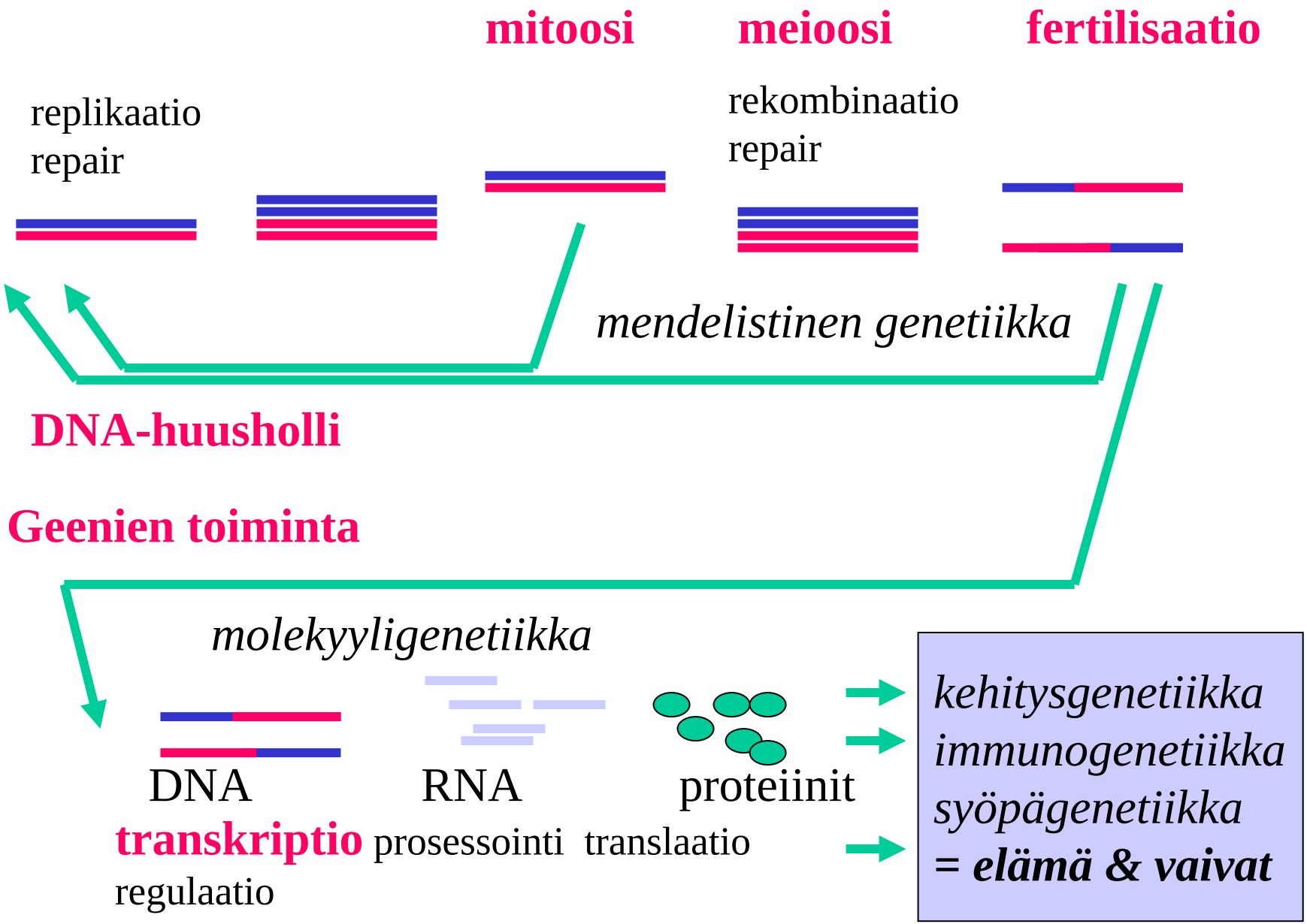
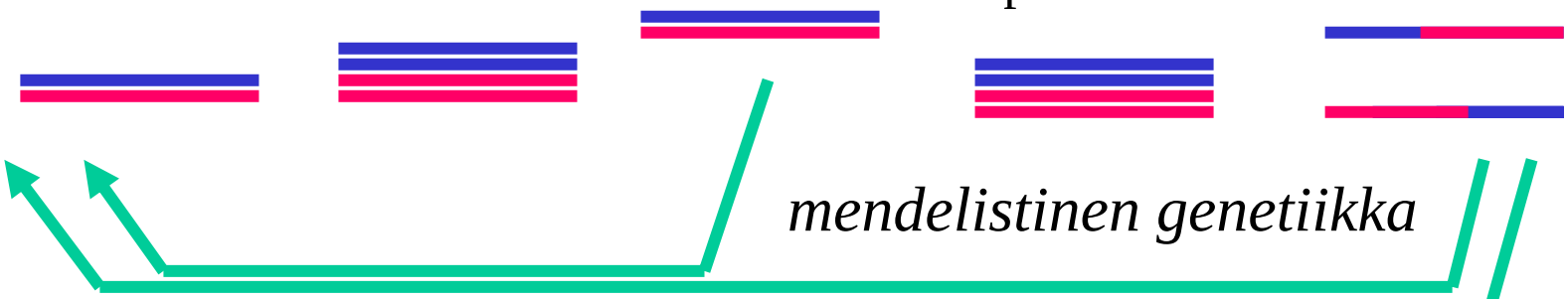

mitoosi
meioosi
fertilisaatio
replikaatio
repair
rekombinaatio
repair
mendelistinen genetiikka
DNA-huusholli
Geenien toiminta
molekyyligenetiikka
DNA
RNA
proteiinit
transkriptio
prosessointi
translaatio
regulaatio
kehitysgenetiikka
immunogenetiikka
syöpägenetiikka
= elämä & vaivat

CELL 309

mitoosi **meioosi** **fertilisaatio**

rekombinaatio
repair



mendelistinen genetiikka

DNA-huusholli

Geenien toiminta

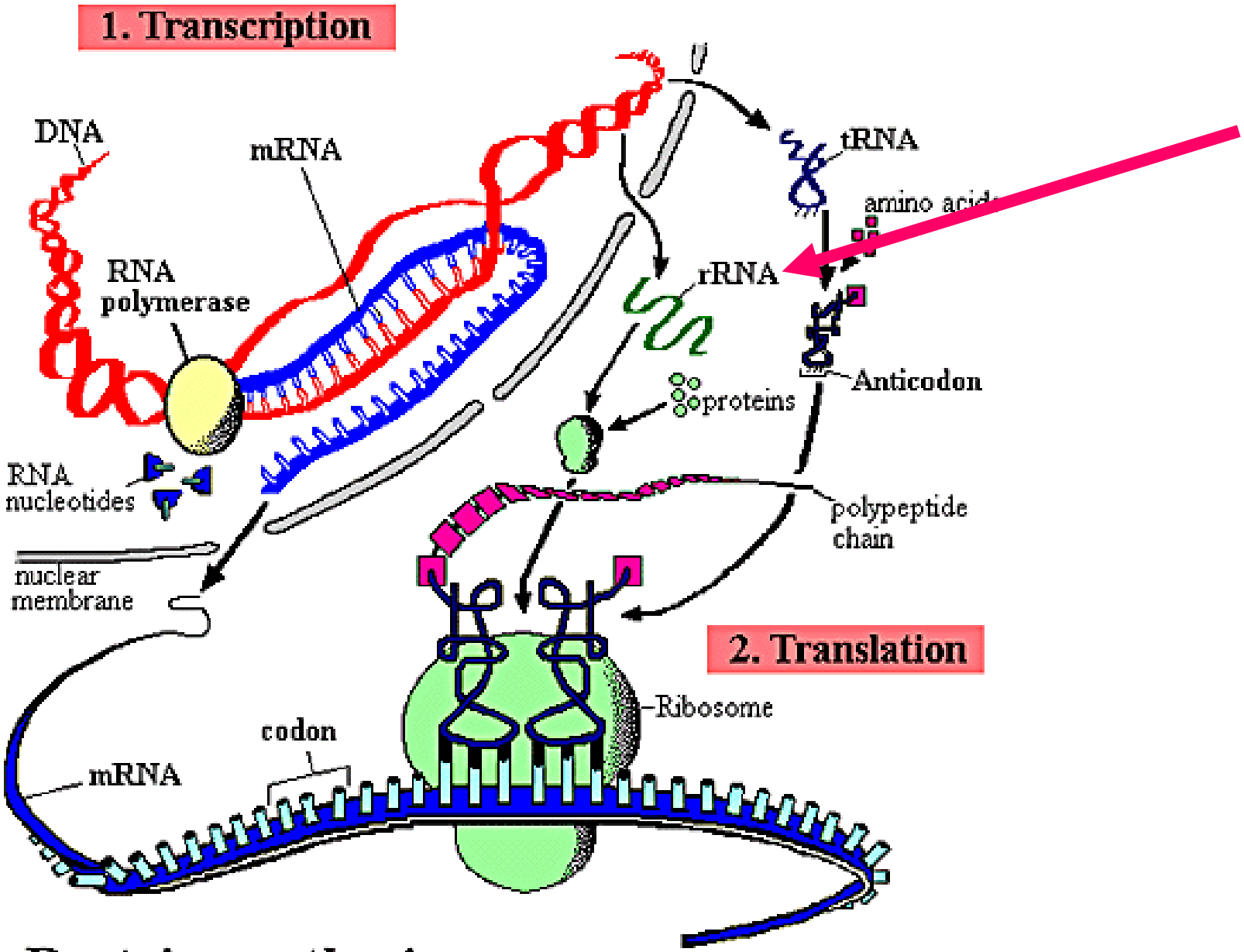
molekyyligenetiikka

kehitysgenetiikka
immunogenetiikka
syöpägenetiikka
ELÄMÄ!

Genetiikan perusteiden miellekartta: transkriptio jatkuu

Ribosomin RNA eli rRNA

- Koodia sanotaan rDNA:ksi
- Ei mitään tekemistä triplettikoodin kanssa
- Useita alayksiköitä
- Eräs kaikkein konservatiivisimmista informaatiopaketeista
- Todistaa vahvasti *kaikkien* eliöiden yhteisestä alkuperästä



Protein synthesis

Table 6–1 Principal Types of RNAs Produced in Cells

TYPE OF RNA	FUNCTION
mRNAs	messenger RNAs, code for proteins
rRNAs	ribosomal RNAs, form the basic structure of the ribosome and catalyze protein synthesis
tRNAs	transfer RNAs, central to protein synthesis as adaptors between mRNA and amino acids
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snoRNAs	small nucleolar RNAs, used to process and chemically modify rRNAs
scaRNAs	small cajal RNAs, used to modify snoRNAs and snRNAs
miRNAs	microRNAs, regulate gene expression typically by blocking translation of selective mRNAs
siRNAs	small interfering RNAs, turn off gene expression by directing degradation of selective mRNAs and the establishment of compact chromatin structures
Other noncoding RNAs	function in diverse cell processes, including telomere synthesis, X-chromosome inactivation, and the transport of proteins into the ER

Table 6-1 Molecular Biology of the Cell 5/e (© Garland Science 2008)

CELL 336

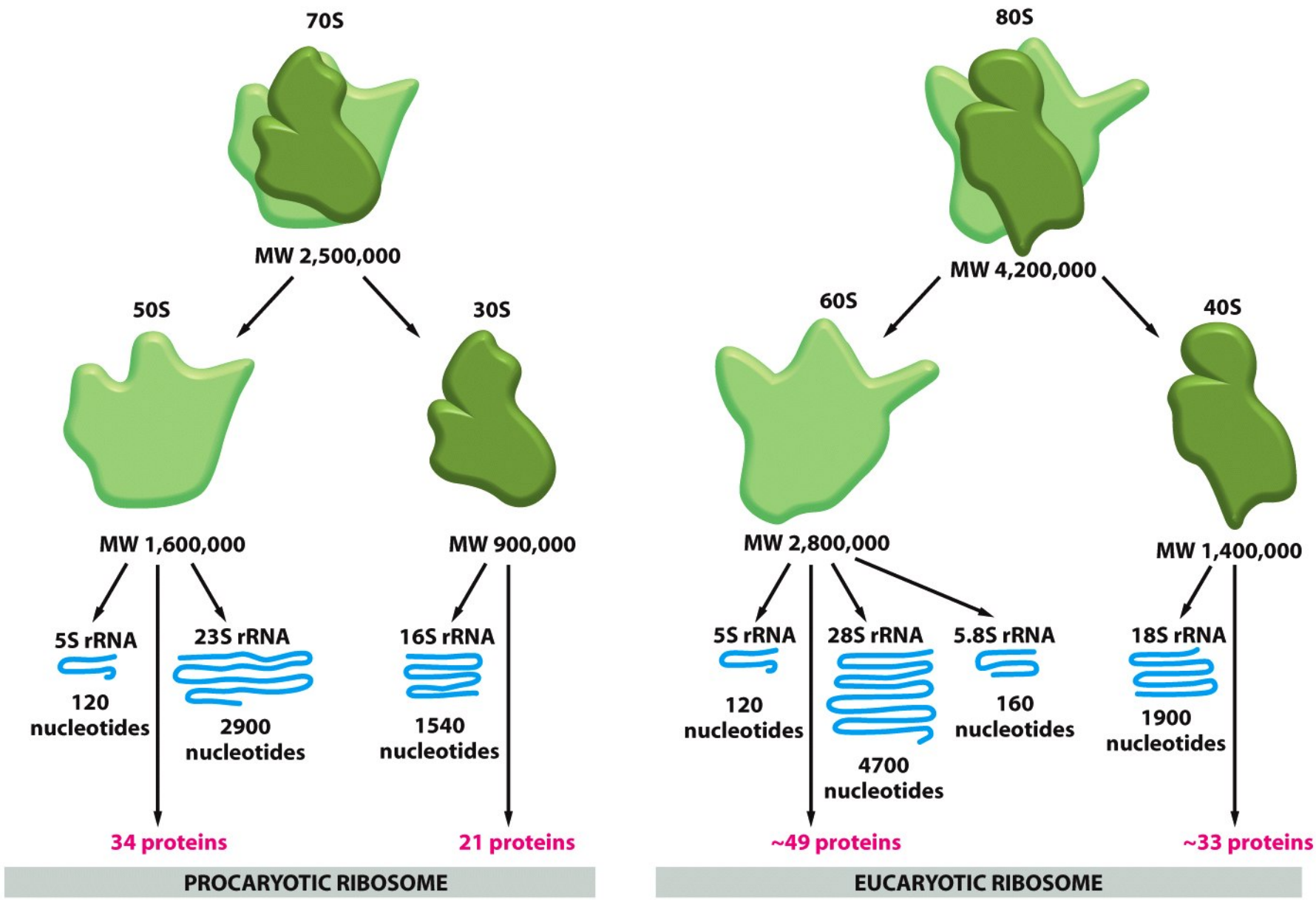

Figure 6-63 Molecular Biology of the Cell 5/e (© Garland Science 2008)

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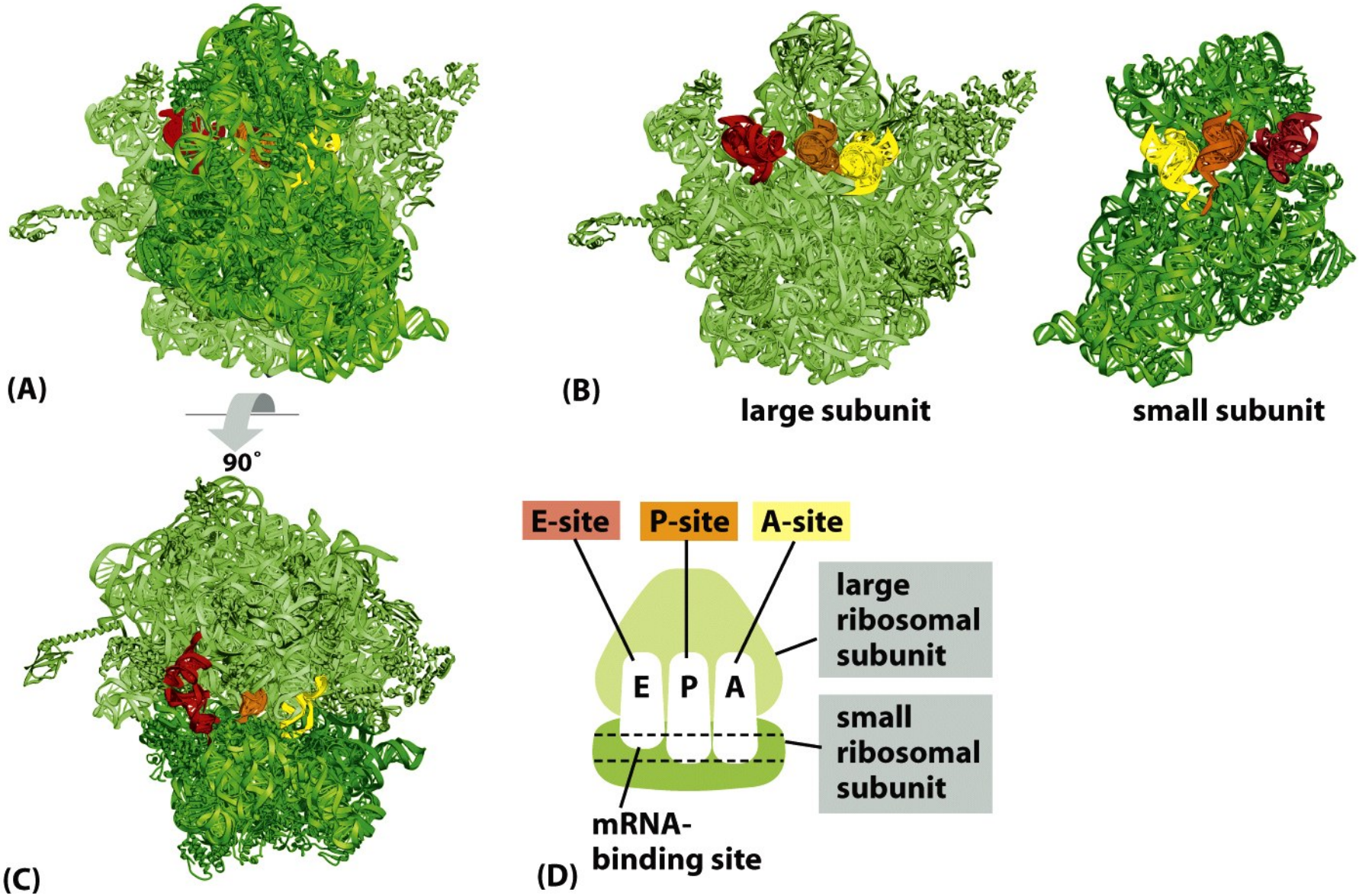
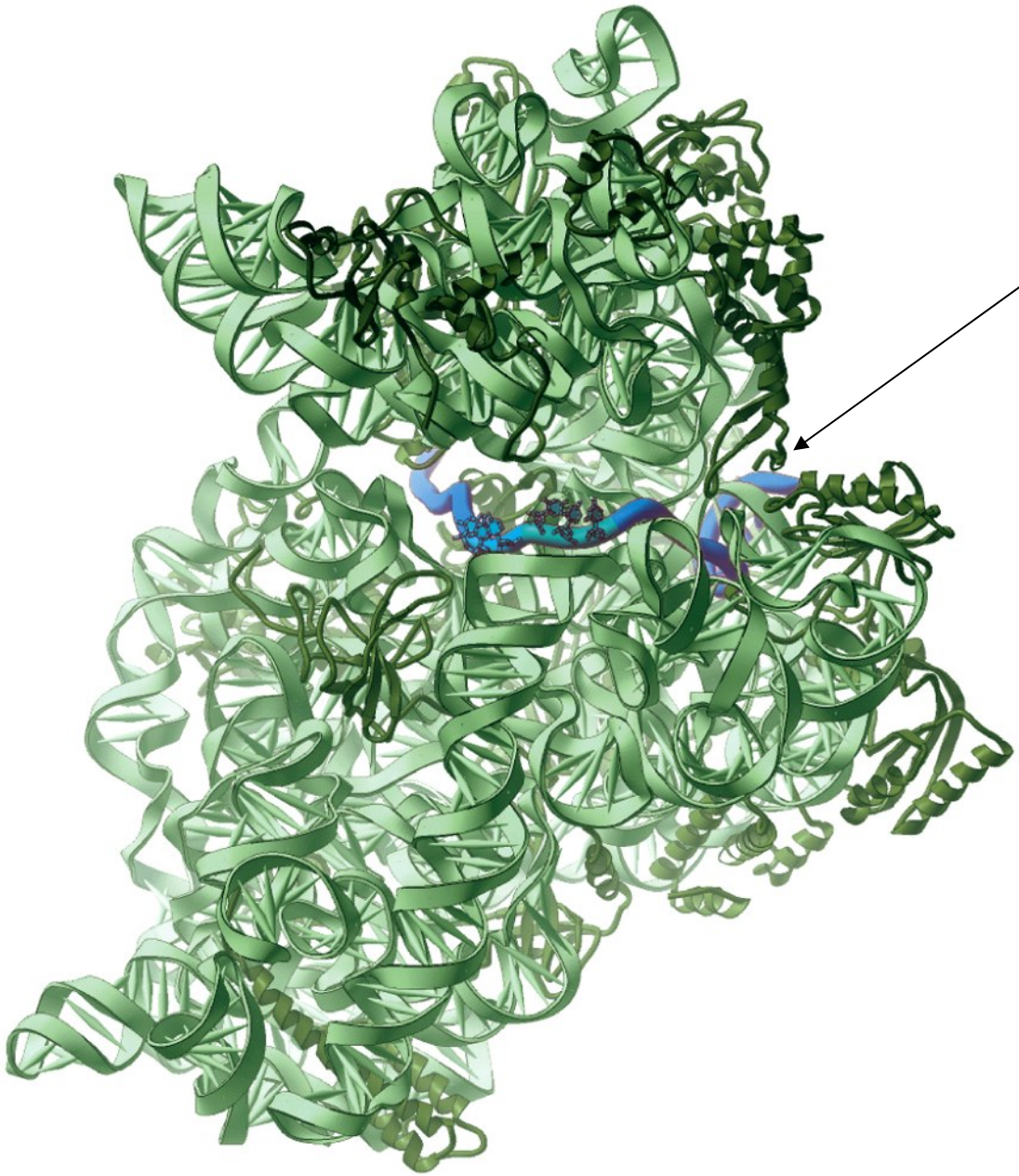


Figure 6-64 Molecular Biology of the Cell 5/e (© Garland Science 2008)

CELL 375



Small subunit

mRNA luikertaa

Figure 6-65 Molecular Biology of the Cell 5/e (© Garland Science 2008)

CELL 376

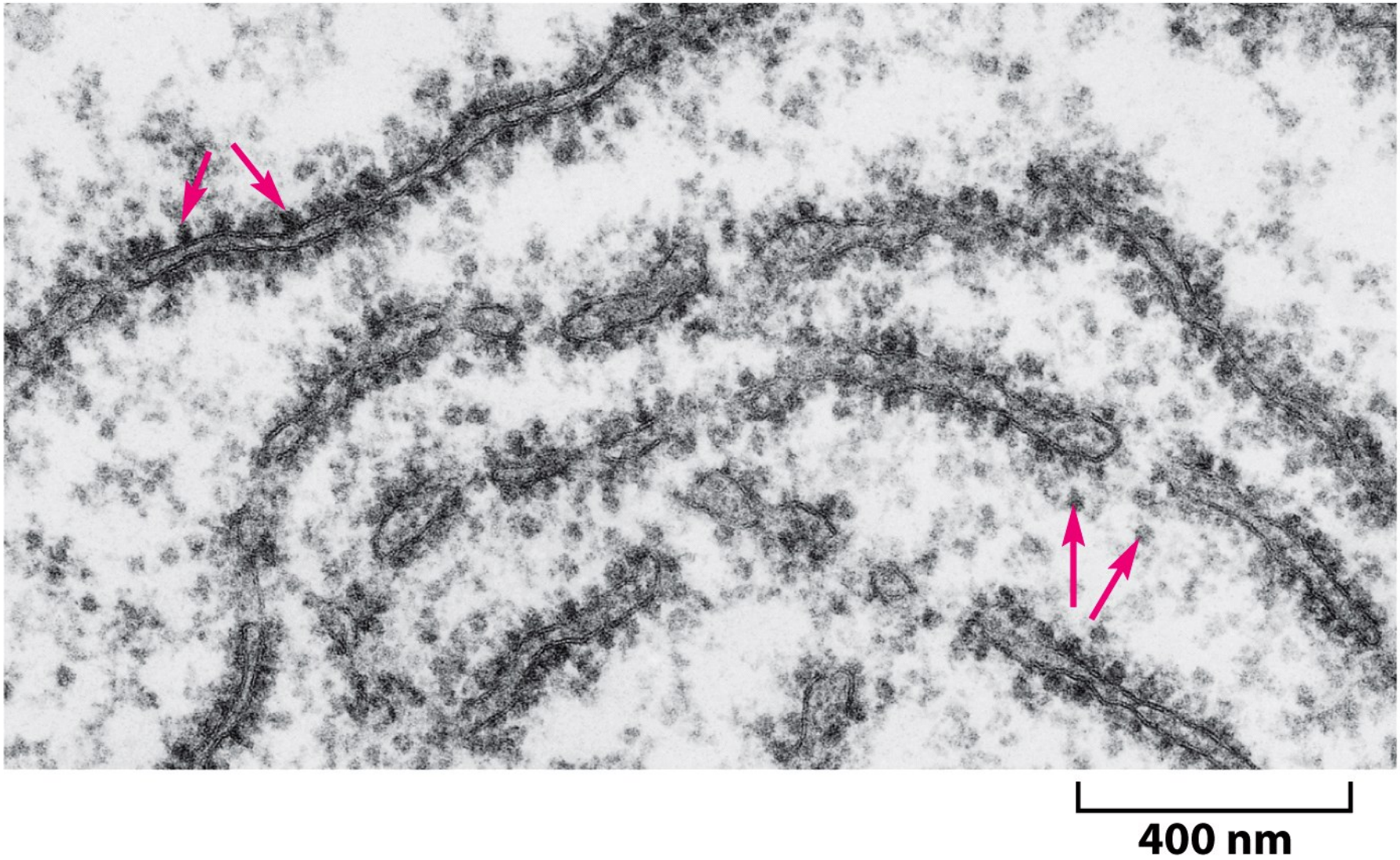


Figure 6-62 Molecular Biology of the Cell 5/e (© Garland Science 2008)

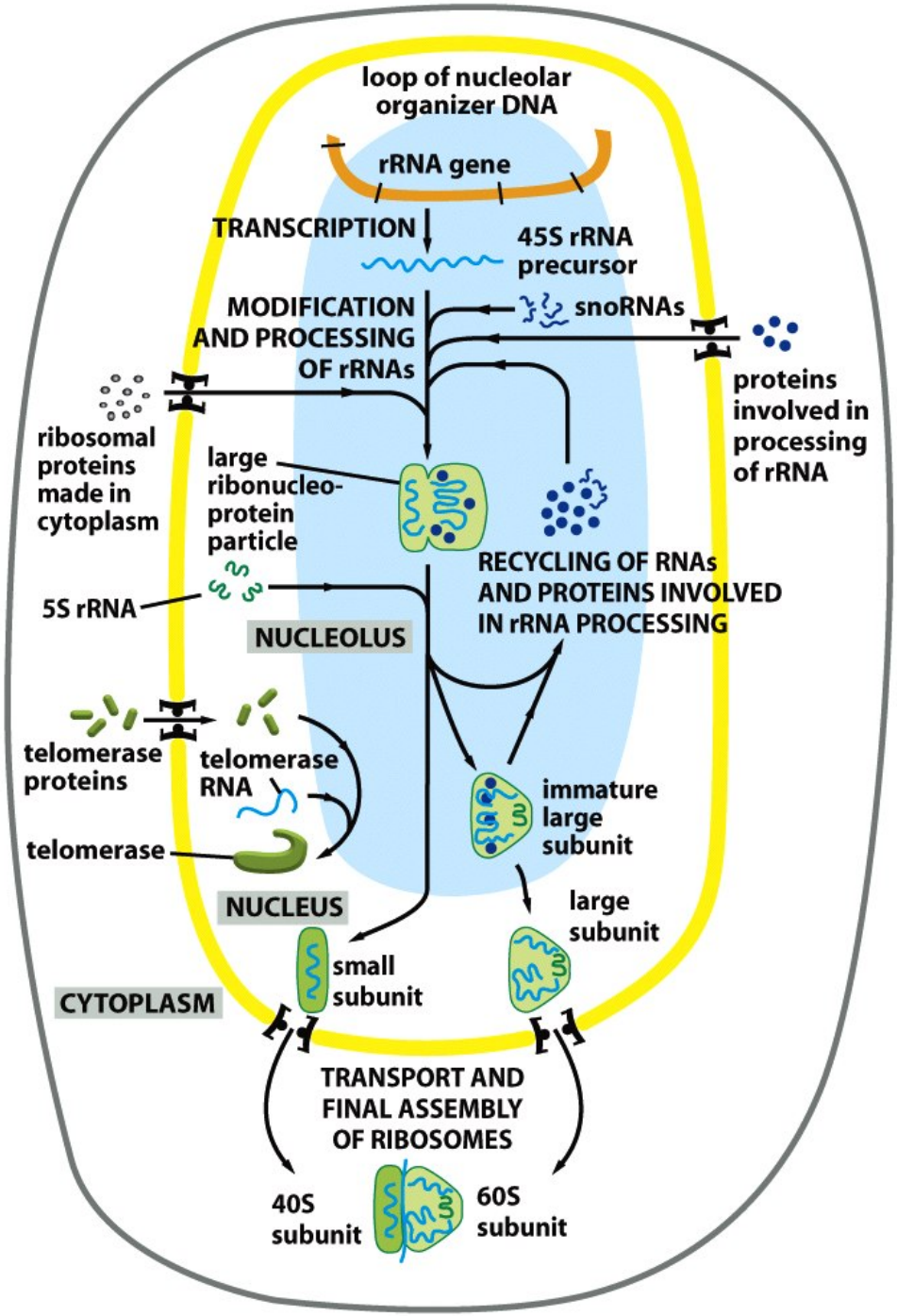


Figure 6–47 The function of the nucleolus in ribosome and other ribonucleoprotein synthesis. The 45S precursor rRNA is packaged in a large ribonucleoprotein particle containing many ribosomal proteins imported from the cytoplasm. While this particle remains in the nucleolus, selected pieces are added and others discarded as it is processed into immature large and small ribosomal subunits. The two ribosomal subunits are thought to attain their final functional form only as each is individually transported through the nuclear pores into the cytoplasm. Other ribonucleoprotein complexes, including telomerase shown here, are also assembled in the nucleolus.

CELL 364

Figure 6-47 Molecular Biology of the Cell 5/e (© Garland Science 2008)

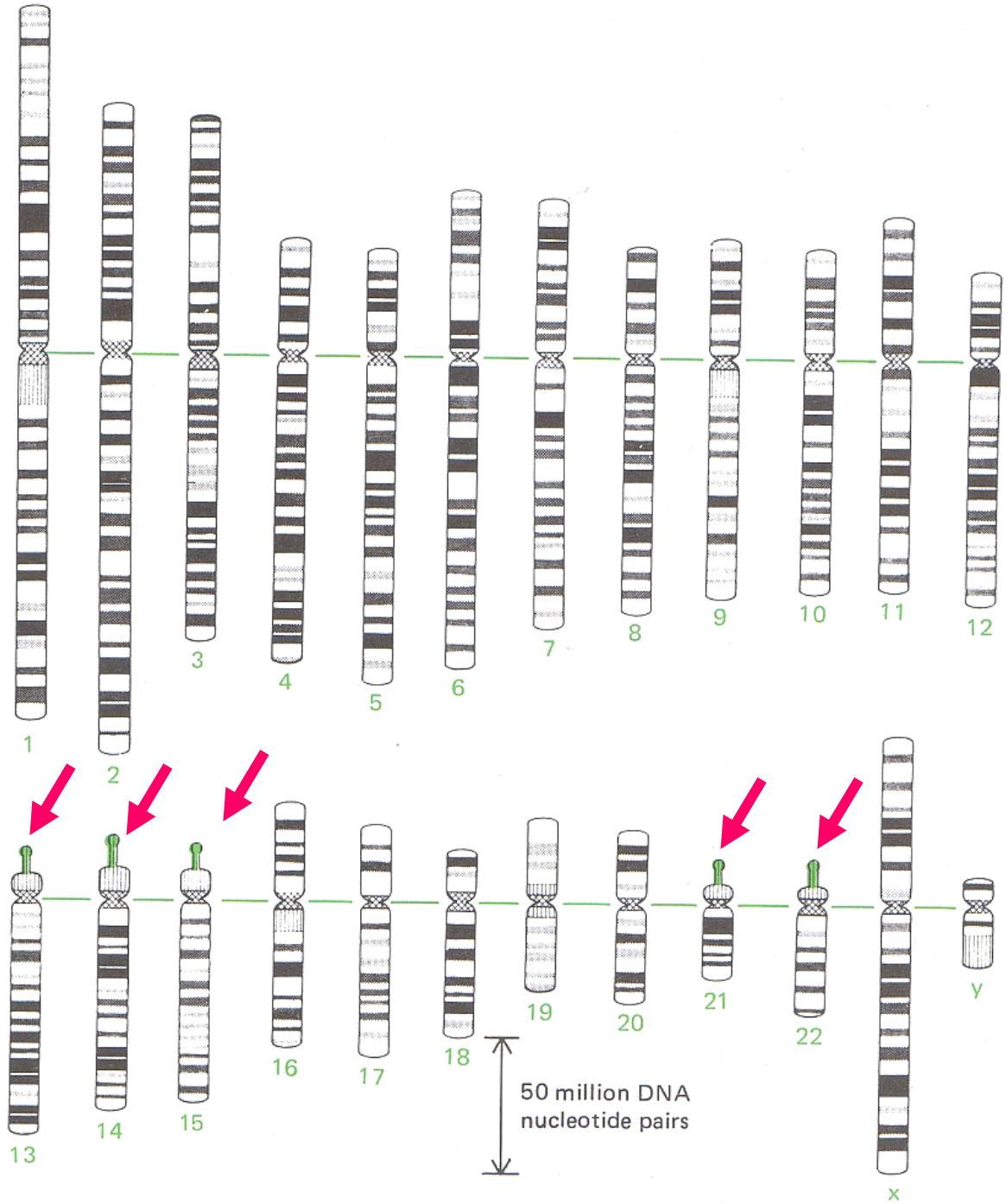


Figure 8–32 A standard map of the banding pattern of each chromosome in the human karyotype. This map was determined at the prometaphase stage of mitosis. Chromosomes 1 through 22 are labeled in the approximate order of their size; a diploid cell contains two of each of these autosomes plus two sex chromosomes—two X chromosomes (female) or an X and a Y chromosome (male). The 850 bands shown here are G bands, which stain with reagents that appear to be specific for A-T-rich DNA sequences. The *red knobs* on chromosomes 13, 14, 15, 21, and 22 indicate the positions of the genes that encode the large ribosomal RNAs; the *red lines* mark the centromere on each chromosome. (Adapted from U. Franke, *Cytogenet. Cell Genet.* 31:24–32, 1981.)

CELL 203

Fragile X :
Musco et al Cell85:

rDNA-geenit

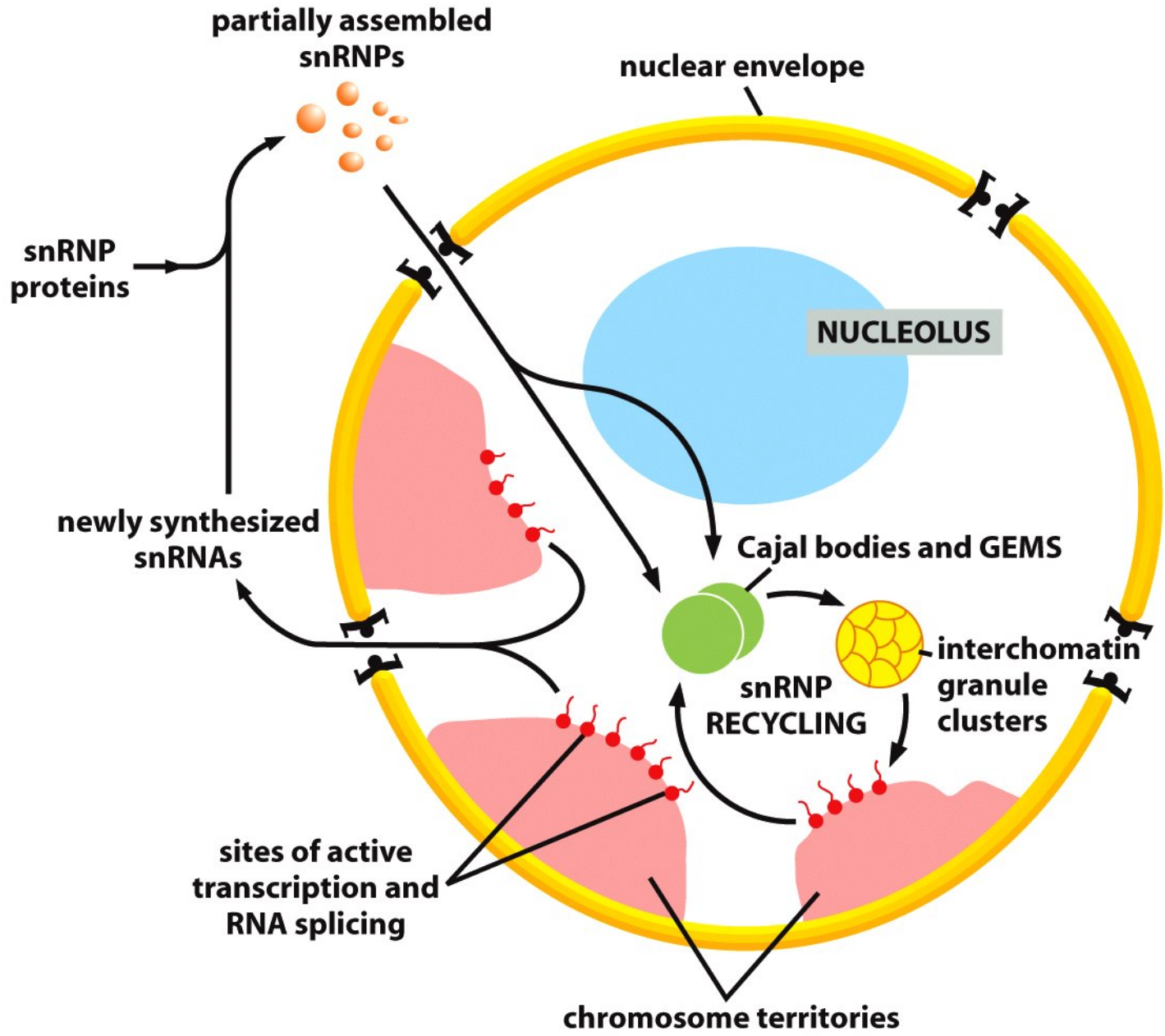


Figure 6-49 Molecular Biology of the Cell 5/e (© Garland Science 2008)

CELL 365

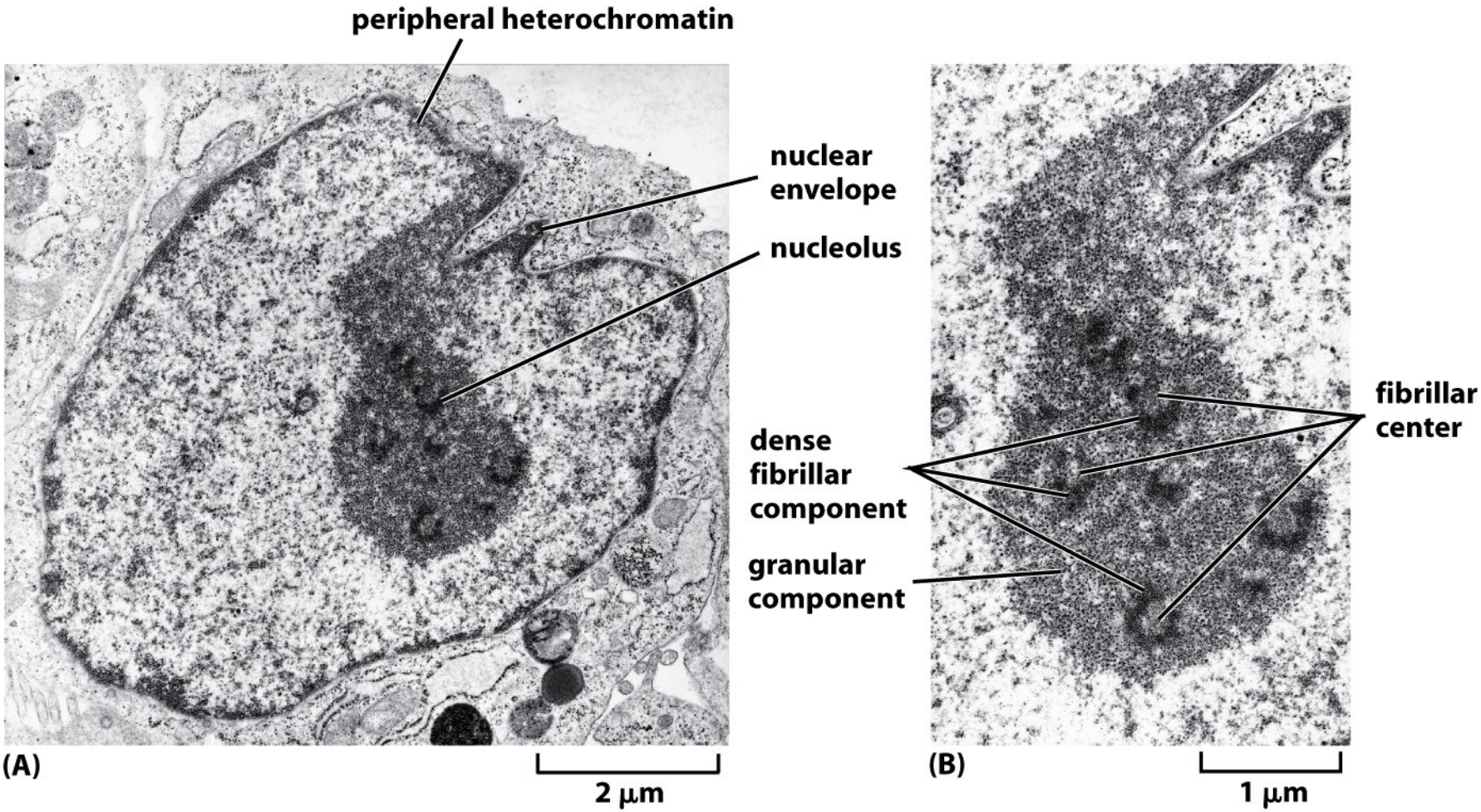


Figure 6-44 Molecular Biology of the Cell 5/e (© Garland Science 2008)

CELL 362

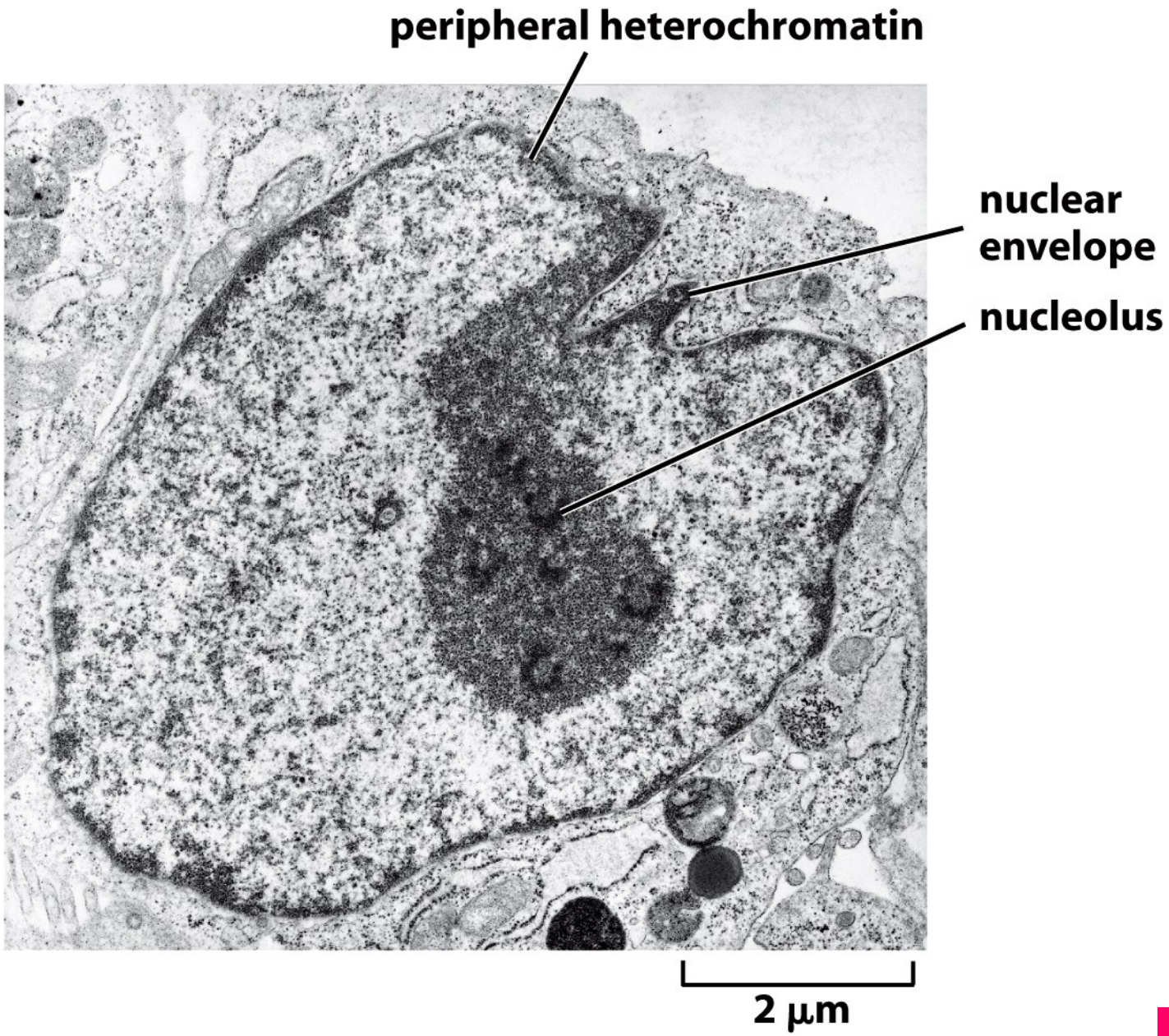


Figure 6-44a Molecular Biology of the Cell 5/e (© Garland Science 2008)

CELL 362

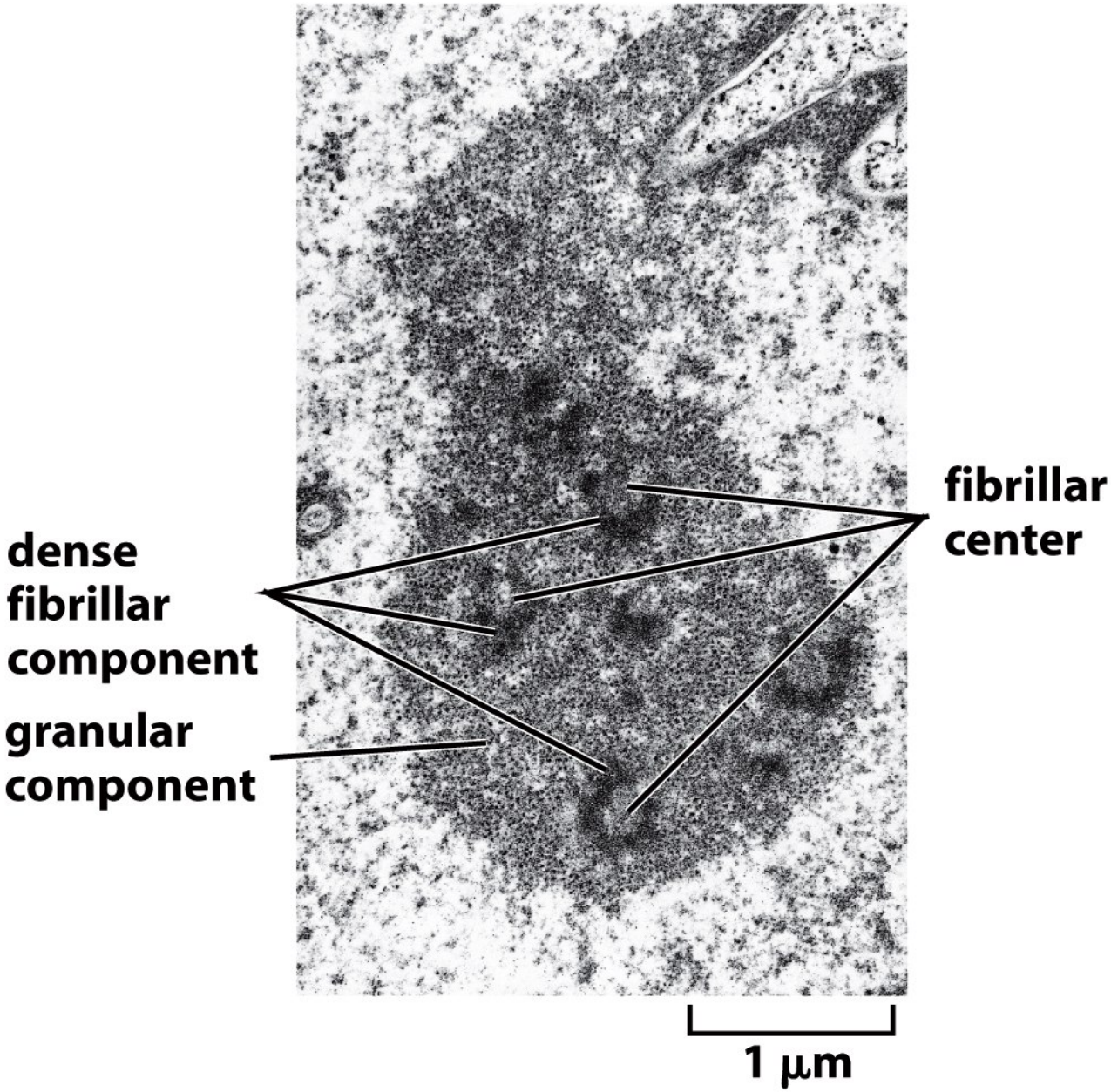


Figure 6-44b Molecular Biology of the Cell 5/e (© Garland Science 2008)

CELL 362

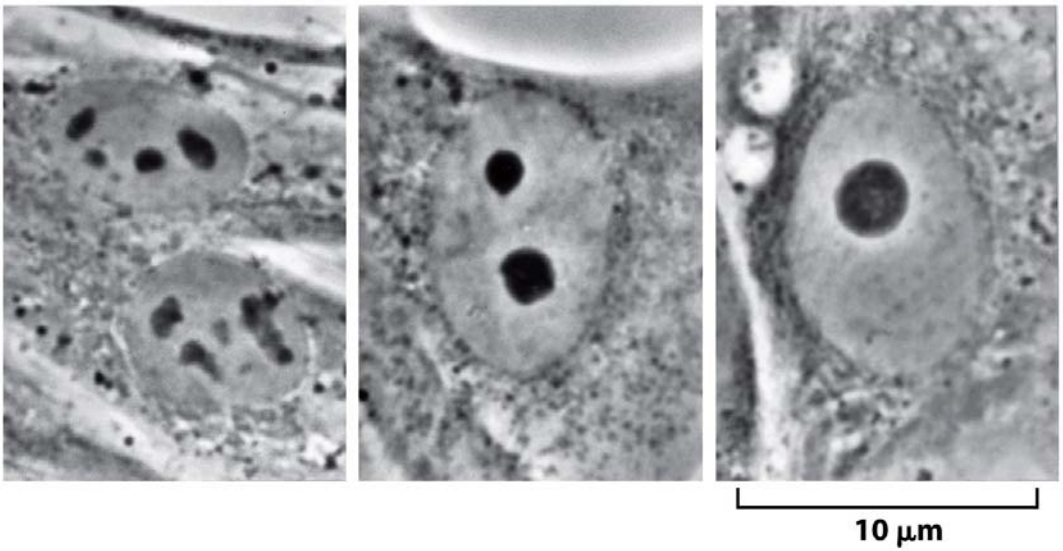


Figure 6-46 Molecular Biology of the Cell 5/e (© Garland Science 2008)

Mitoosin jälkeen kaikki 10 ihmisen NOR- segmenttiä alkavat muodostaa nukleolusta, jotka sitten fuusioituvat yhdeksi kalvottomaksi rykelmäksi

CELL 363

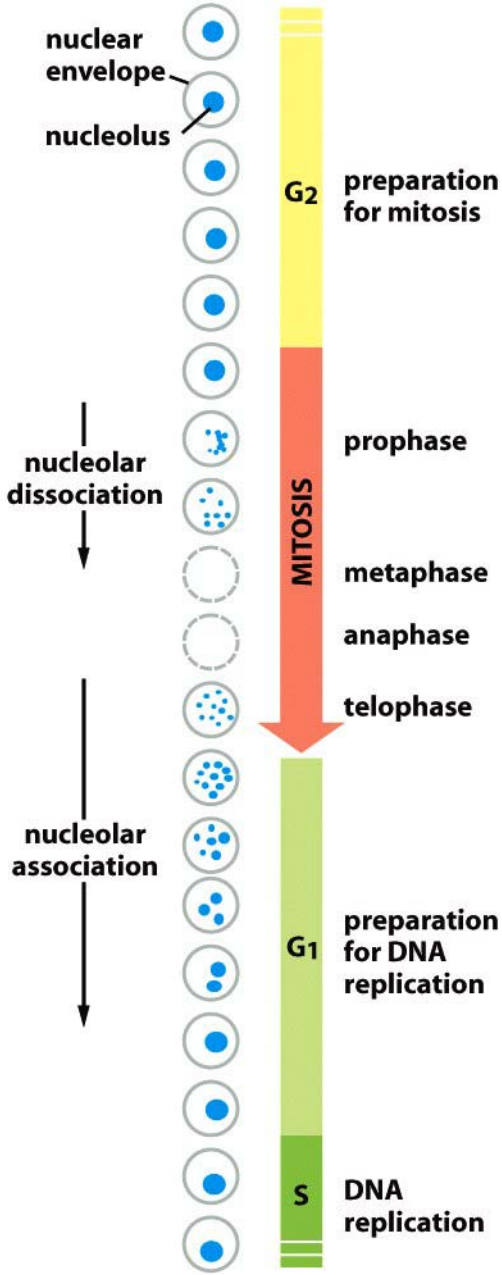


Figure 6-45 Molecular Biology of the Cell 5/e (© Garland Science 2

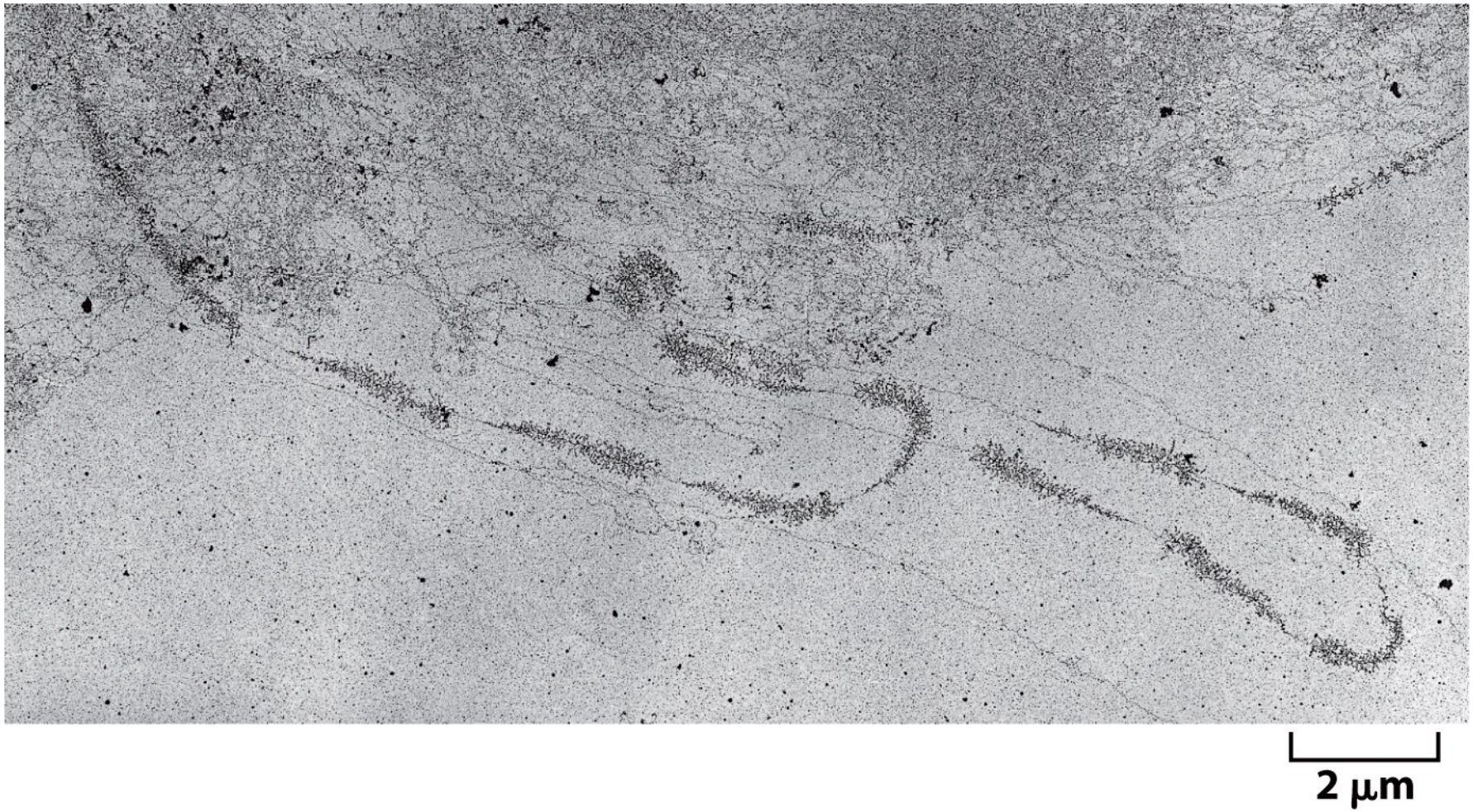


Figure 6-41 Molecular Biology of the Cell 5/e (© Garland Science 2008)

CELL 361

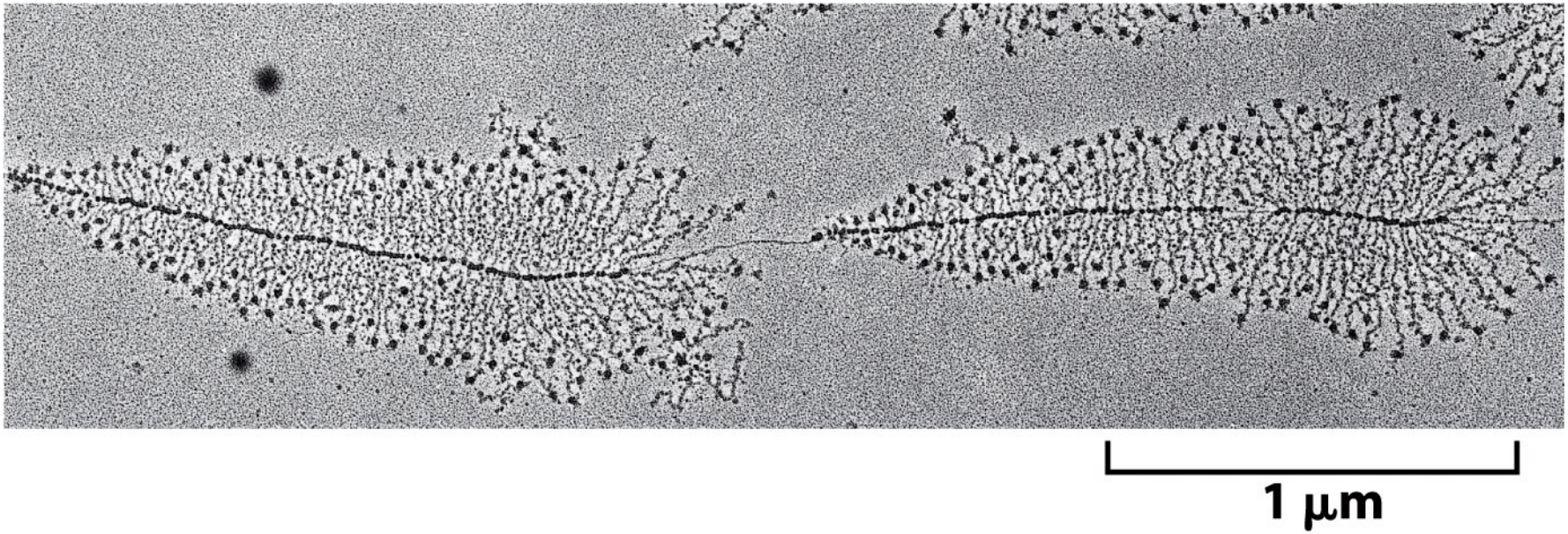


Figure 6-9 Molecular Biology of the Cell 5/e (© Garland Science 2008)

CELL 335

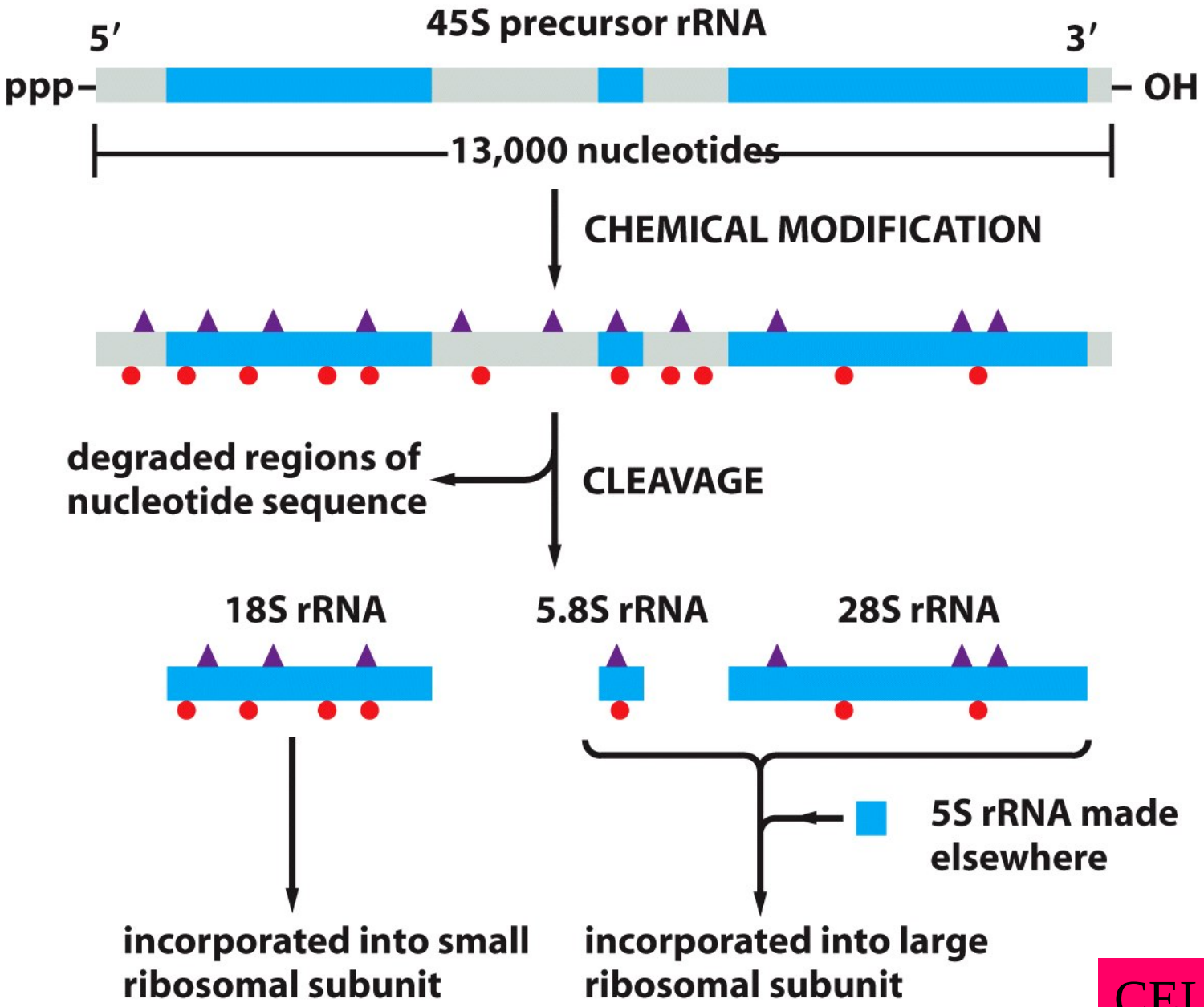
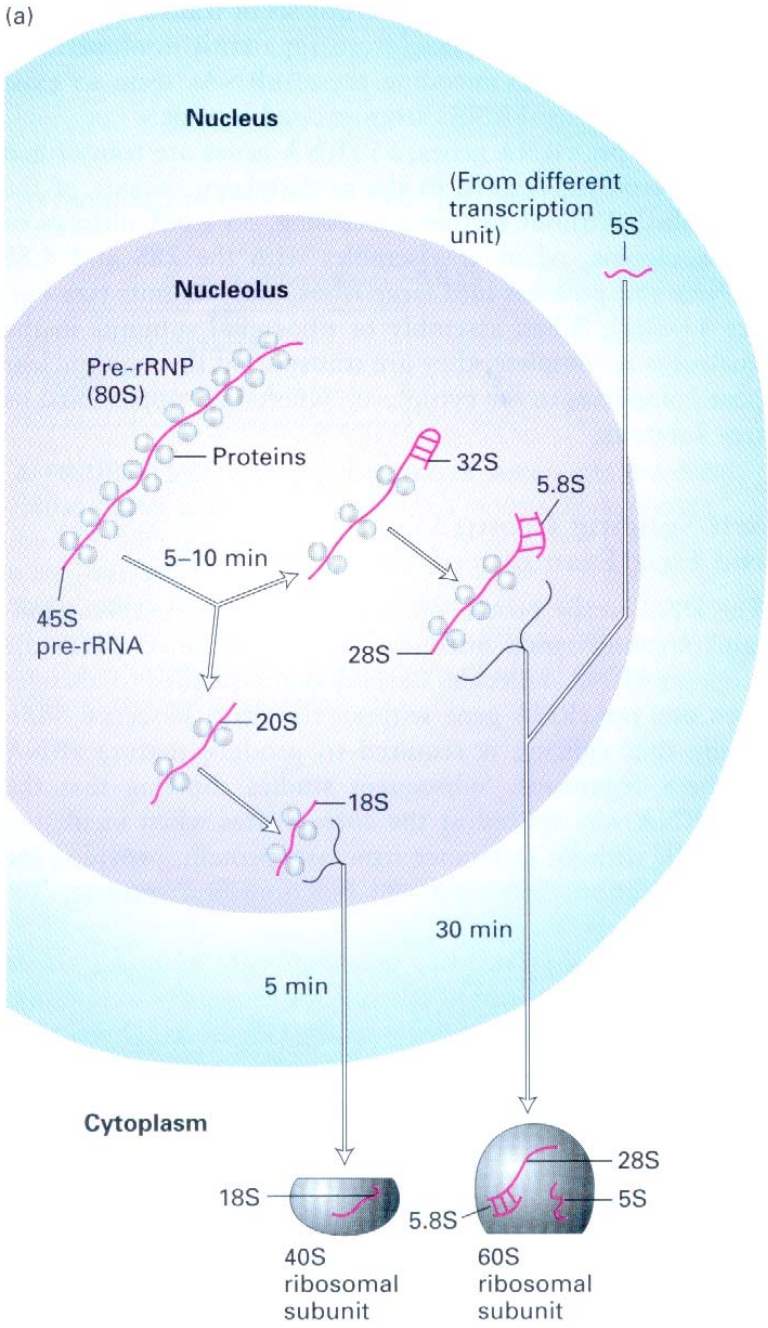


Figure 6-42 Molecular Biology of the Cell 5/e (© Garland Science 2008)

CELL 361



Tumajyvänen (nucleolus) on ribosomi- tehdas

CELL 364

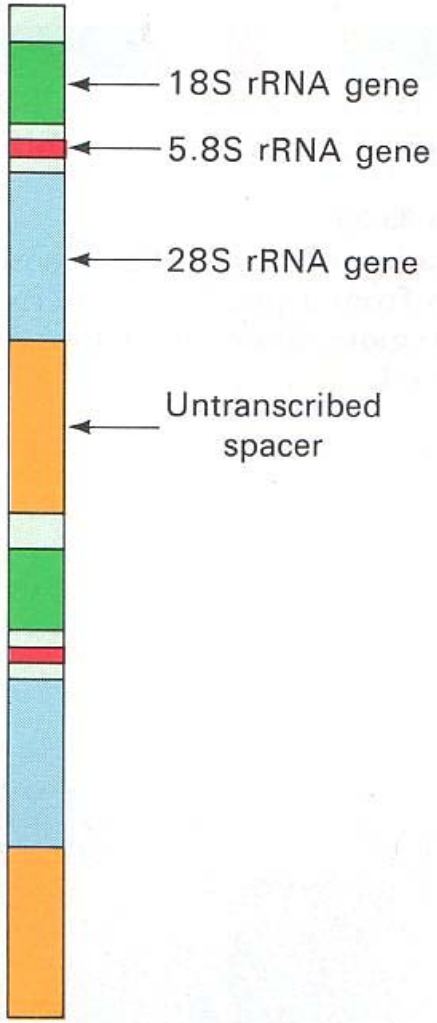


Figure 33-26
Organization of the genes for the 40S precursor of 18S, 5.8S, and 28S rRNAs in *Xenopus*. The tandemly repeated genes are separated by untranscribed spacers (yellow). The repeating unit is about 13 kb long.

RibosomiRNA –geenit ovat järjestäytyneet tandem-toistoiksi

Evoluution kuluessa jotkut voimat tai tapahtumat pitävät toistot identtisinä (concerted evolution), mikä on yksi ihmeteltävistä asioista

Ribosomigeenien eri segmenttejä käytetään paljon erilaisissa evoluutiotutkimuksissa

Koodaavat alueet ovat konservatiivisia

Välikkeet nopeasti muuttuvia:

- ITS internal transcribed spacer
- IGS tai UTS intergenic/untranscribed spacer

CELL 361

rDNA transkriboidaan ’kasetteina’

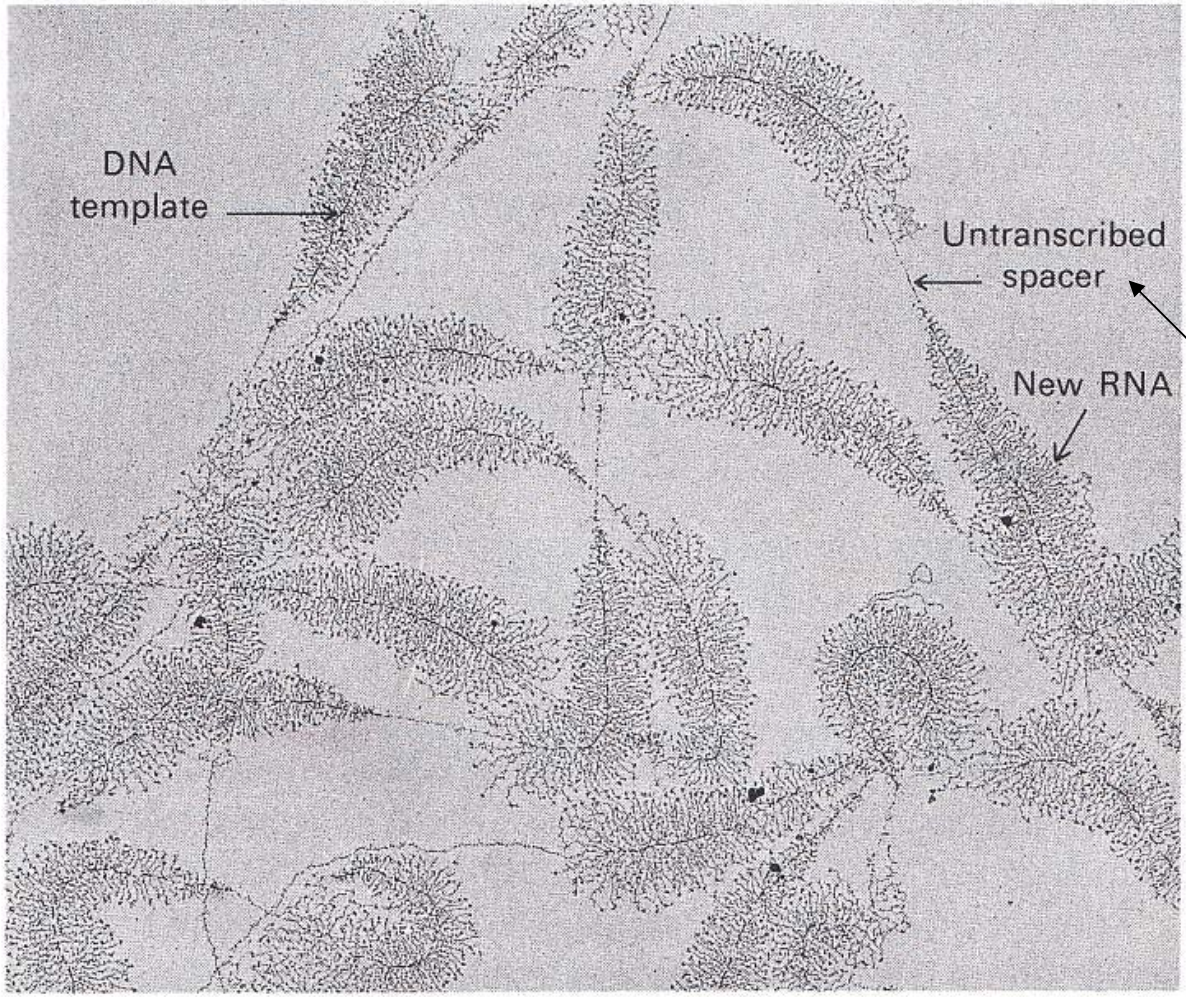
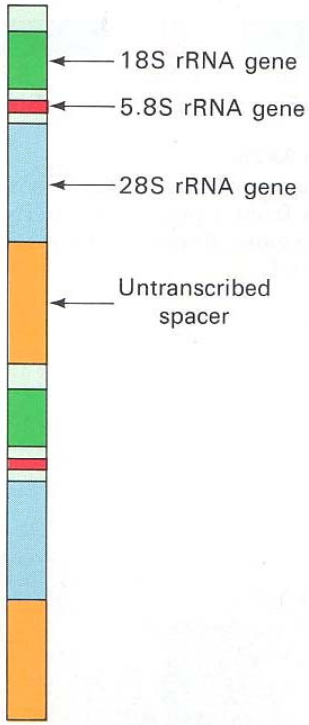


Figure 33-27
The tandem array of genes for 18S, 5.8S, and 28S rRNA is evident in this electron micrograph of nucleolar DNA. The dense axial fiber is DNA. The fine lateral fibers are newly synthesized RNA molecules with bound proteins. The tip of each arrowhead of the RNA molecules corresponds to an initiation point for transcription. The bare regions between arrowheads are the untranscribed spacers. [From O. L. Miller, Jr., and B. R. Beatty. Portrait of a gene. *J. Cell Physiol.* 74(supp. 1, 1969):225.]



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CELL 361

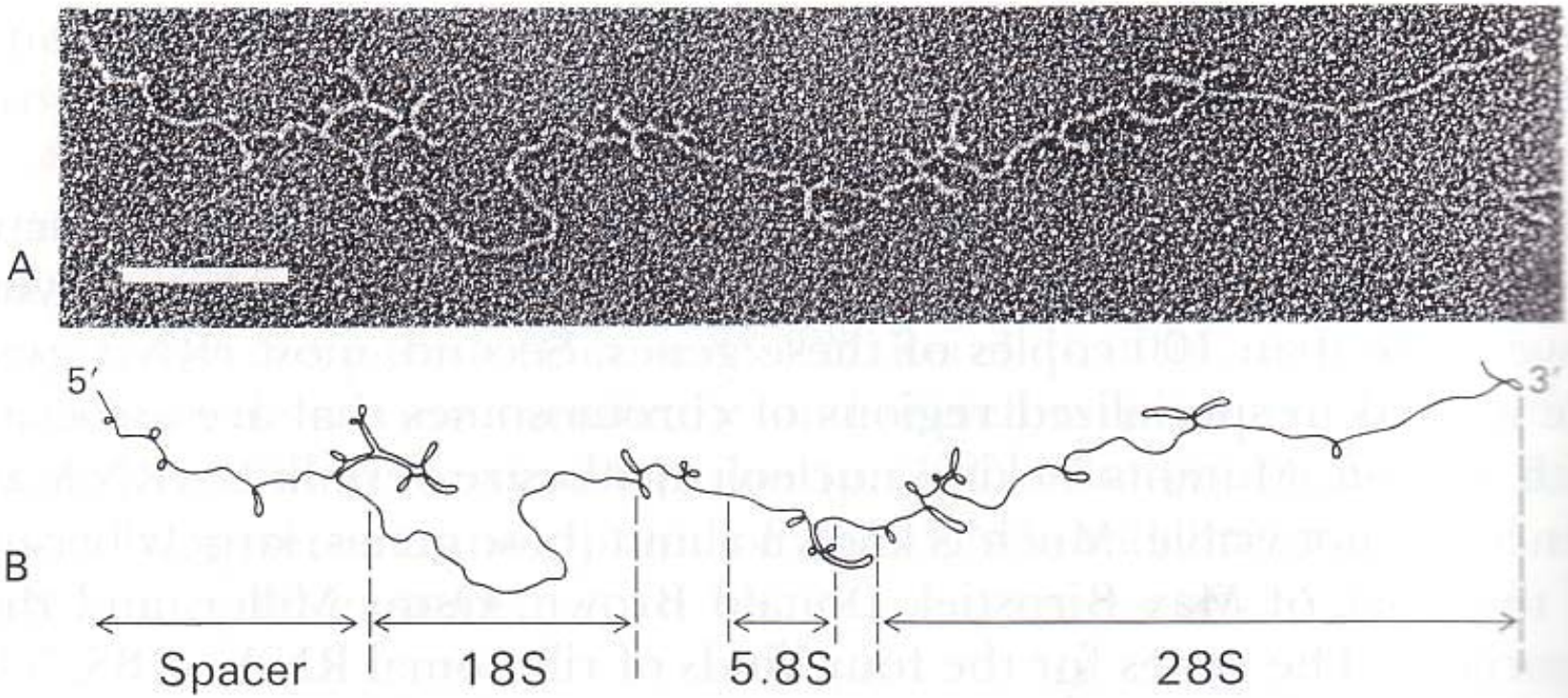
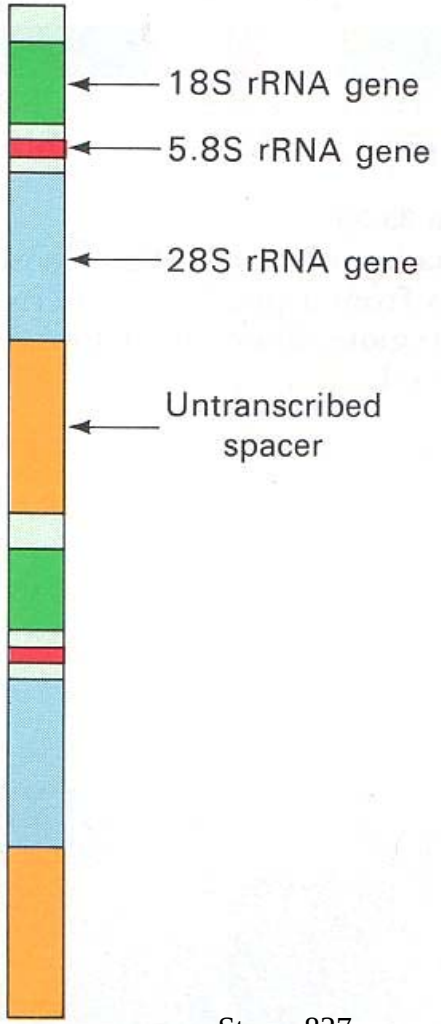


Figure 33-28
The 45S precursor of ribosomal RNA in HeLa cells has a highly distinctive pattern of hairpin loops when spread and examined by electron microscopy. This pattern makes it feasible to map the linear arrangement of 28S and 18S RNA molecules derived from this precursor. Part A is an electron micrograph of the 45S precursor (bar = 2000 Å). Part B is a tracing of the molecule shown in part A. [From P. K. Wellauer and I. B. Dawid. *Proc. Nat. Acad. Sci.* 70(1973):2828.].

Primaaritranskripti on 45S RNA, joka silputtautuu alayksiköiksi tumajyväsessä sijaitsevan tehtaan koneissa



Stryer 837

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Organization of the genes for the 40S precursor of 18S, 5.8S, and 28S rRNAs in *Xenopus*. The tandemly repeated genes are separated by untranscribed spacers (yellow). The repeating unit is about 13 kb long.

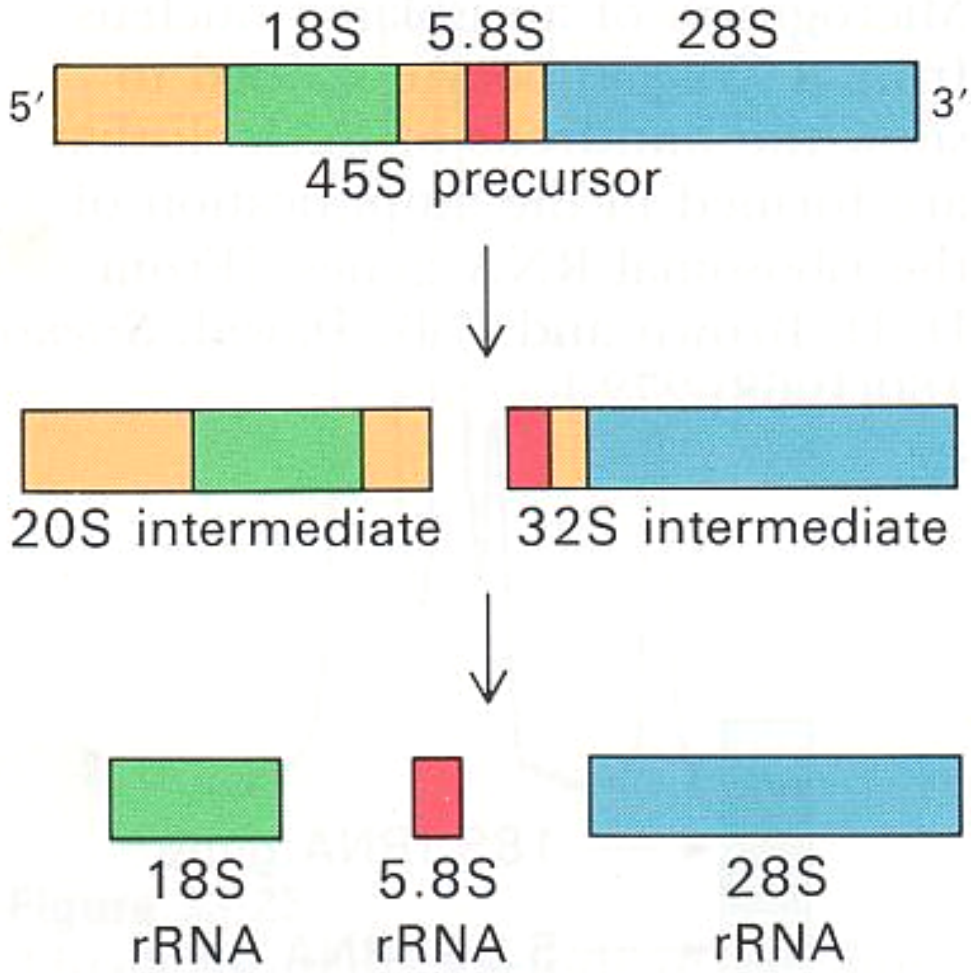


Figure 33-29
Formation of mammalian ribosomal RNAs from a primary transcript. The regions shown in yellow are removed.

Stryer 838

CELL 361

Opas-RNAt muuntelee rRNA:ta

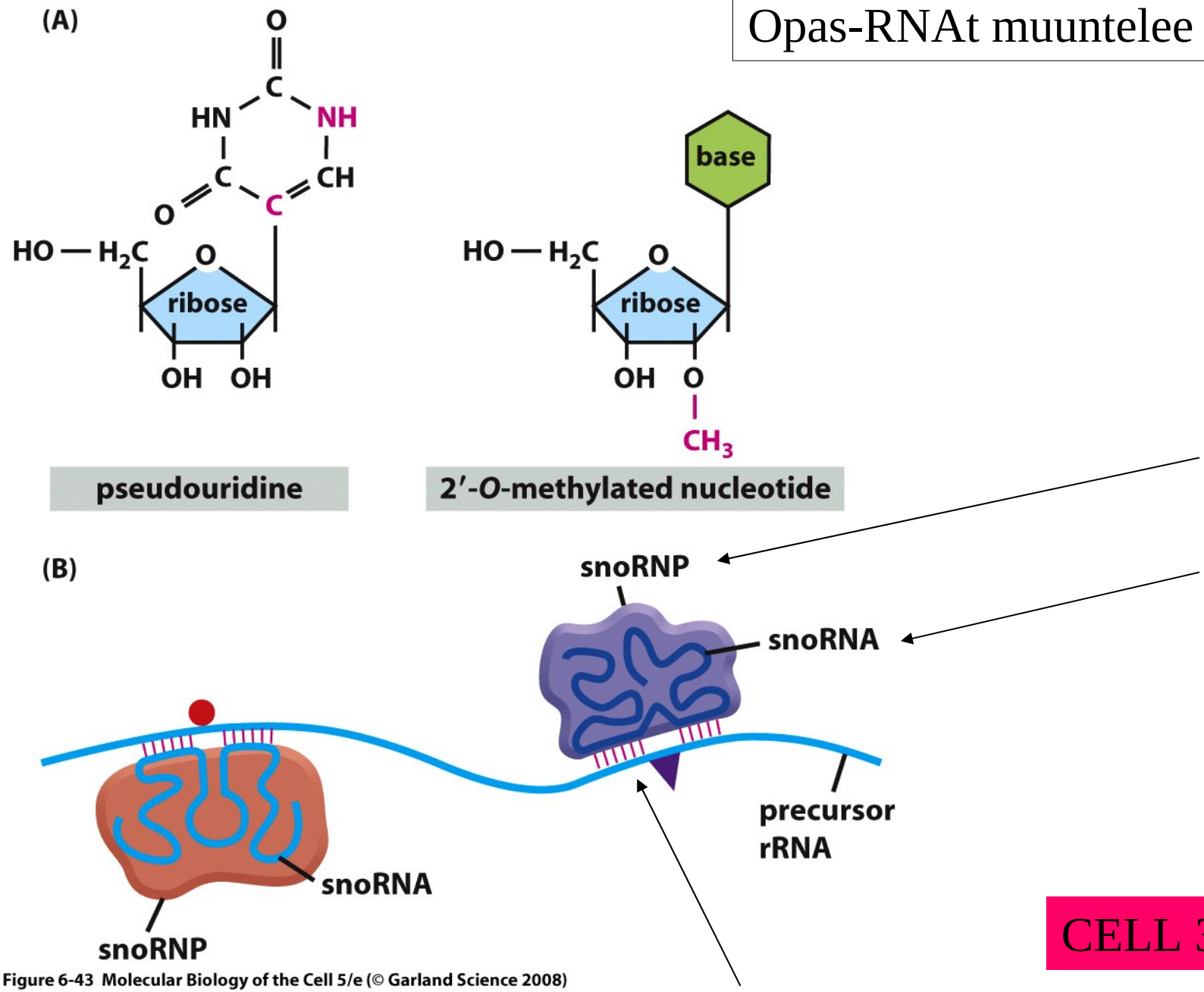
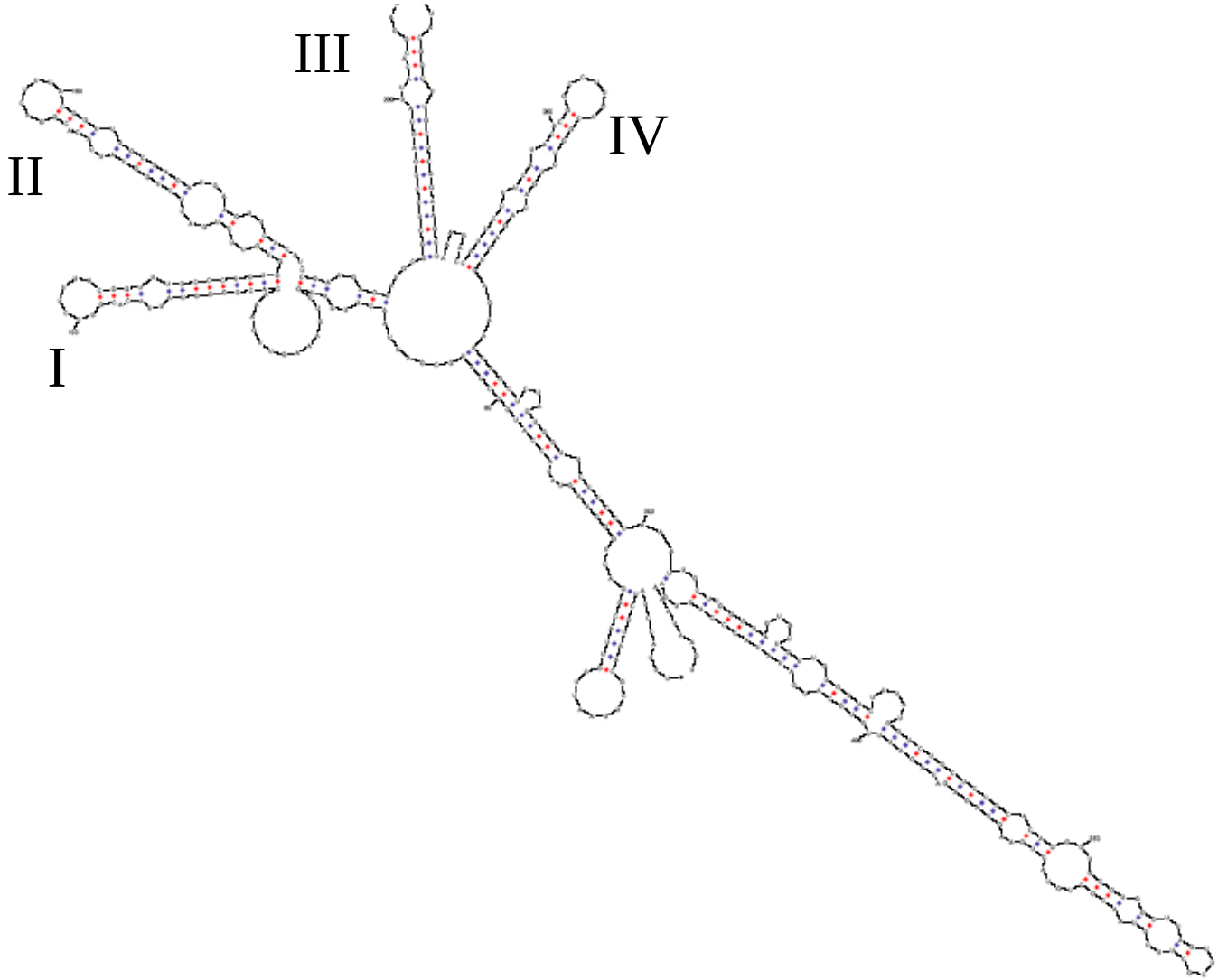


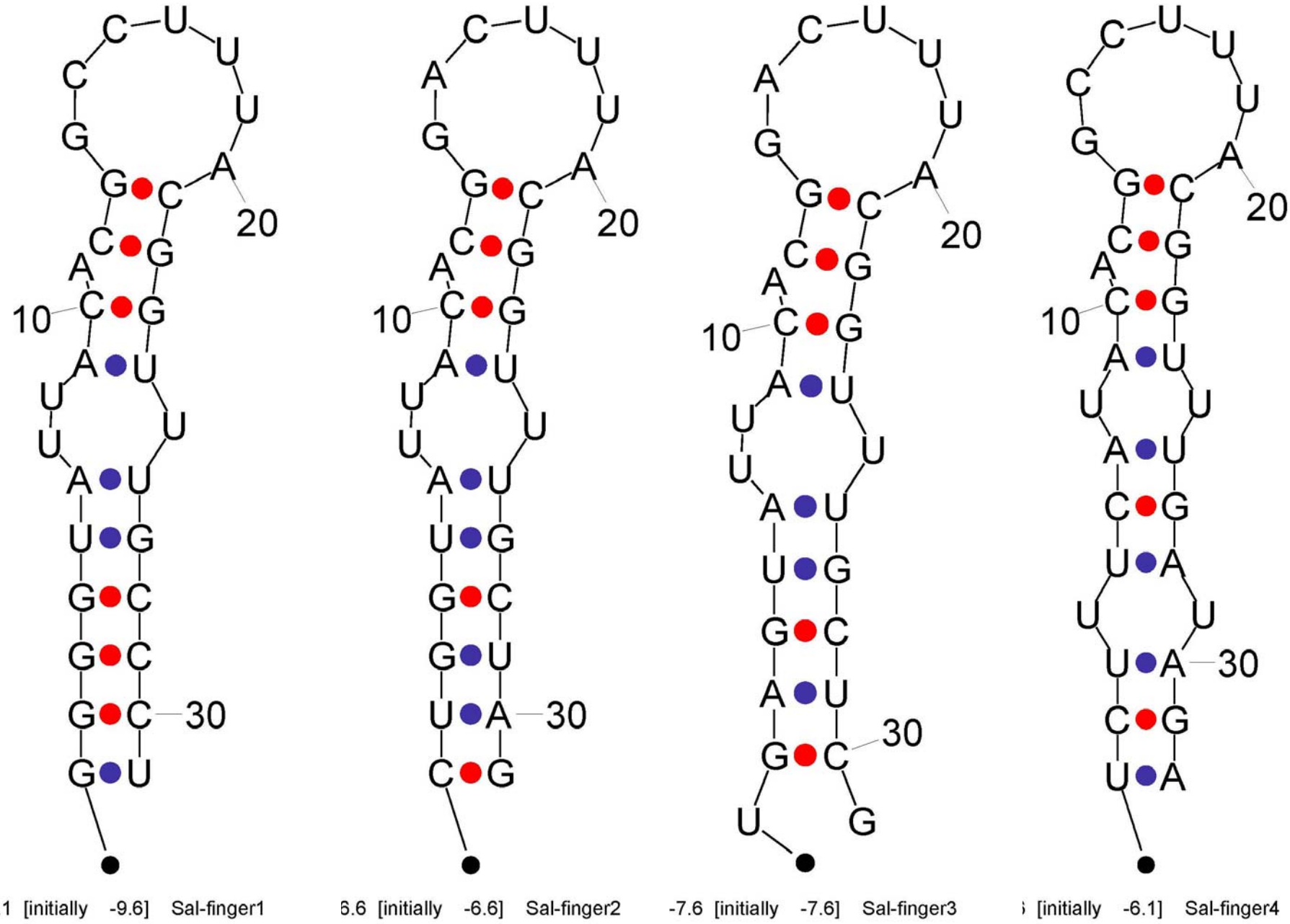
Figure 6-43 Molecular Biology of the Cell 5/e (© Garland Science 2008)

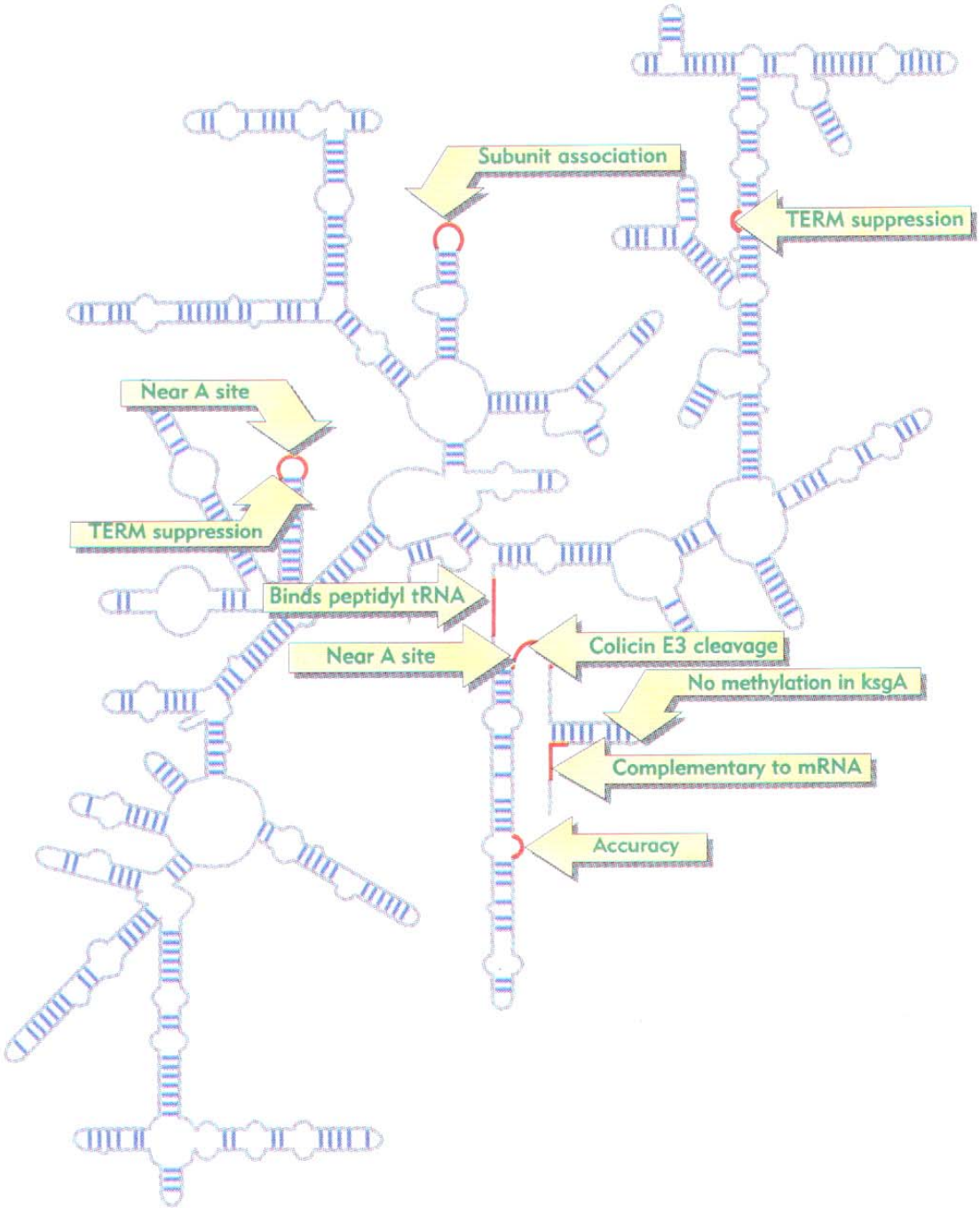
CELL 362



Lohen *Gyrodactylus*-loisen ITS2 rRNA:n sekundaaristruktuuri. Ajatellaan, että tiukat hiuspinnit helpottavat tämän jakson poistoa, mutta voi se toimittaa jotain aktiivistakin. Ei tiedetä.

Yksityiskohtia edellisestä: neljä samanlaisena toistuvaa sormea





Silputuksen jälkeen rRNA alayksiköt hankkiutuvat omiin omituisiin sekundaariasentoihin, missä tietysti niillä lukuisilla proteiineilla on oma osuutensa

CELL 378

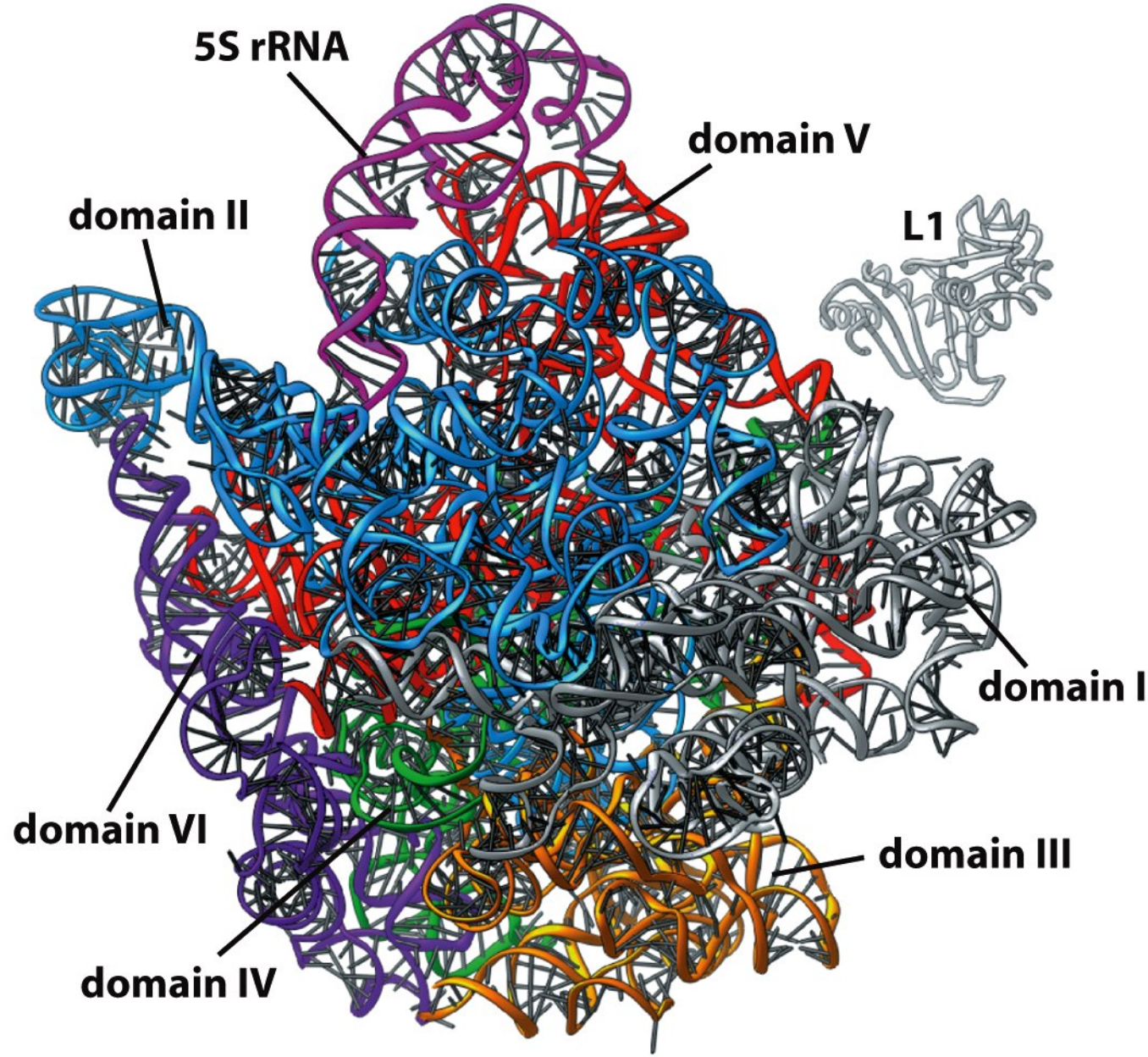


Figure 6-69a Molecular Biology of the Cell 5/e (© Garland Science 2008)

CELL 378

Bakteeri-ribosomin isomman alayksikön RNA:n laskostukset

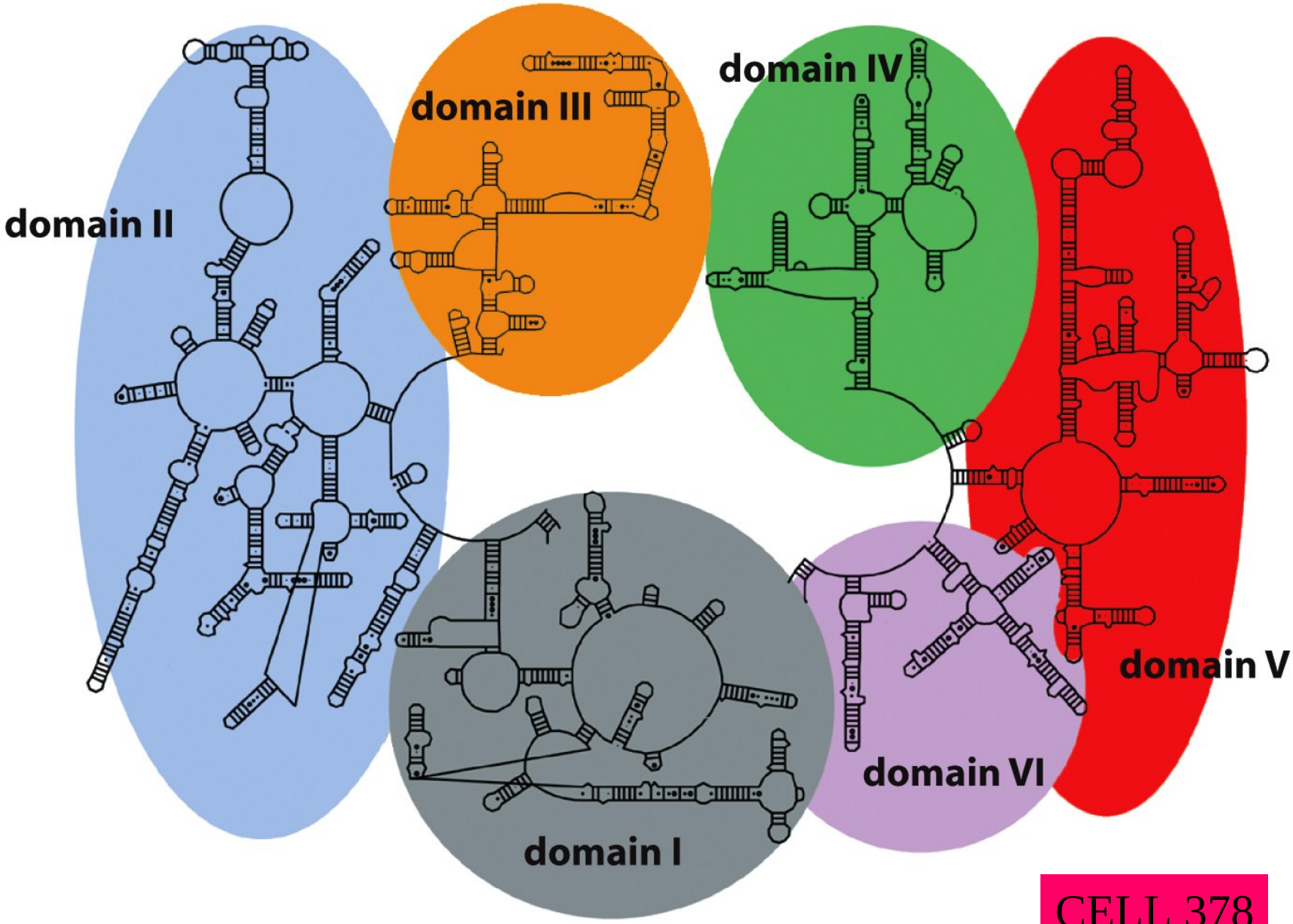


Figure 6-69b Molecular Biology of the Cell 5/e (© Garland Science 2008)

CELL 378

Bakteeri-ribosomin isomman alayksikön RNA:n laskostukset

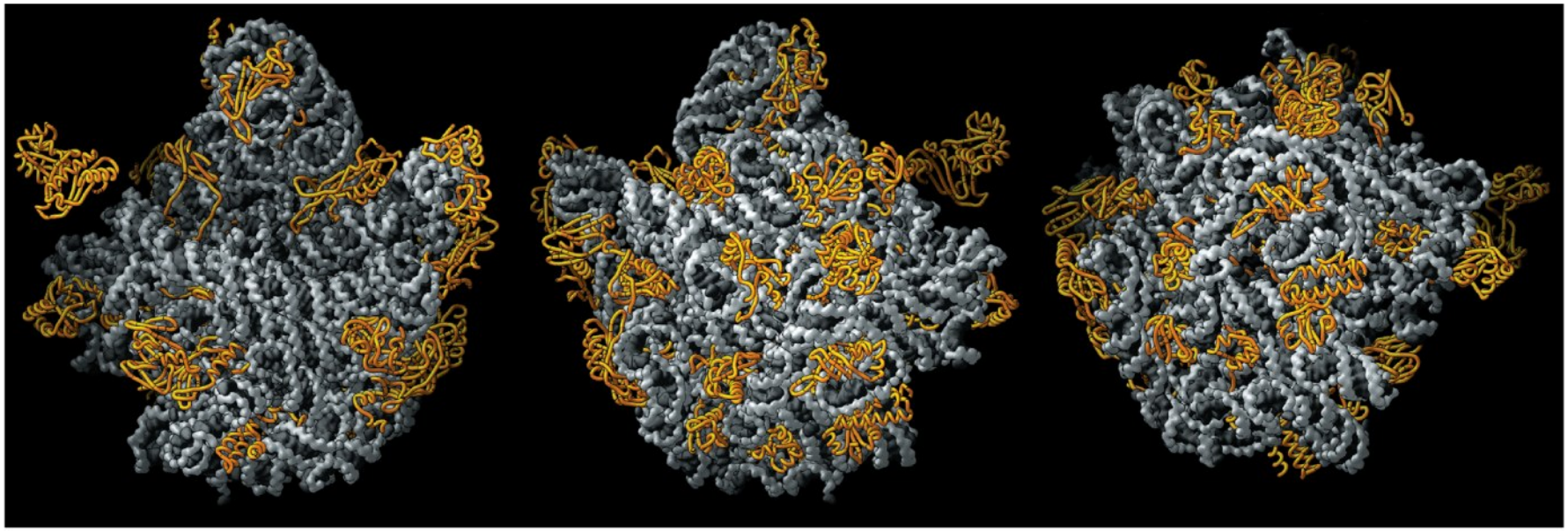


Figure 6-70 Molecular Biology of the Cell 5/e (© Garland Science 2008)

Proteiinit bakteerin LSU:ssa. Harmaa RNA, keltaiset proteiineja

CELL 379

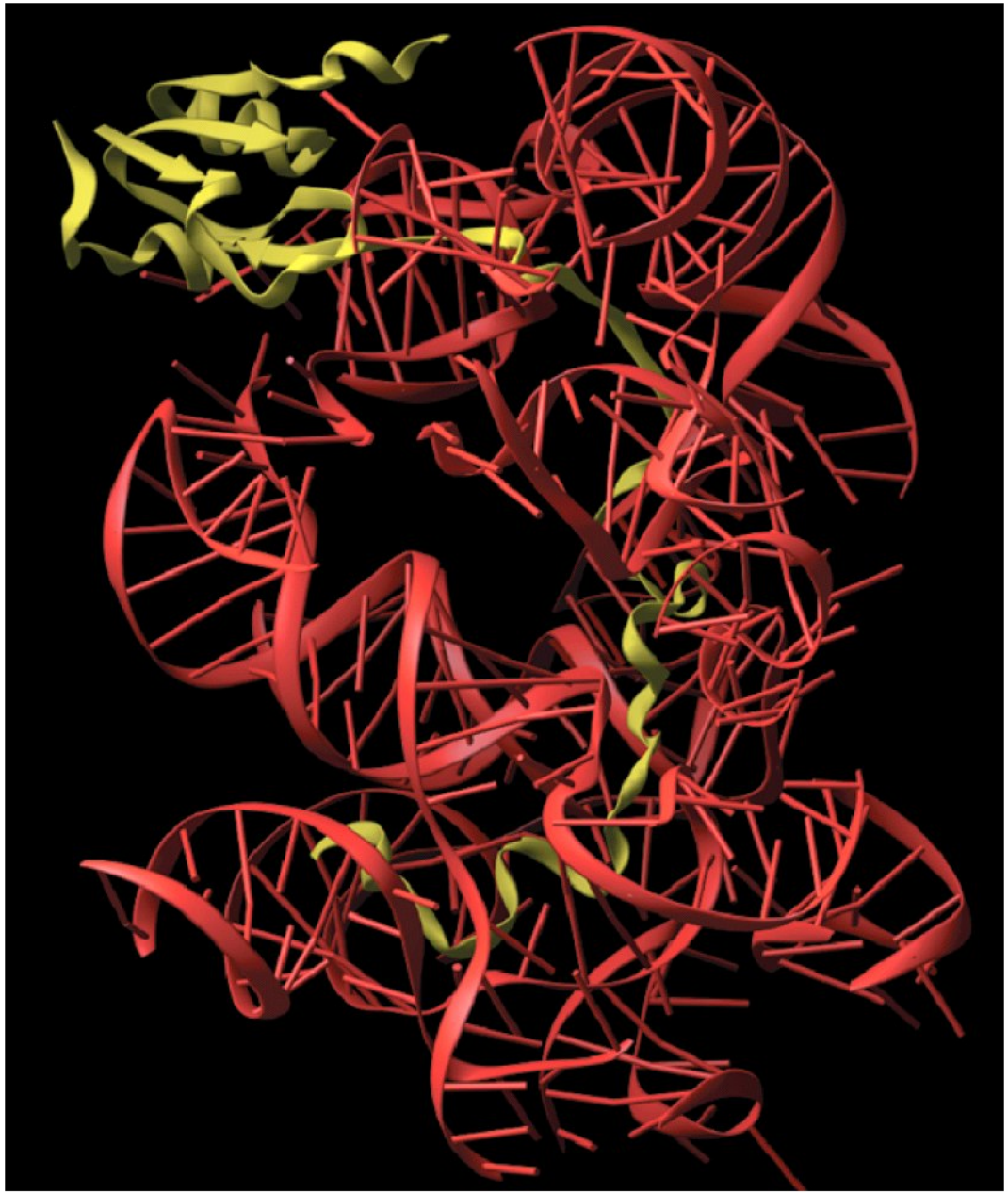


Figure 6-71 Molecular Biology of the Cell 5/e (© Garland Science 2008)

Yksi ribosomin proteiineista malliksi

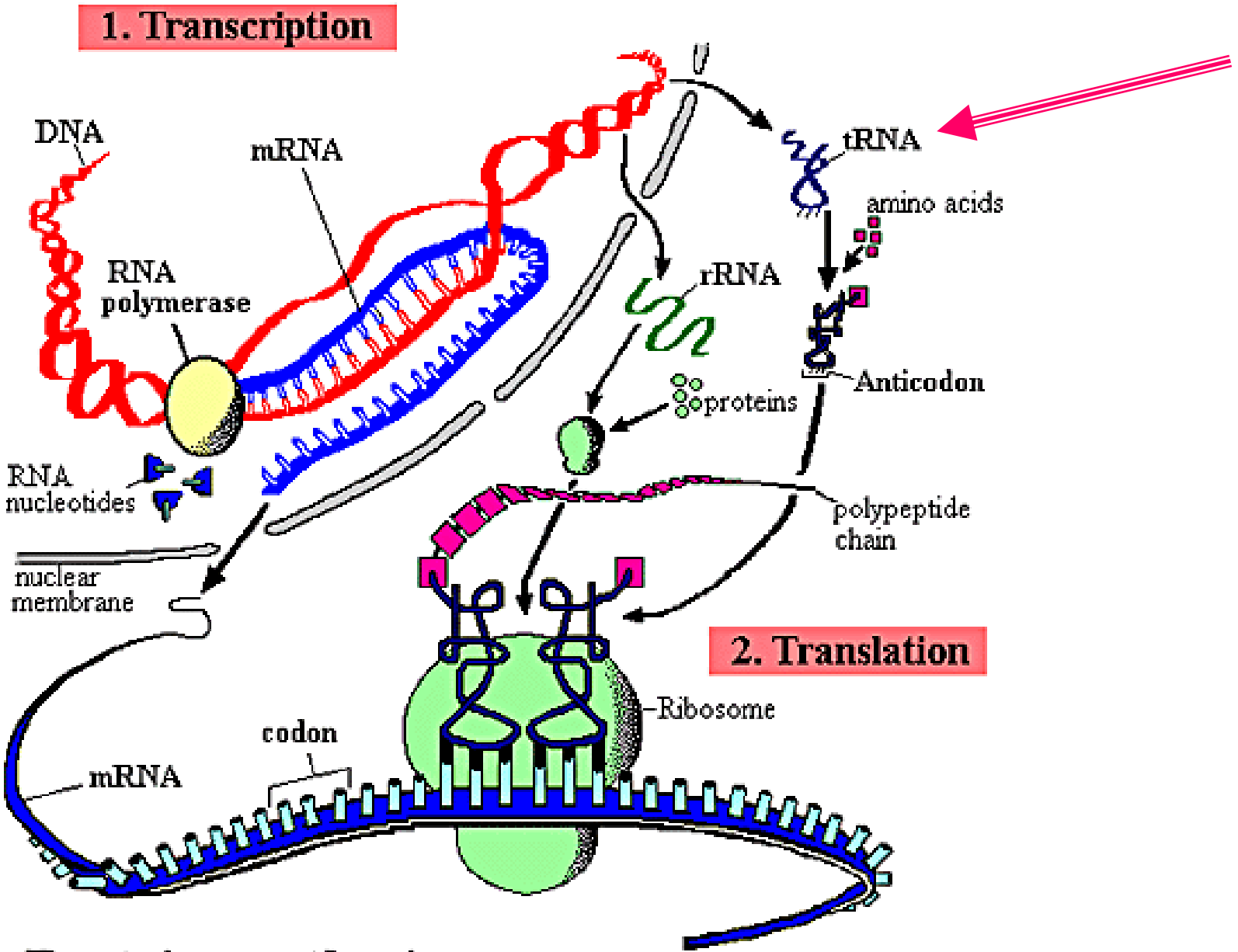
L15 (keltainen) bakteerin LSU:ssa (osa)

CELL 379

Siirtäjä-RNAeli tRNA

Siirtäjä-RNA:n keksi Robert Holley, Nobel 1968

Nobel-luento



Protein synthesis

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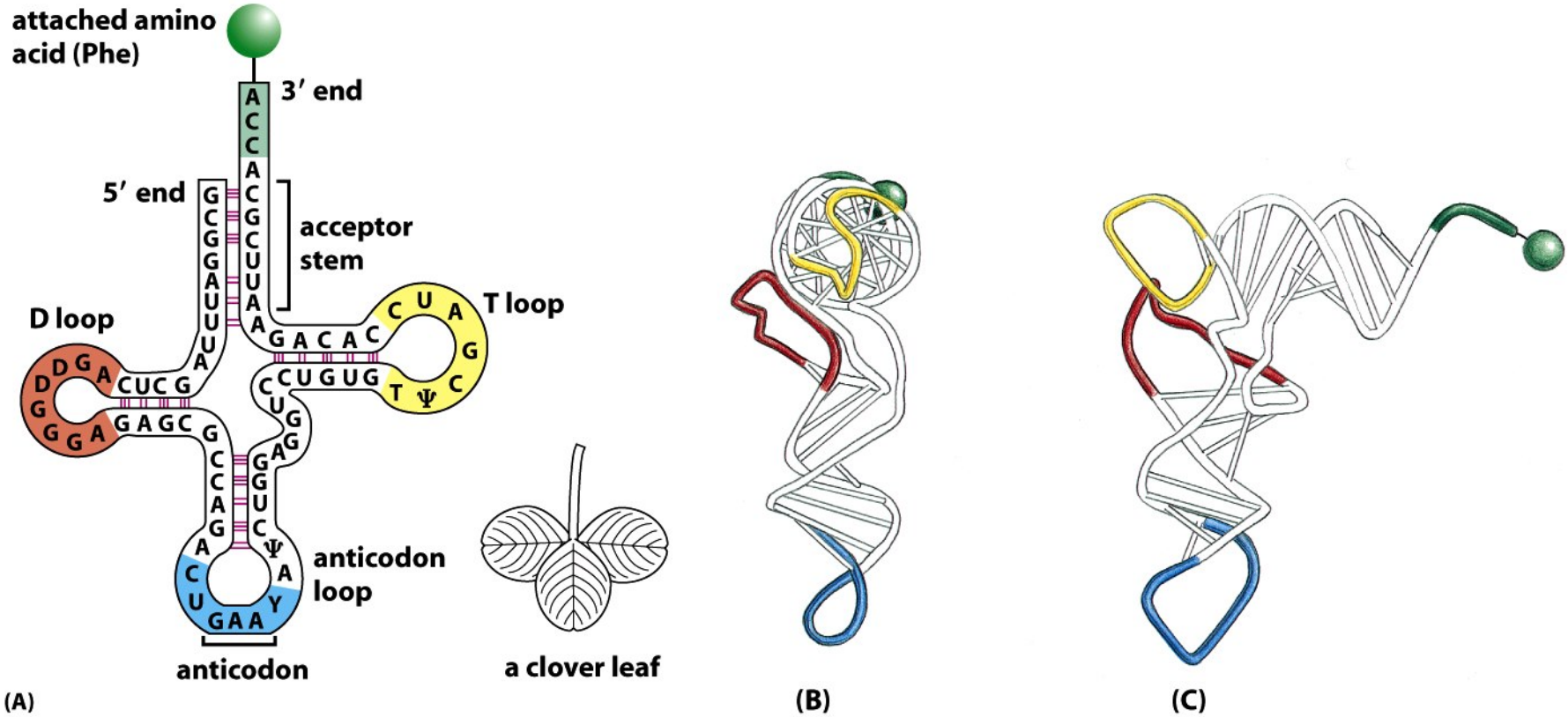
Other noncoding RNAs	function in diverse cell processes, including telomere synthesis, X-chromosome inactivation, and the transport of proteins into the ER

Table 6-1 Molecular Biology of the Cell 5/e (© Garland Science 2008)

CELL 336



5′ GCGGAUUUAGCUCAGDDGGGAGAGCGCCAGACUGAAYAΨCUGGAGGUCCUGUGTΨCGAUCCACAGAAUUCGCACCA 3′

anticodon

(D)

Figure 6-52 Molecular Biology of the Cell 5/e (© Garland Science 2008)

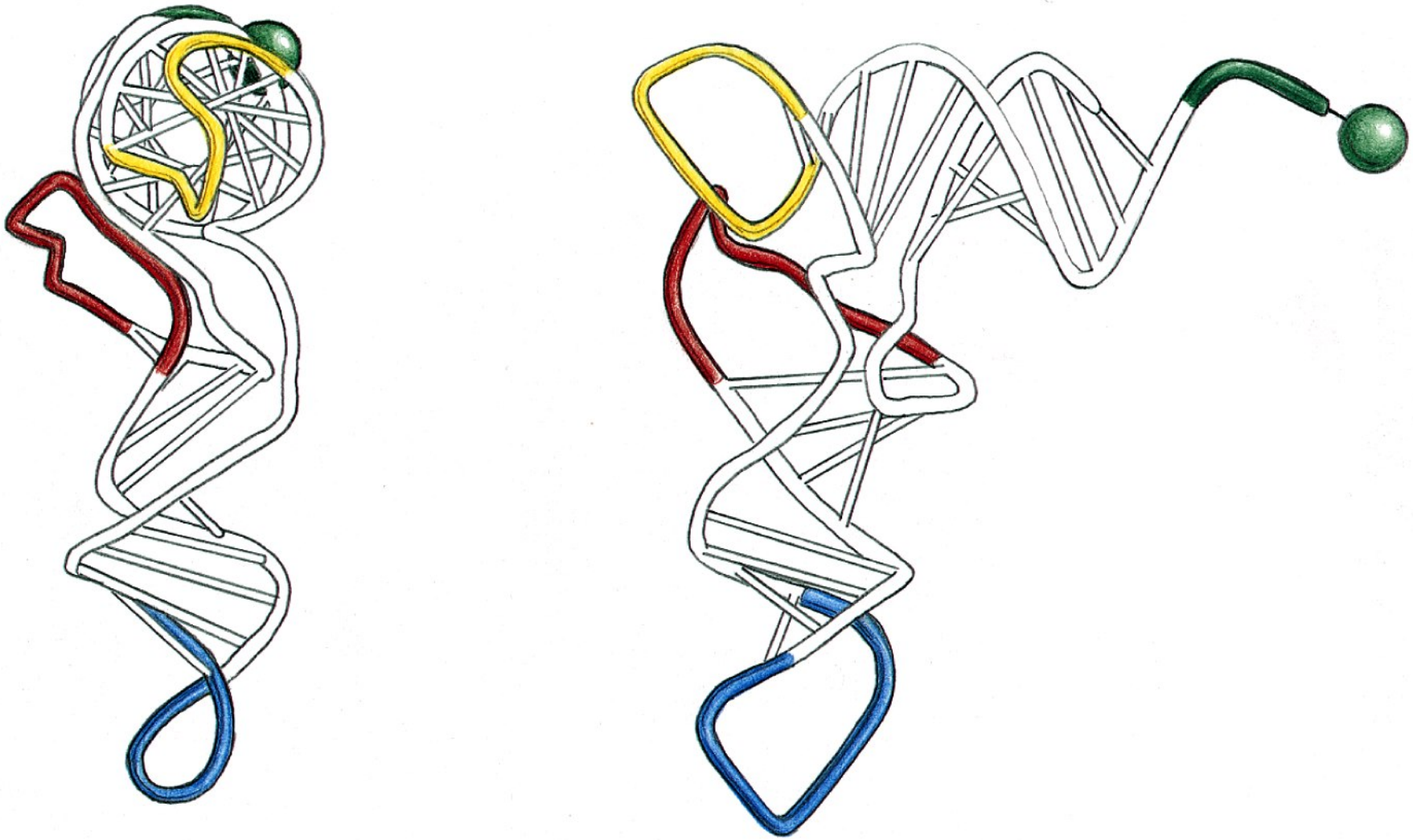
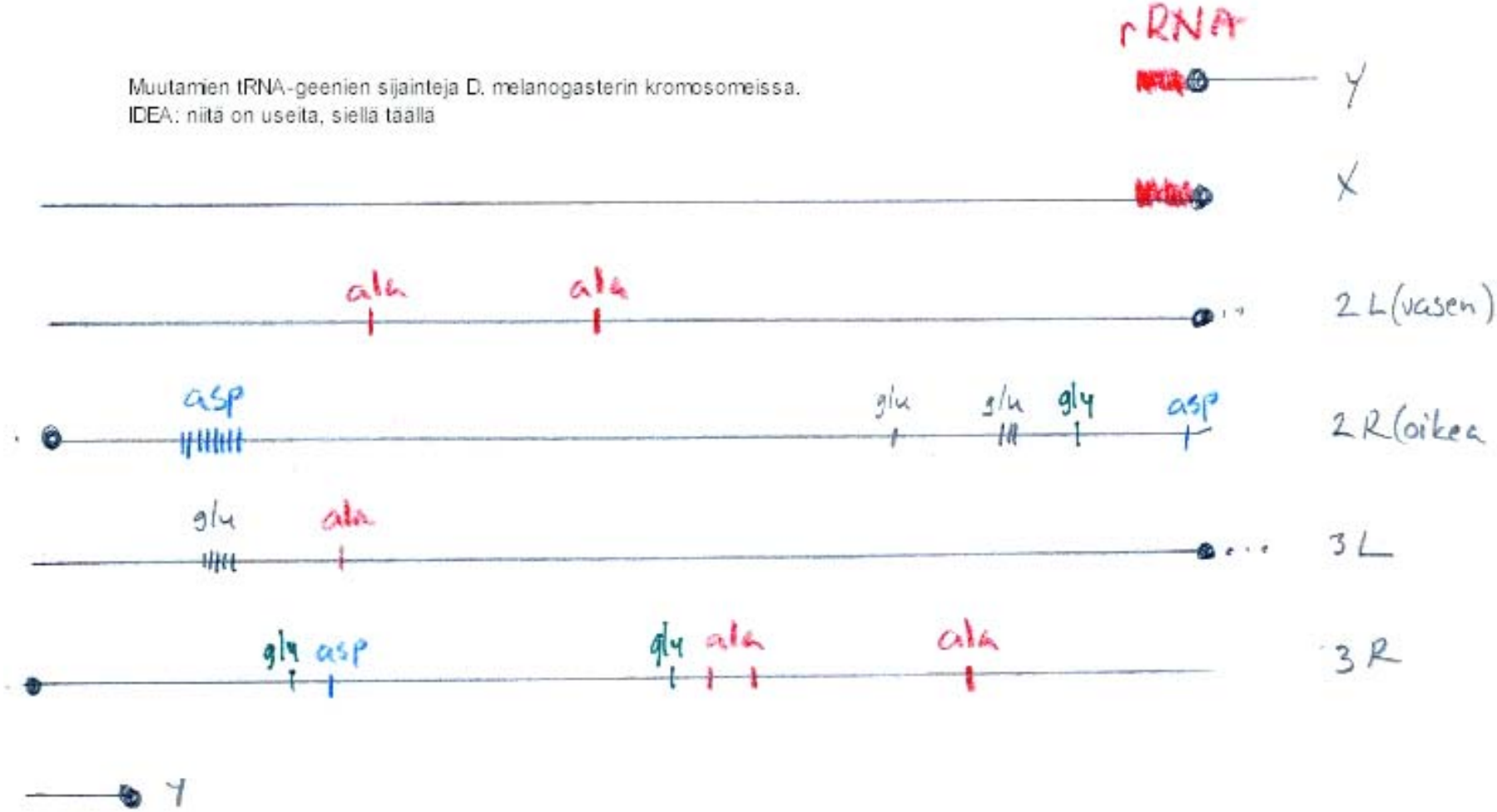


Figure 6-52d Molecular Biology of the Cell 5/e (© Garland Science 2008)



Drosophilalla on 292 erilaista tRNA-geeniä, jotka sijaitsevat ihan hajallaan kromosomeissa, ilman mitään loogista järjestystä. rDNA on kahtena blokkina, X kromosomissa ja Y:ssä

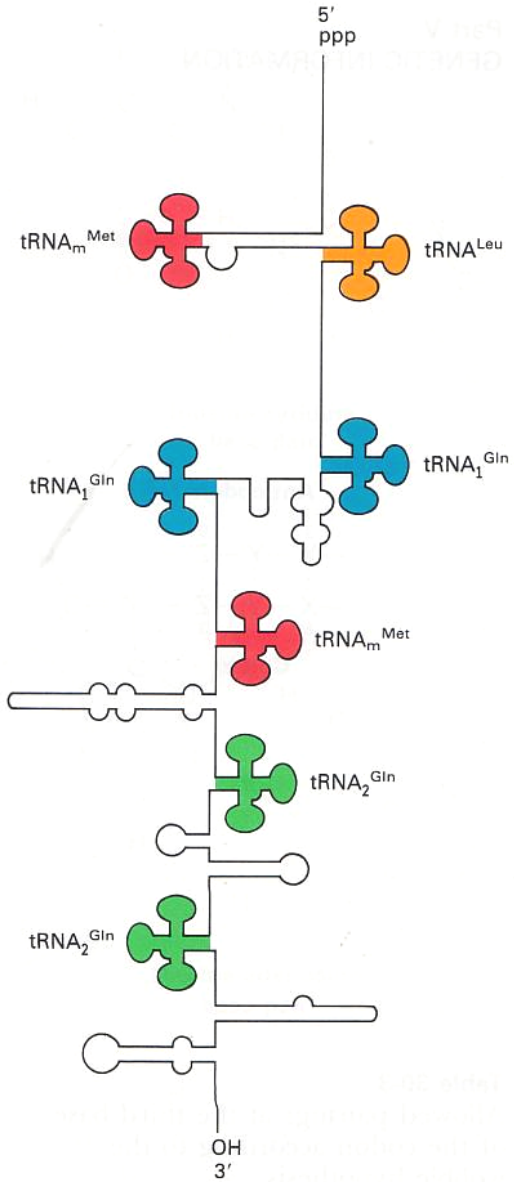


Figure 30-13
Seven tRNA molecules are formed by cleavage of this 950-nucleotide primary transcript. [After N. Nakajima, H. Ozeki, and Y. Shimura. *Cell* 23(1981):245.]

tRNA eli siirtäjäRNA geenejä transkriboidaan pitkinä hnRNA-laatuina, joista sitten silputaan varsinaisia tRNA molekyylejä

Figure 6-54 Molecular Biology of the Cell 5/e (© Garland Science 2008)

tRNA:ssa voi olla introneita, jotka silputaan pois (sininen)

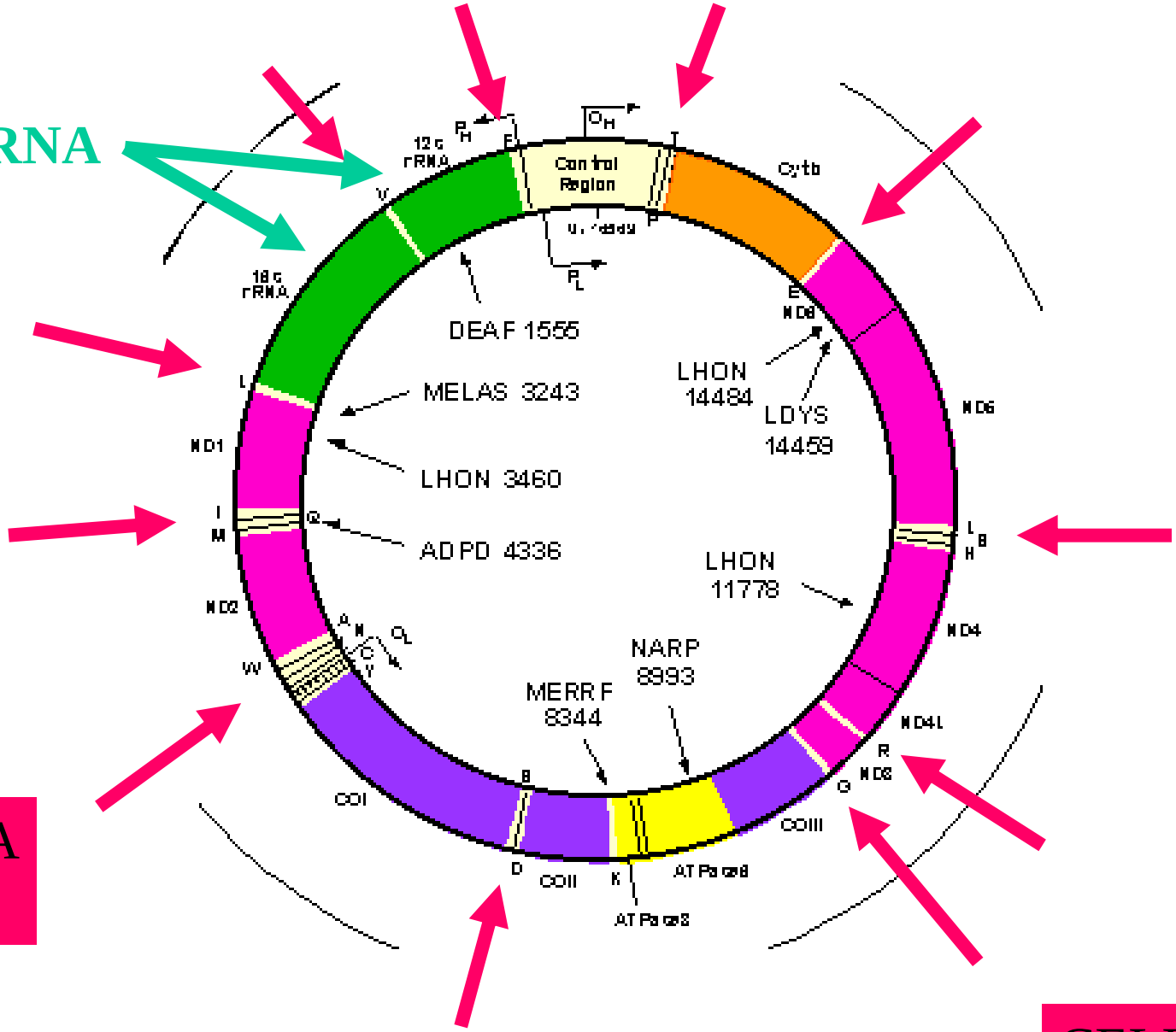
CELL 370

Figure 6-54 Molecular Biology of the Cell 5/e (© Garland Science 2008)

tRNA:ssa voi olla introneita, jotka silputaan pois (sininen)

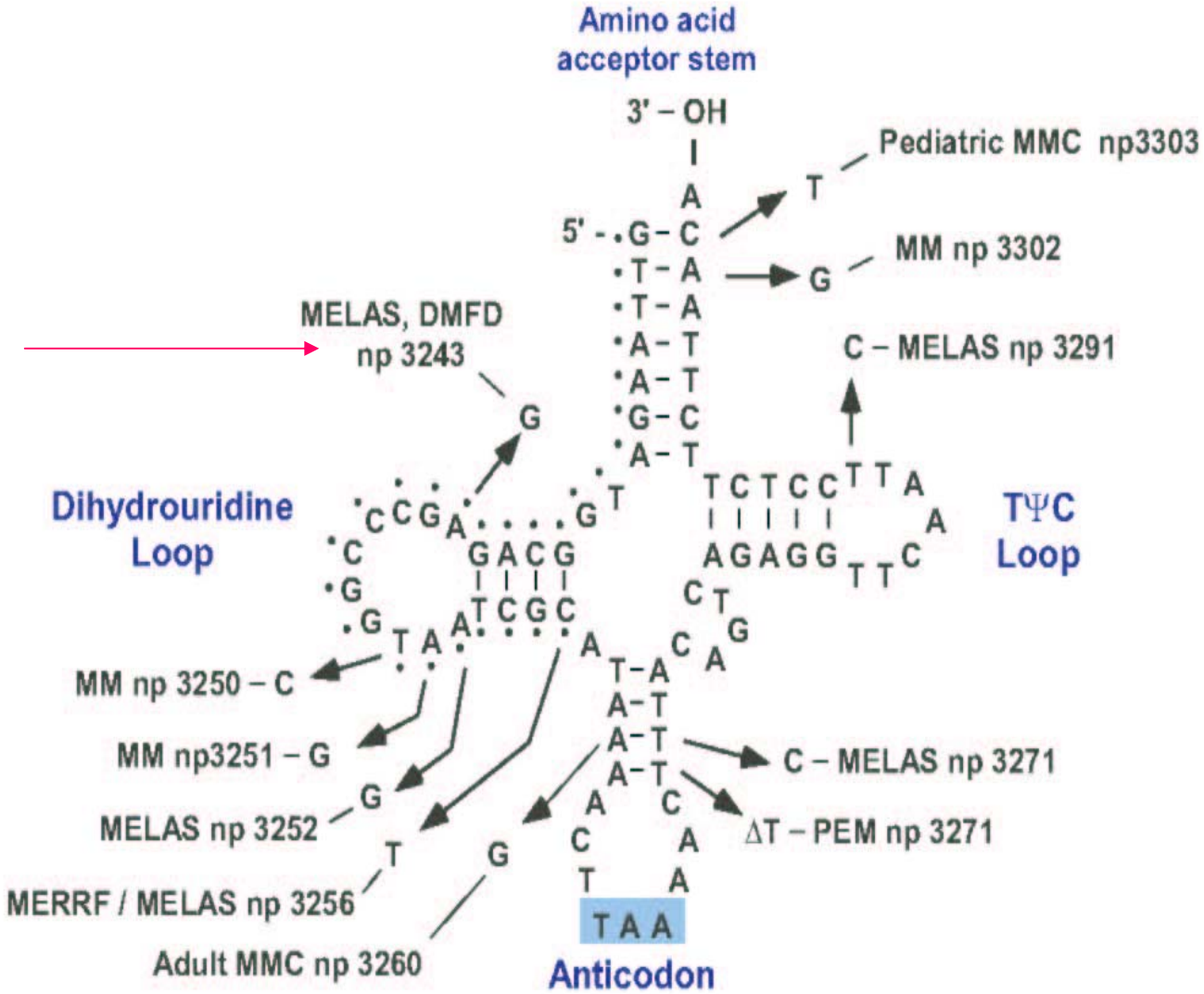
Ihmisen mitokondrio

ribosomiRNA



transferRNA
22 kpl

CELL 861



Pathological Mutations in tRNALeu(UUR)

http://www.gen.emory.edu/MITOMAP/tRNAleu.pdf

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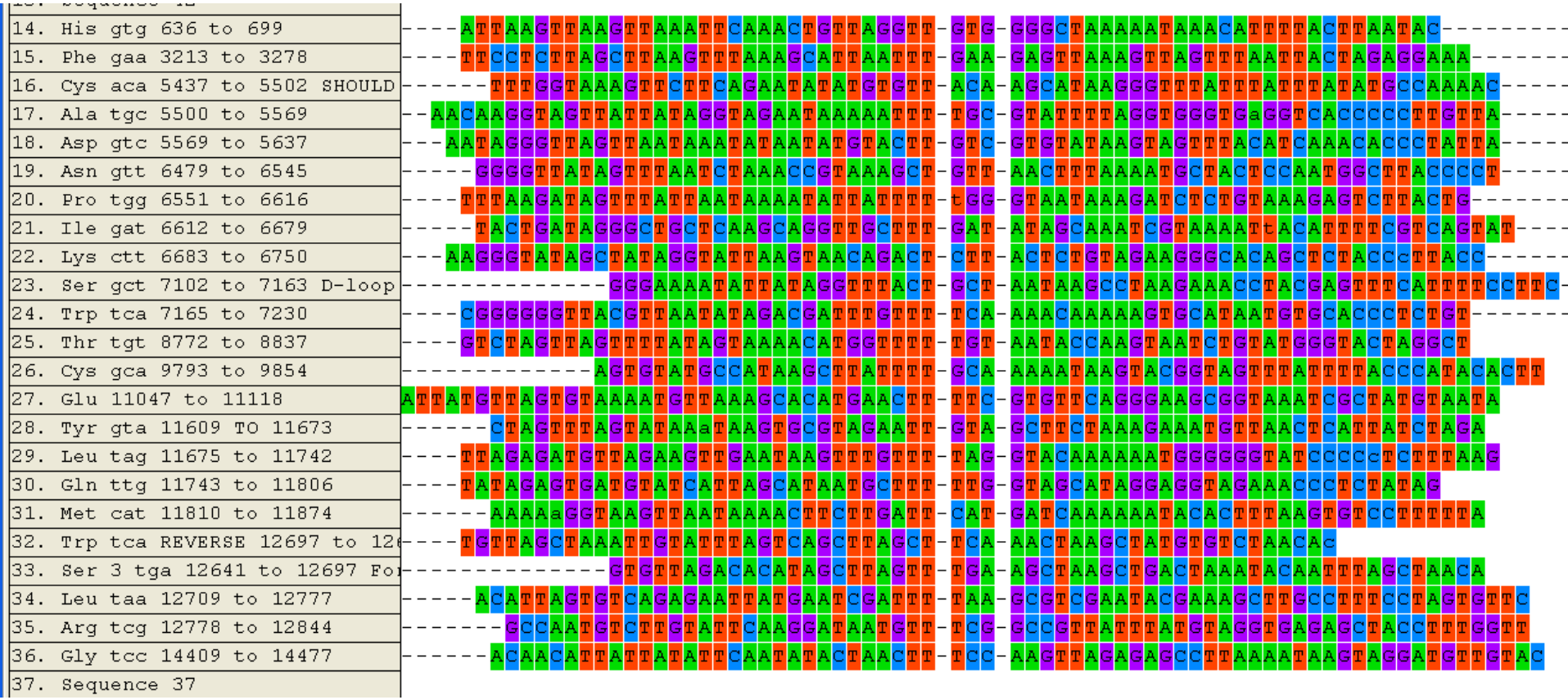
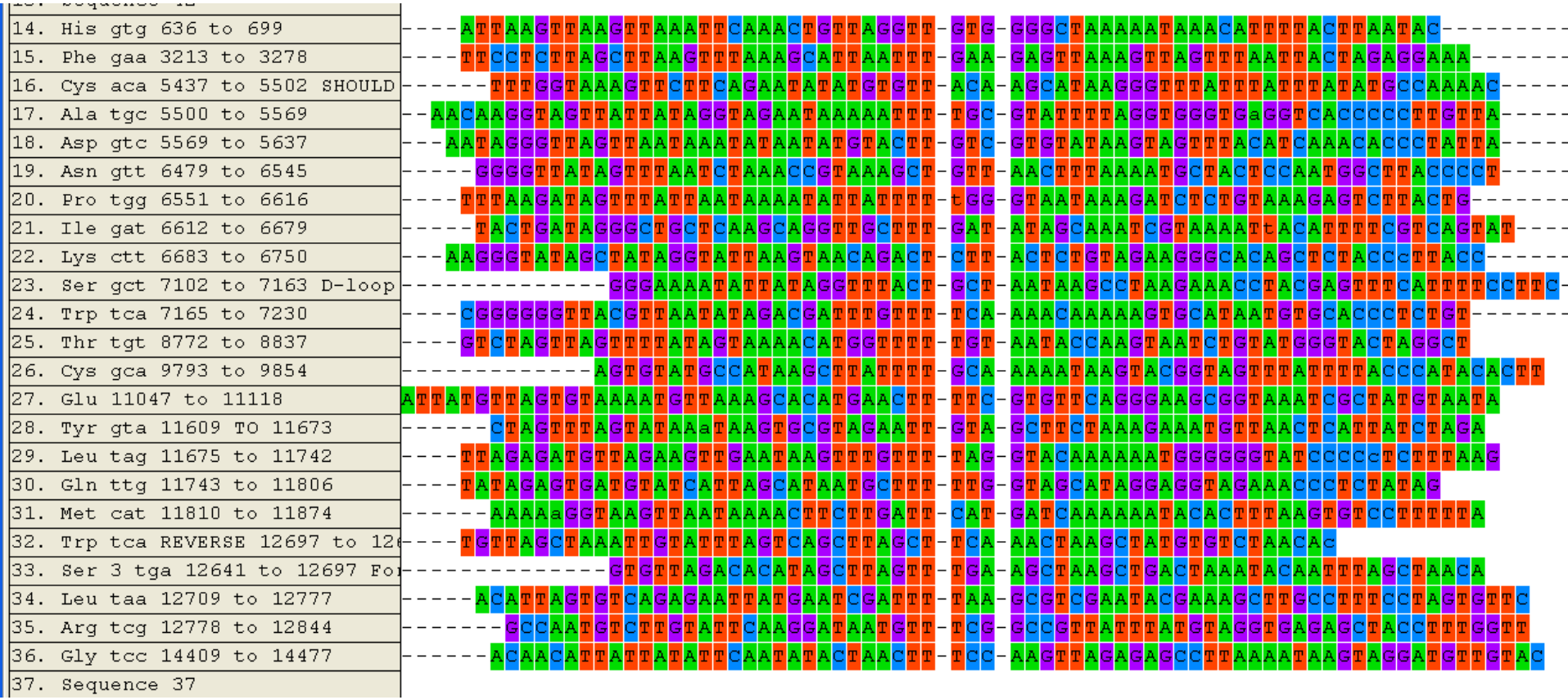
One of the most perplexing aspects of mitochondrial genetics is the marked clinical variability associated with mtDNA mutations. This is dramatically demonstrated for the MTTL1 gene where base substitutions at np 3243, 3252, 3271, and 3291 give MELAS, but the np 3243 mutation can also result in maternally inherited diabetes and deafness. The MTTL1 mutations at np 3303 and 3260 result in hypertrophic cardiomyopathy and myopathy, but the former has a pediatric onset while the latter is adult-onset. While alteration of functions embedded within the gene such as the MTTER for the np 3243 mutation and a muscle specific tRNA processing activity for the np 3302 mutation have been proposed as partial explanations for these phenotypic differences, much of the clinical variability remains unexplained. The mitochondrial transcription terminator binding site is indicated by black dots.



Gyrodactylus salariksen mitokondrion tRNA-geenit.
Täsmälleen samat kuin ihmisellä, ei kuitenkaan ihan identtiset.

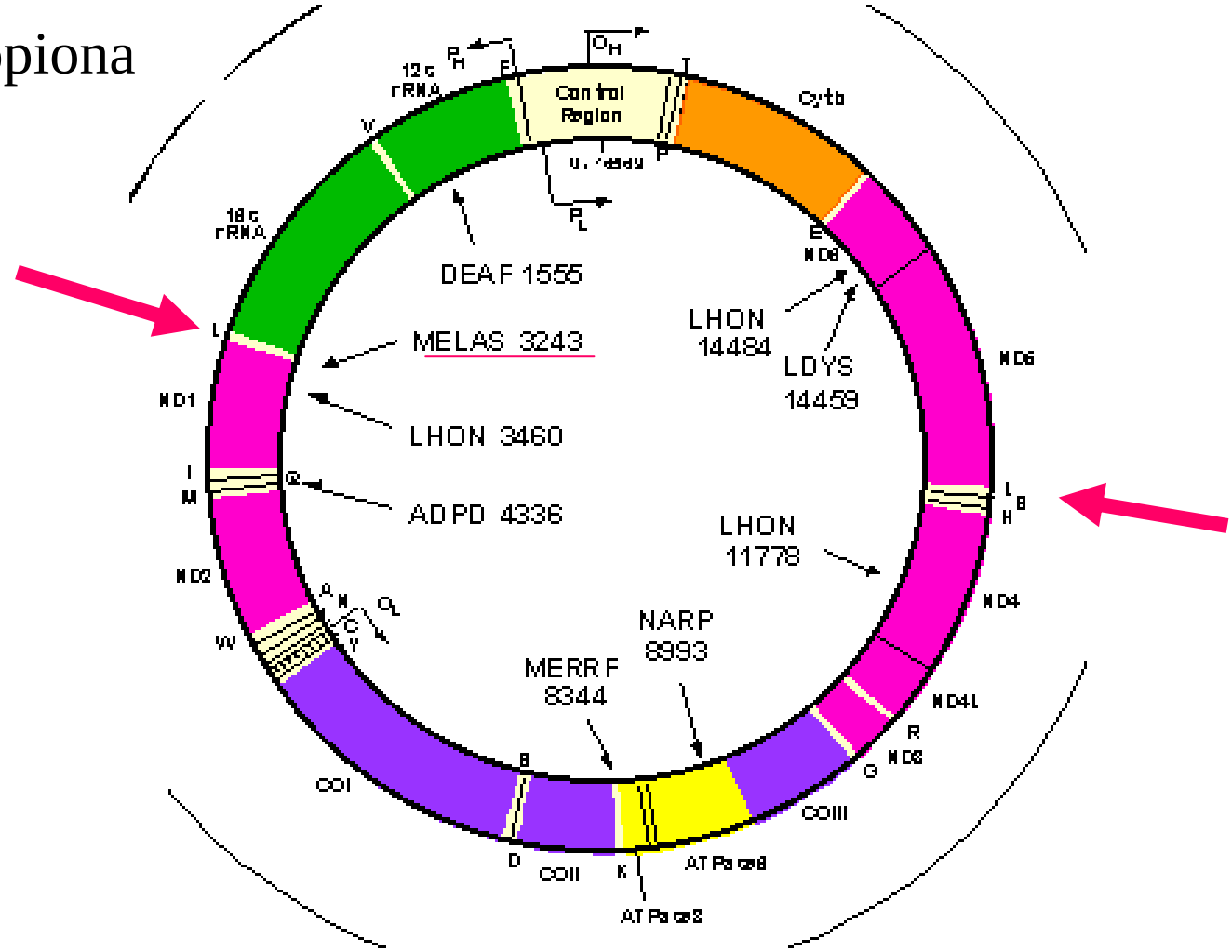
Kahtena ovat seriinin ja leusiinin tRNA:t, ihan niinkuin ihmiselläkin



Kotitehtävä #6: valitse joku tai joitakin ja värkkää sekundaari-struktuurin apilanlehtimalli!

Aloita antikodonista, muista että parit on G=C, A=U(T) ja mahdollinen G=U.

Leusiinin
transferRNA
on kahtena kopiona



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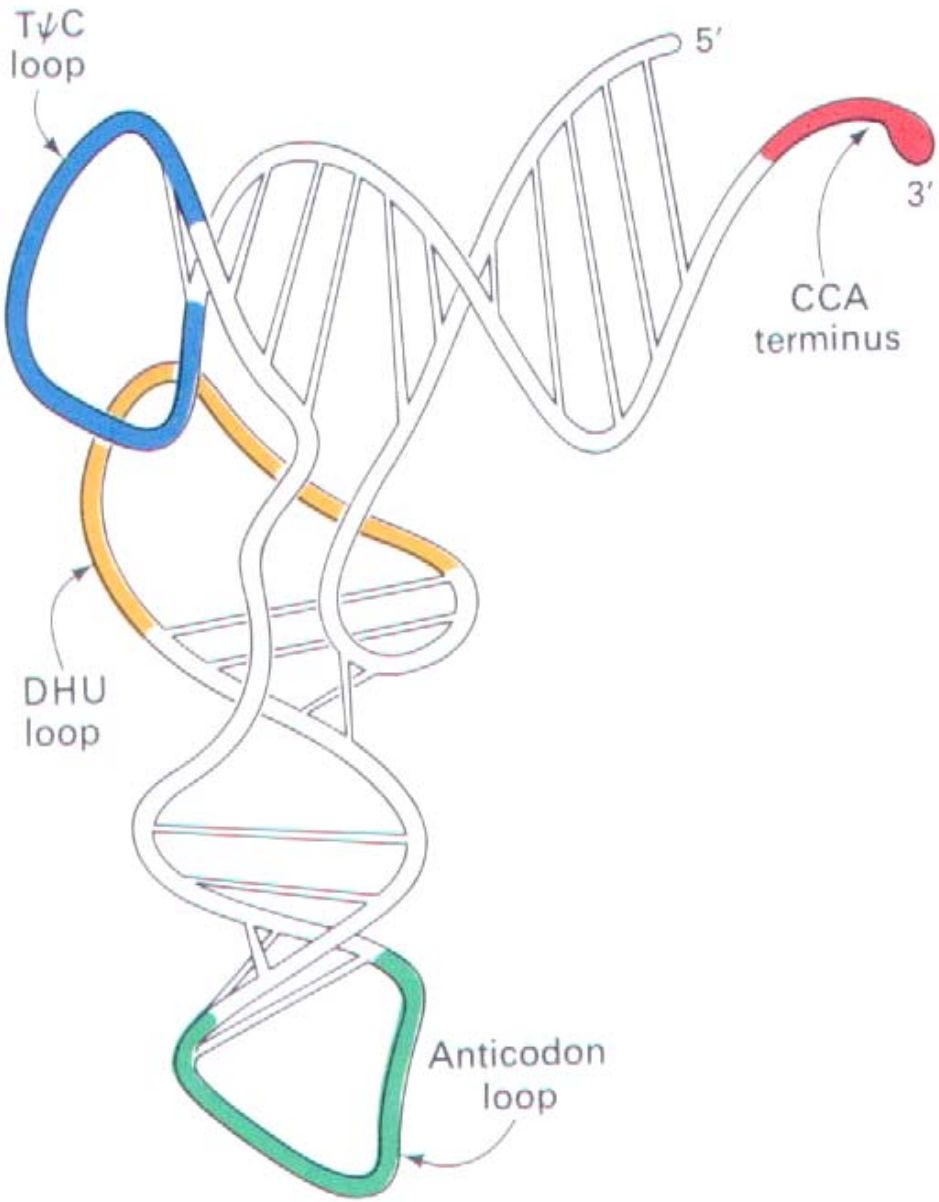

tRNA:n kolmiulotteinen rakenne

Aminohappo sitoutuu CCA-päähän, joka on kaikilla tRNA-laaduilla samanlainen. Miten se aminohappo sitten voi olla oikea????

Otetaan se vielä esille translaatioprosessin yhteydessä: *aminoasyyli-tRNA-syntetaasi*

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