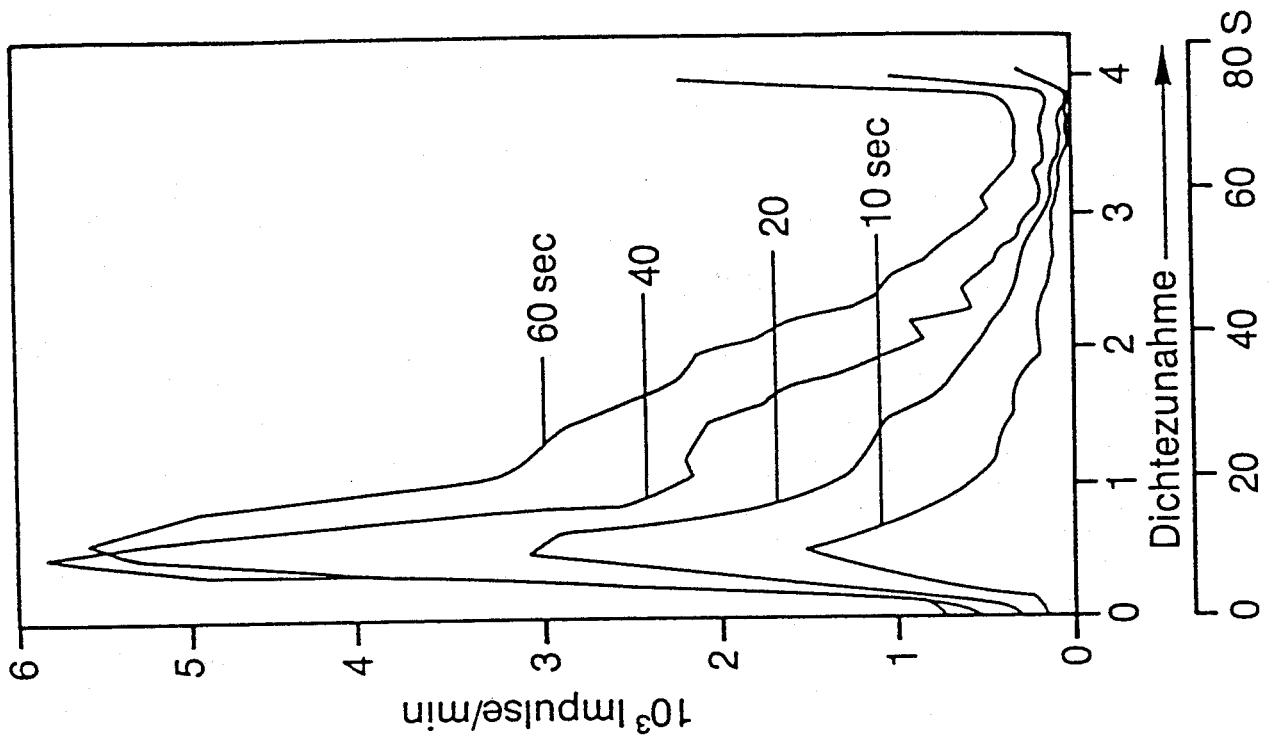
^{14}N - ^{15}N - DNA



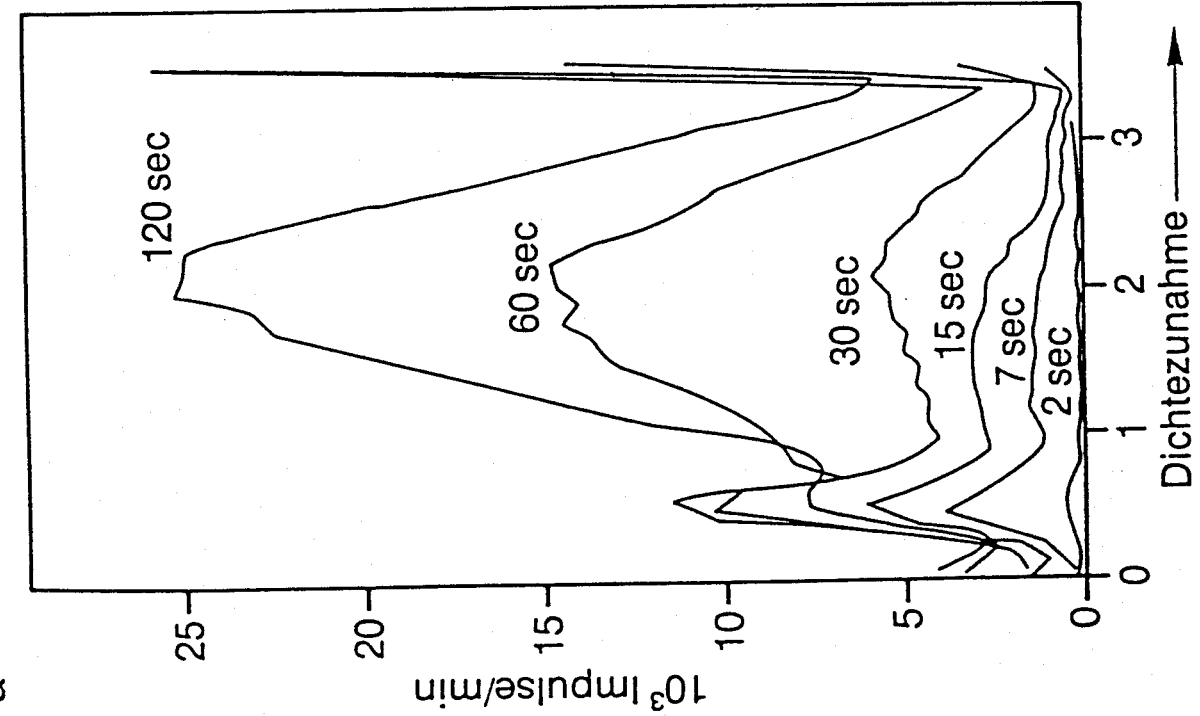
Mischung aus ^{14}N - und ^{15}N - DNA



b



a



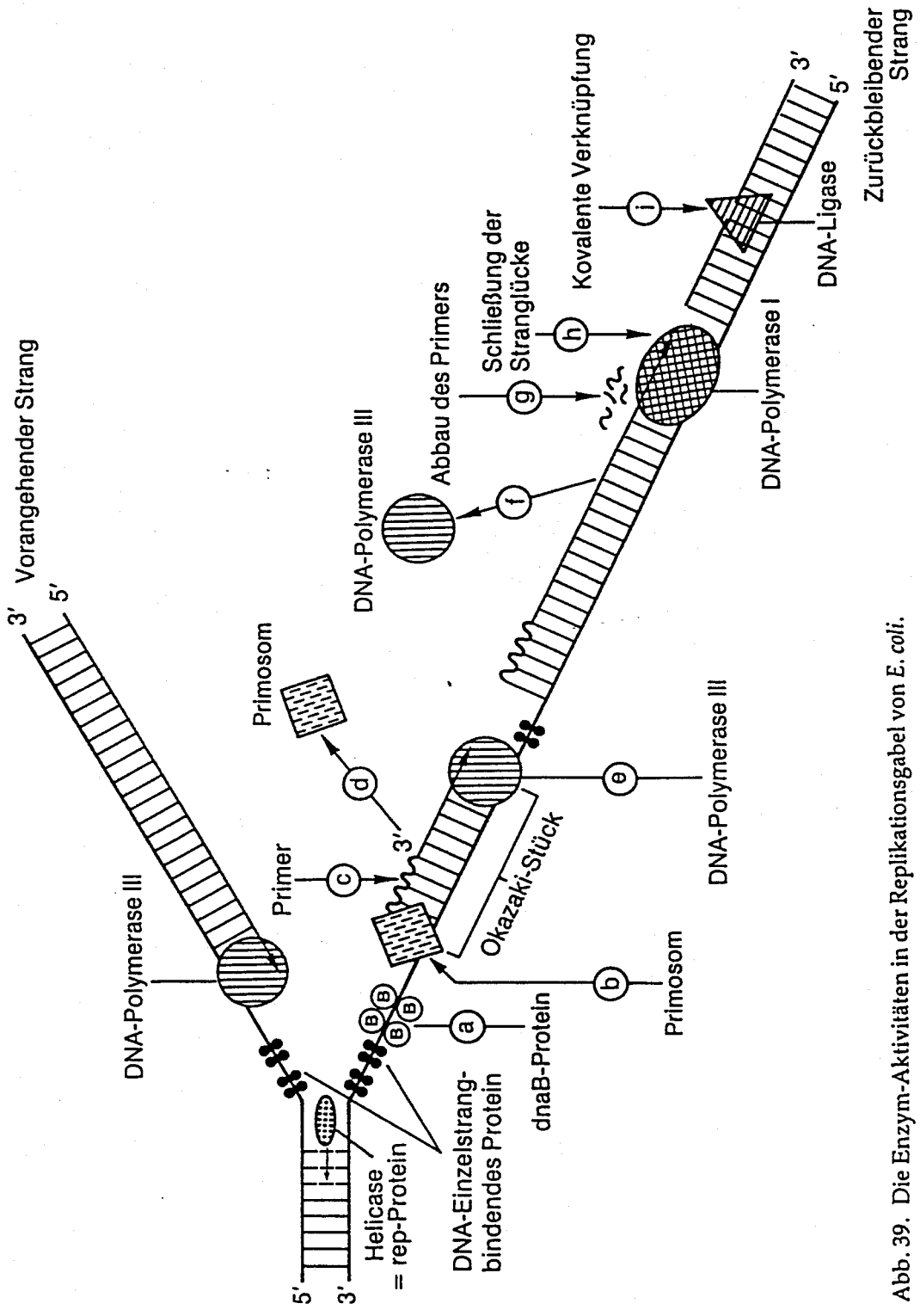
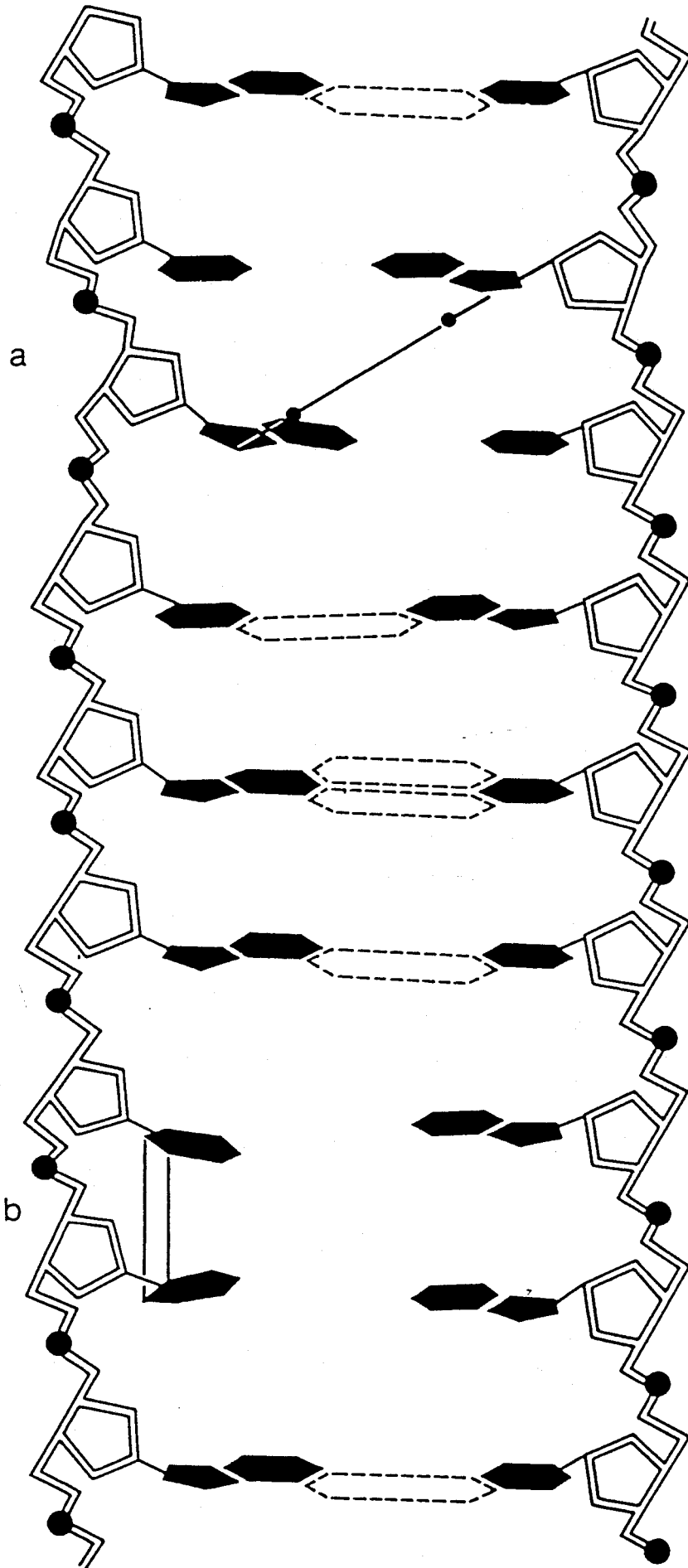
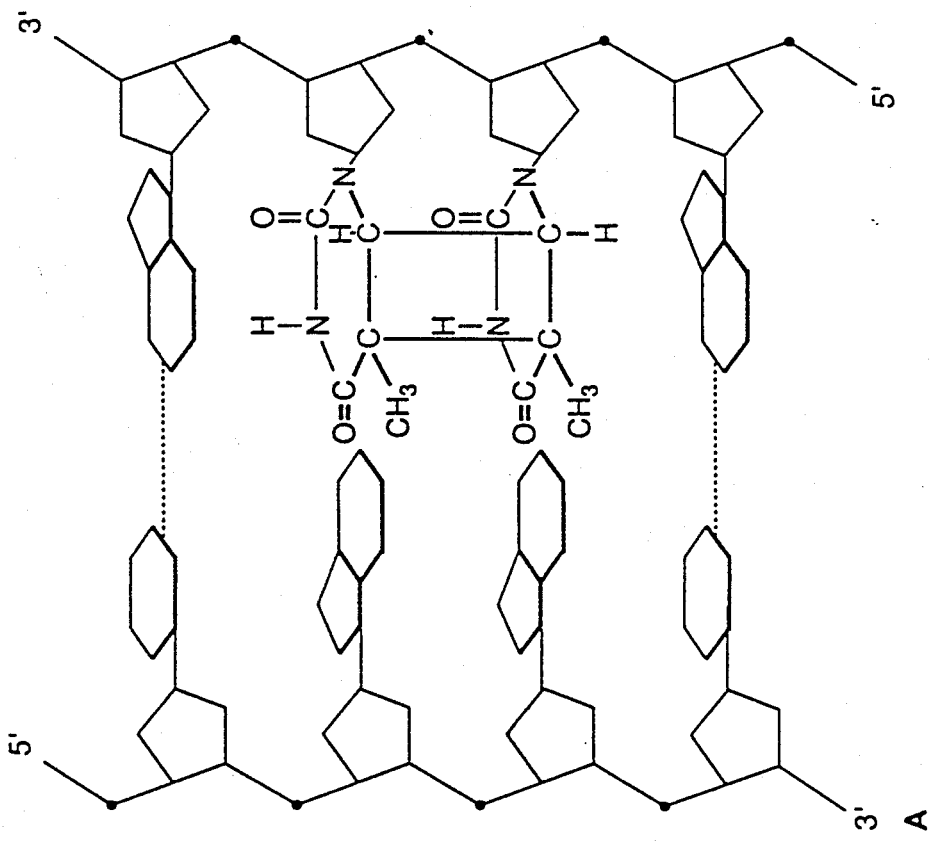
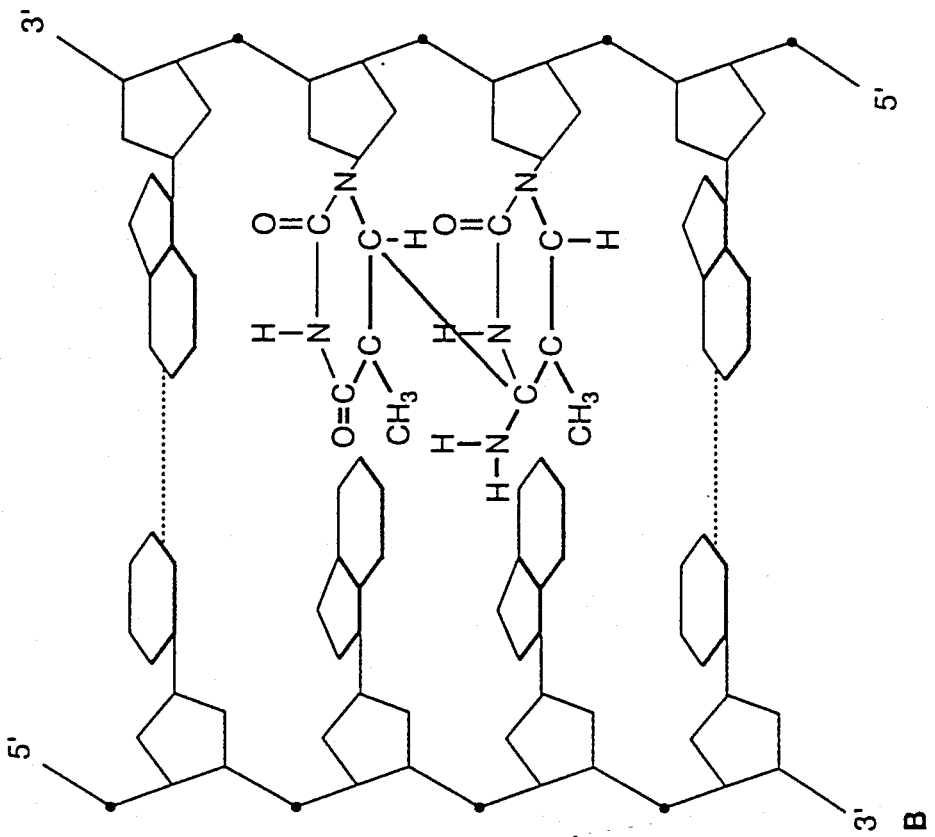
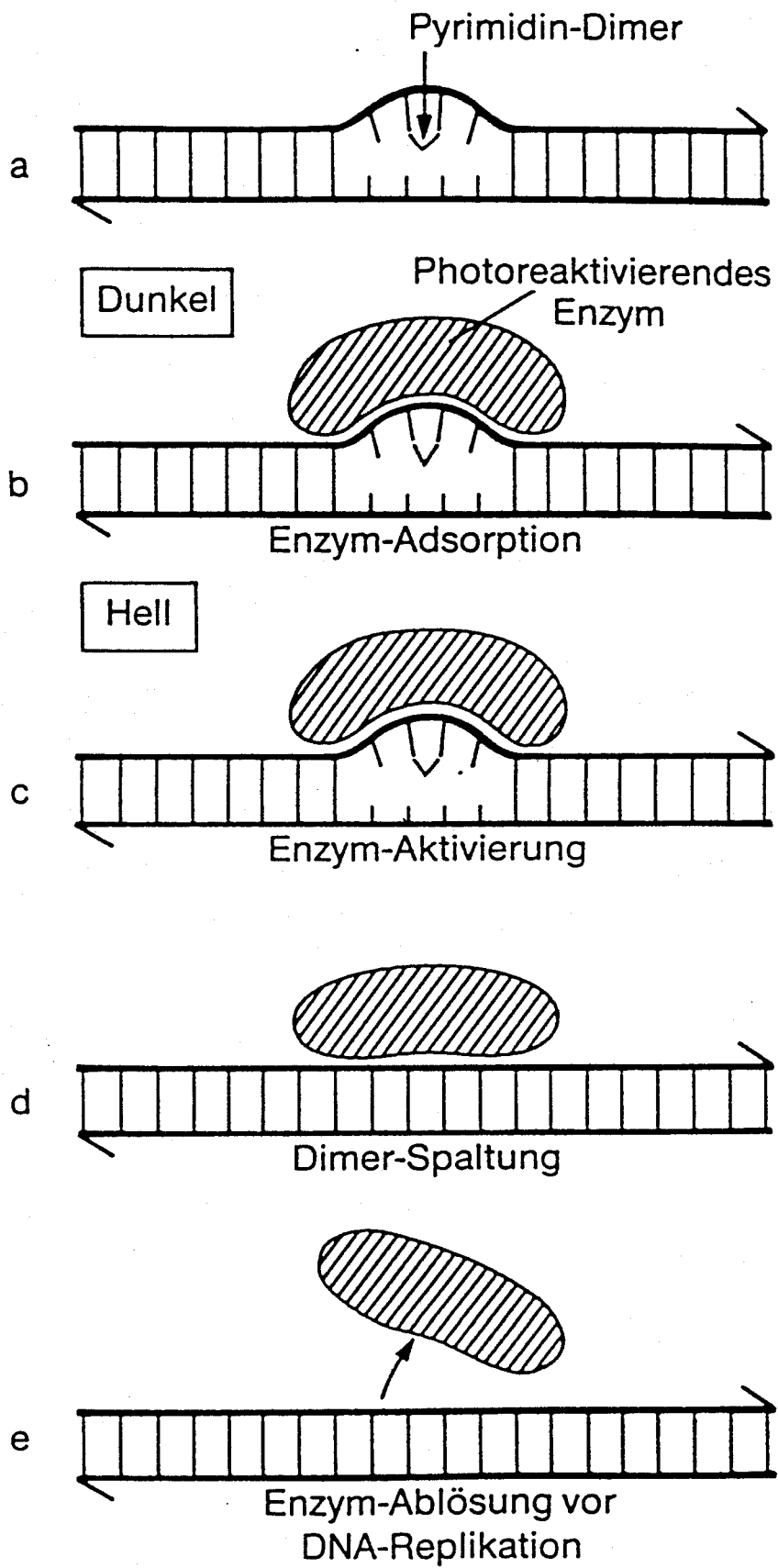
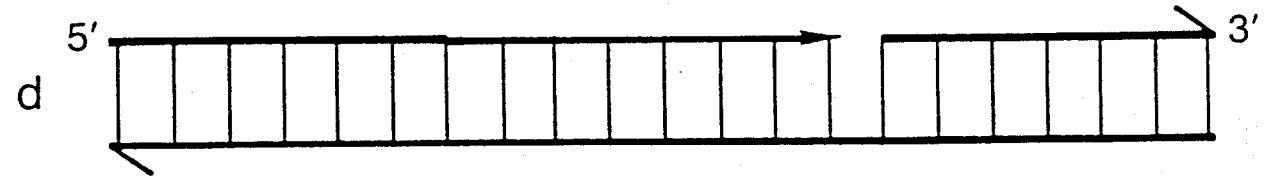
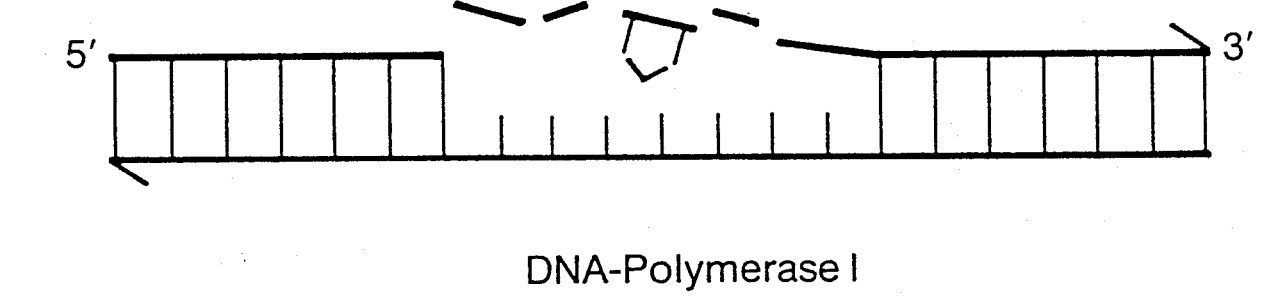
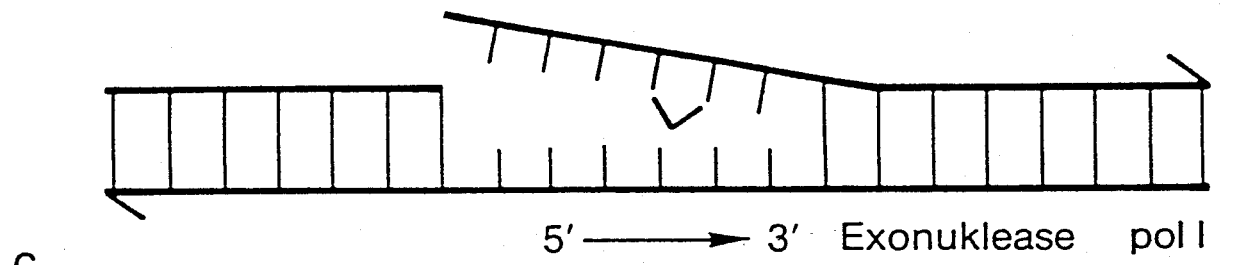
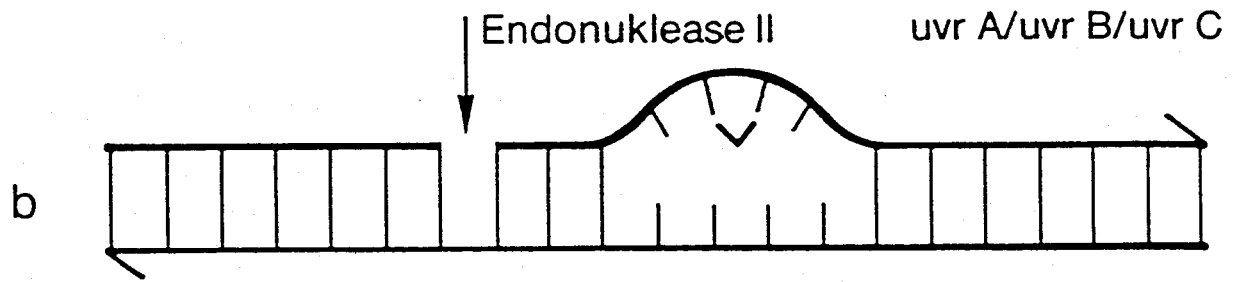
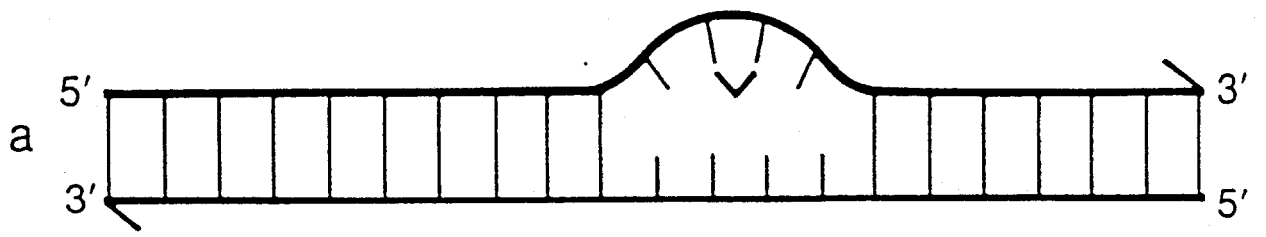


Abb. 39. Die Enzym-Aktivitäten in der Replikationsgabel von *E. coli*.

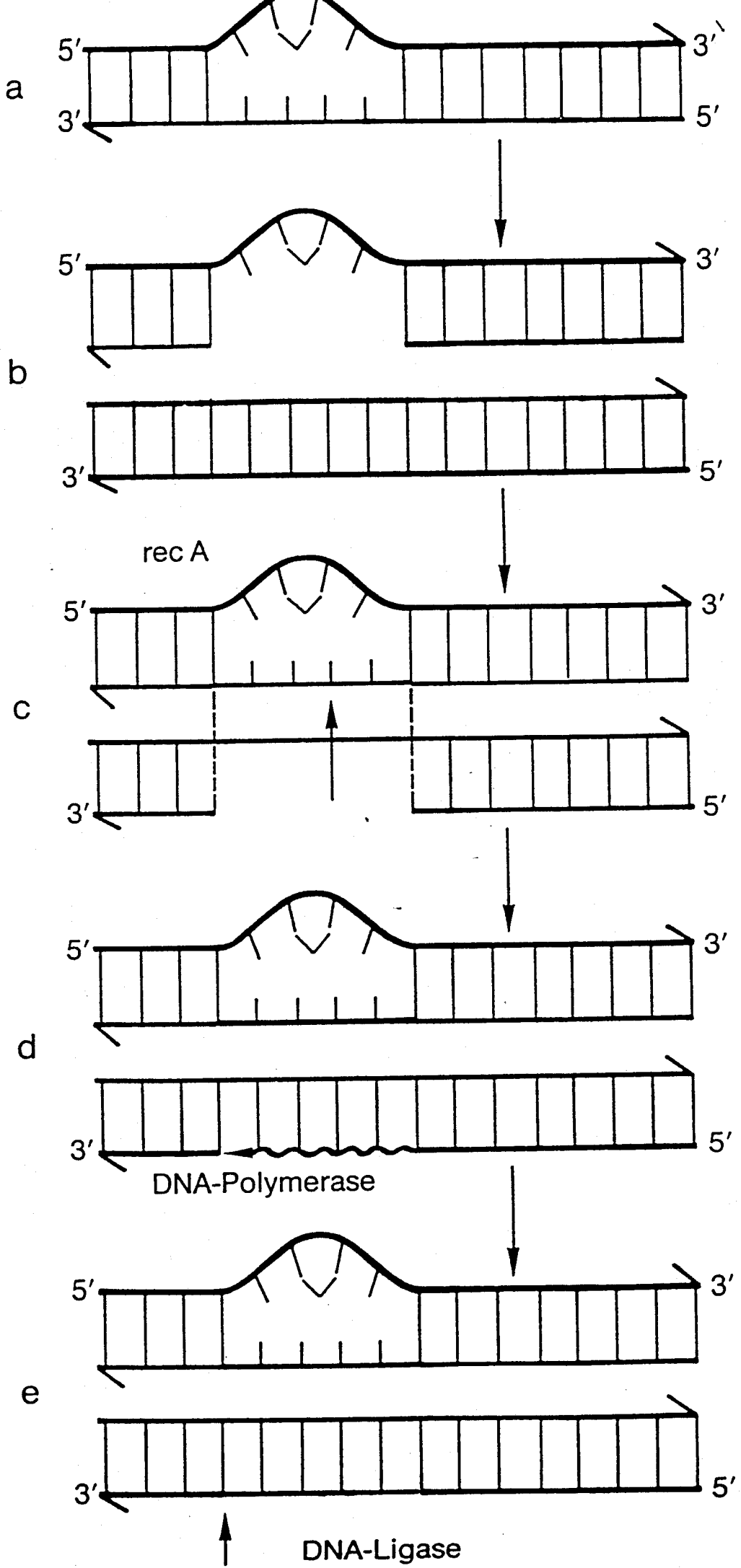


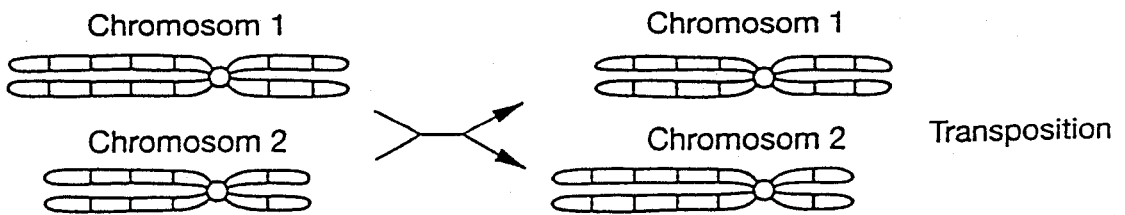
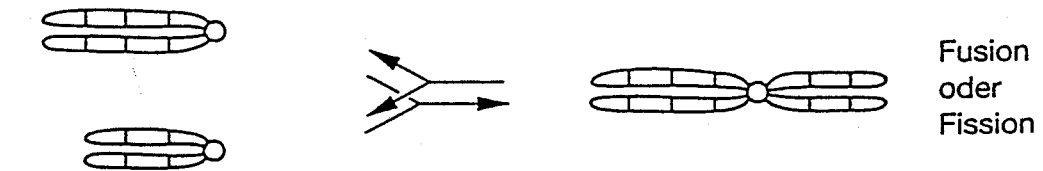
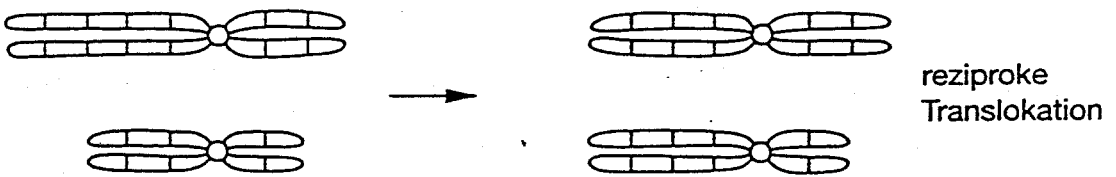
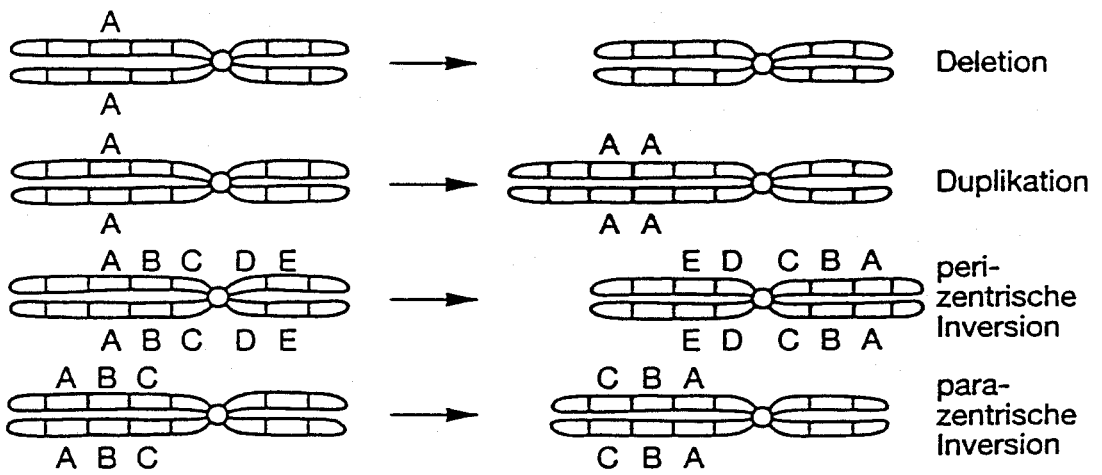


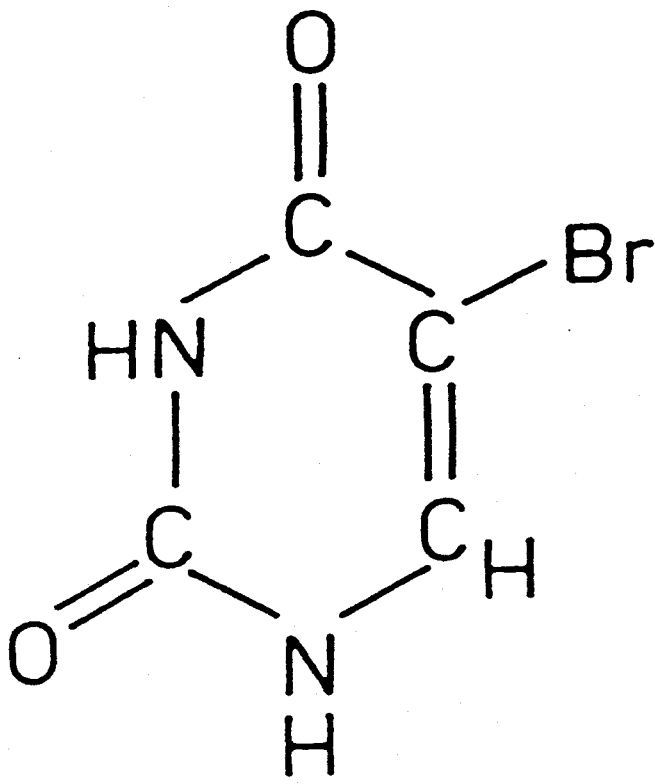




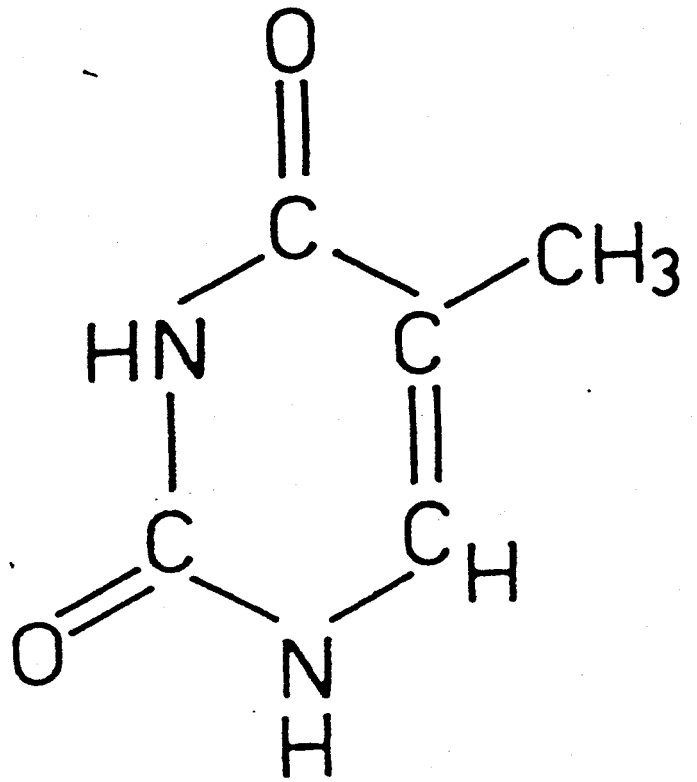
DNA-Replikation



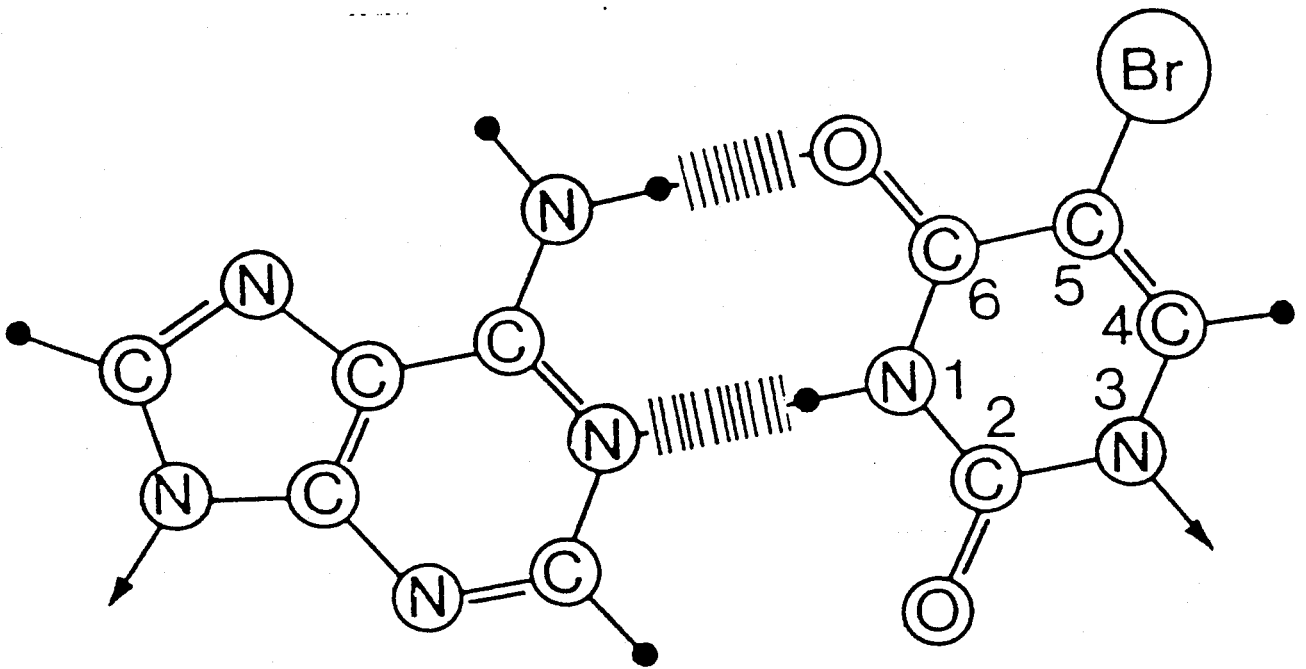




5'-Bromuracil

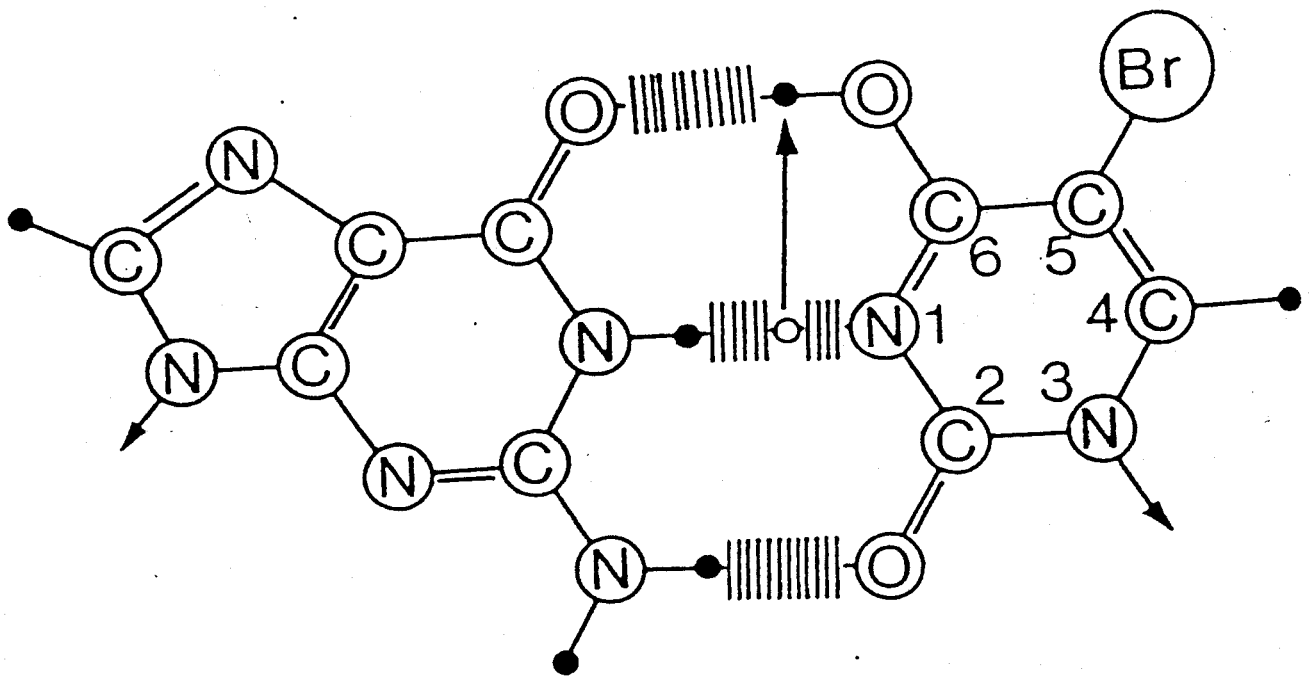


Thymin



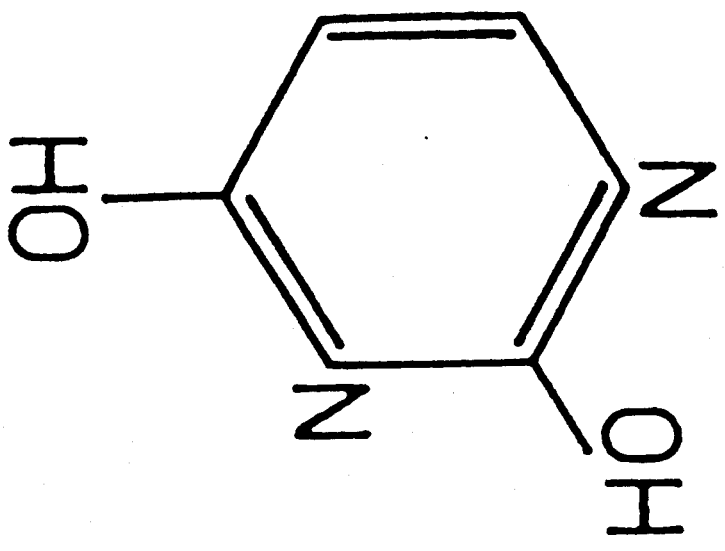
Adenin

5' - Bromuracil
(normaler Ketozustand)

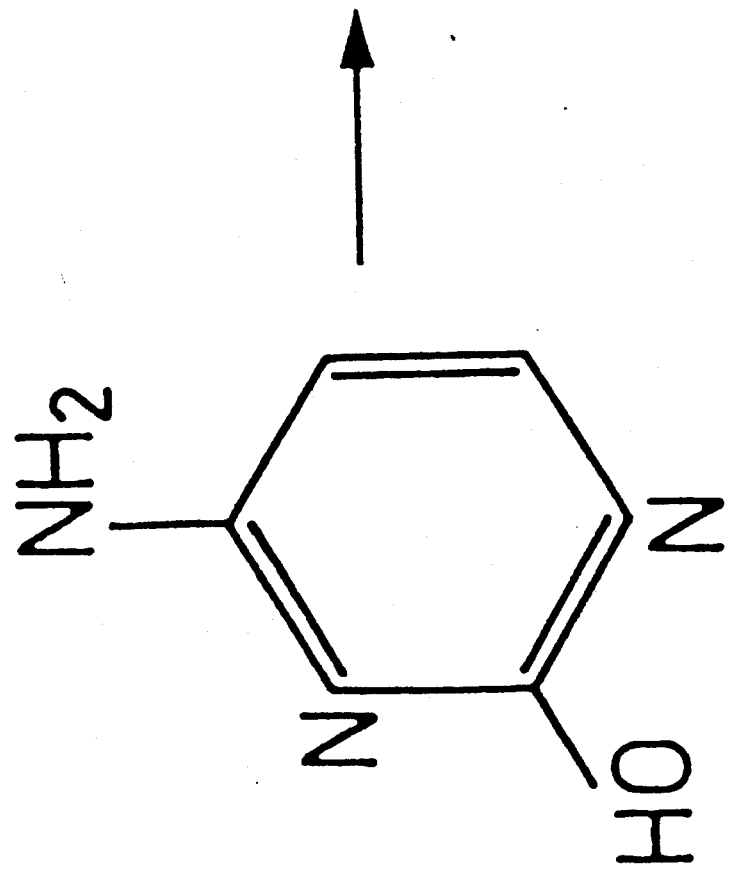


Guanin

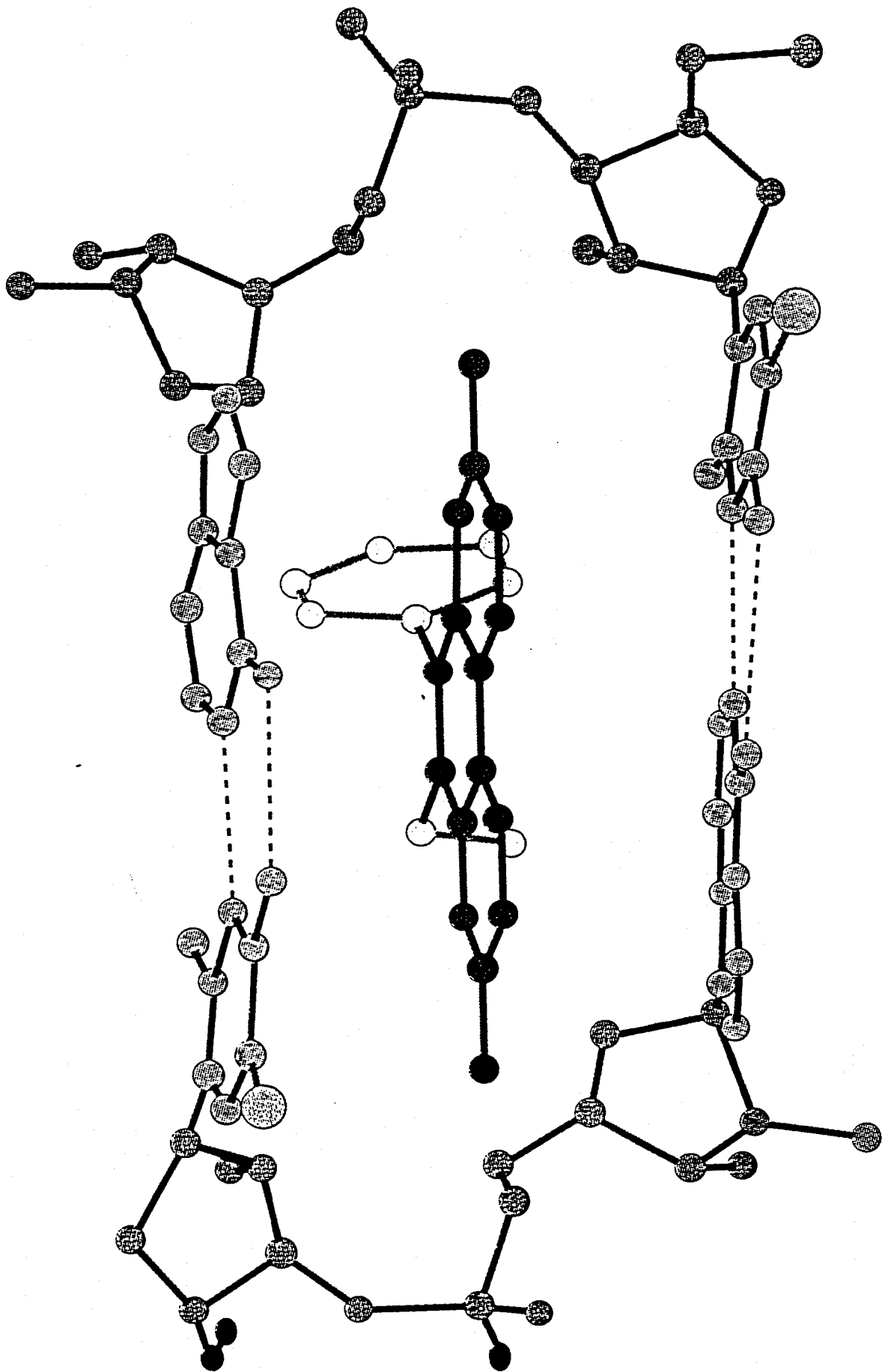
5'-Bromuracil
(seltener Enolzustand)

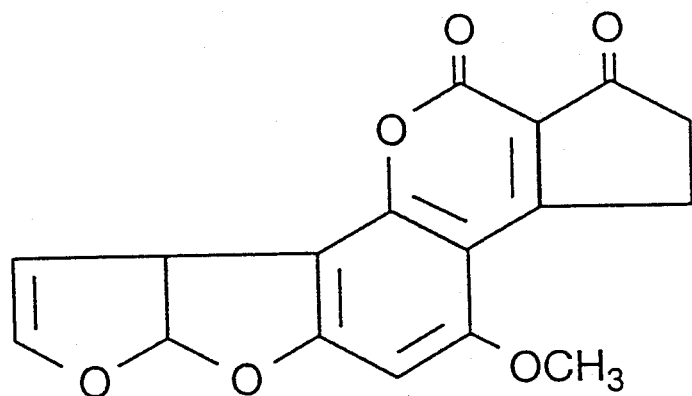


Uracil

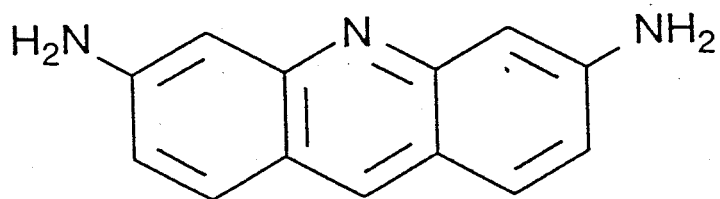


Cytosin

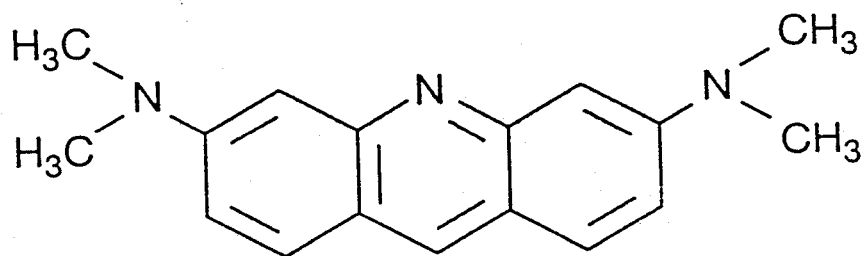




Aflatoxin B₁



Proflavin



Acridinorange