Bioinformatics in practice, week 44+45, 2022

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Globe Institute, KU

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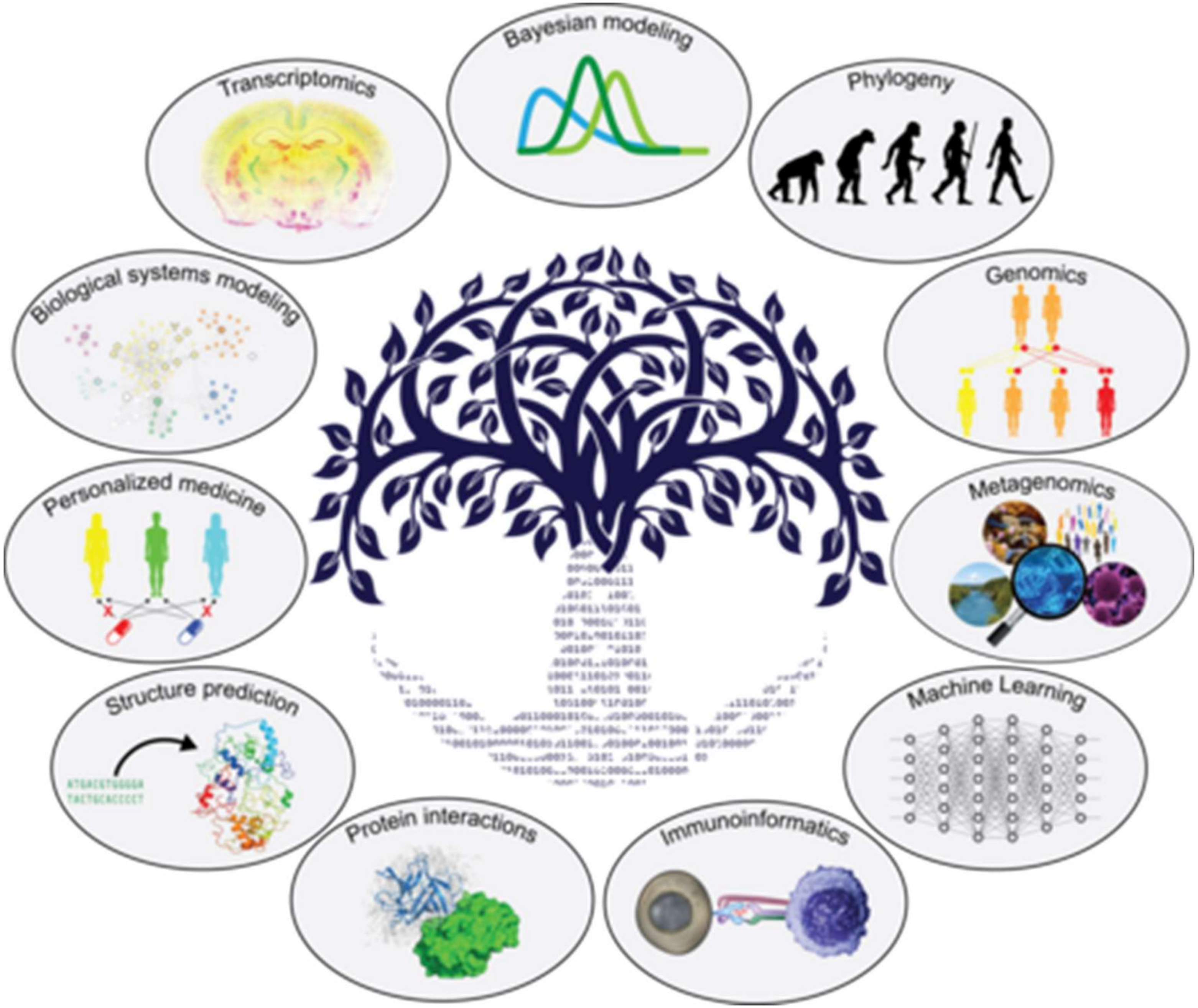
Section for bioinformatics



Section for bioinformatics

Formerly known as:

- **DTU Bioinformatics (own department)**

Formerly formerly known as:

- **Center for Biological Sequence analysis (Department of Systems Biology)**

Center for Biological Sequence analysis (CBS) established 1993

We live in building 204, 2nd floor east (formerly 208)

Week 38 content

Builds on DTU Course
22111: Introduction to Bioinformatics

Course description: https://kurser.dtu.dk/course/22111

Course homepage:
http://teaching.healthtech.dtu.dk/22111

Your week 44+45 homepage:
Go to course homepage → bottom of page →
Bioinformatics in practice, Faroe Islands 2022
(https://teaching.healthtech.dtu.dk/22111/index.php/Bioinformatics_in_practice,_Faroe_Islands_2022)

Week 38 content overview

Data & Databases

- Taxonomy
- DNA
- Proteins
- Protein structure

Methods

- Alignment
  - Pairwise + Multiple
- BLAST (sequence search)
  - DNA / Protein
  - PSI-Blast
- Logos
- Phylogenetic trees
- PyMOL (3D visualization)

Curriculum and structure

Each morning and afternoon:

- Lecture, ~1 hour
  - Copies of slides are linked from the course homepage
- Computer exercise, ~2 hours
  - The exercise guides are the primary curriculum
  - Detailed answers to the exercises are linked from the course homepage. *Don't look at the answers before you have tried to solve the exercise!*

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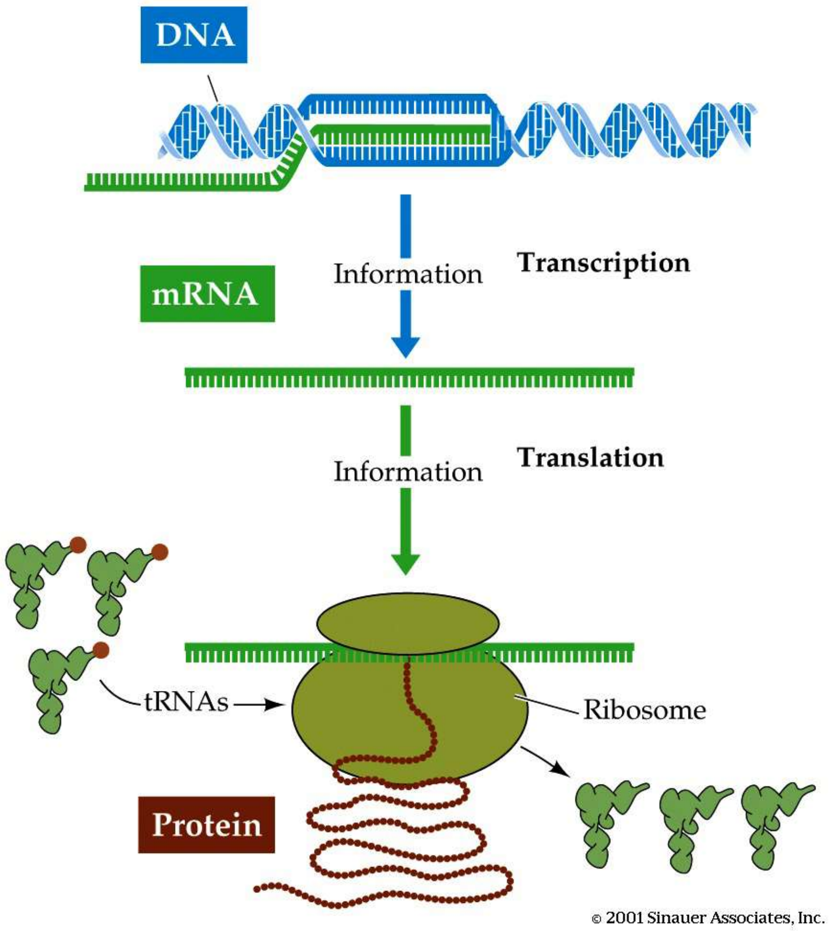
What is bioinformatics?



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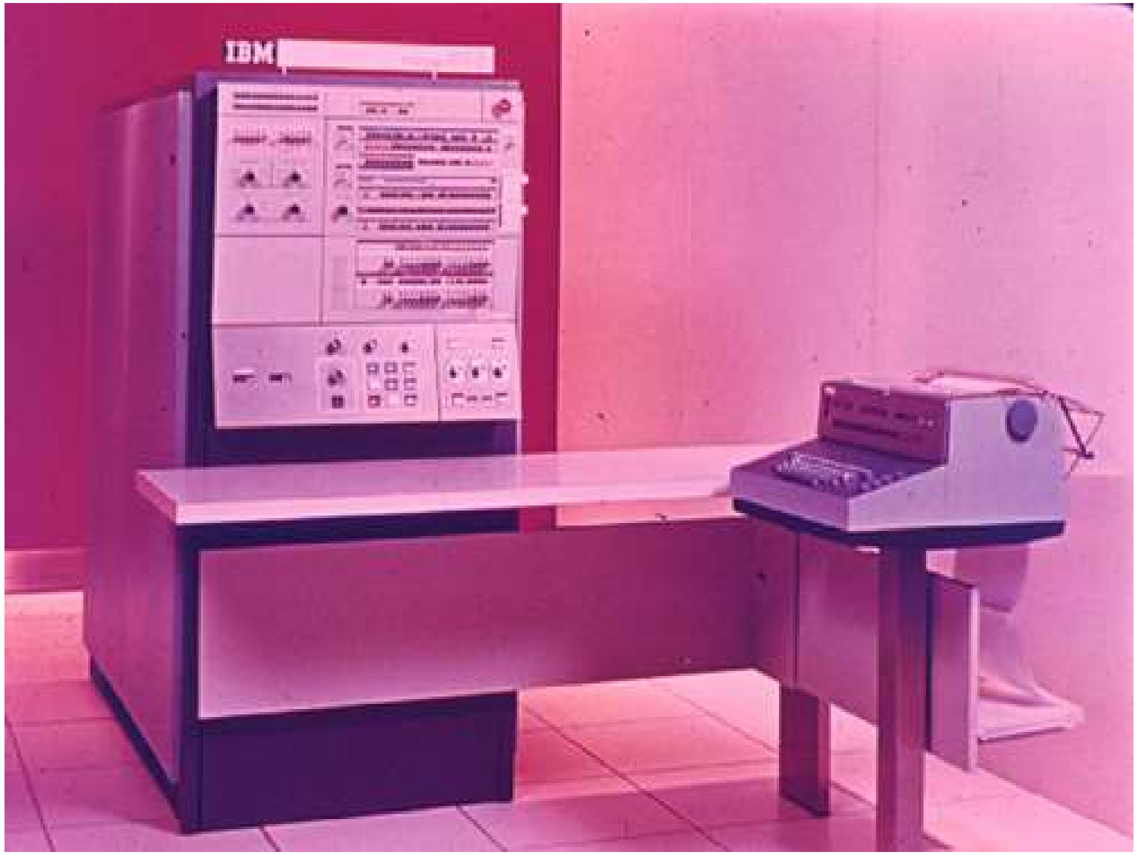

What are bioinformaticians up to, actually?

- *Manage* molecular biological data
  - Store in *databases*, organise, formalise, describe...
- *Compare* molecular biological data
- Find *patterns* in molecular biological data
  - *phylogenies*
  - *correlations* (sequence / structure / expression / function / disease)

Goals:

- *characterise* biological patterns & processes
- *predict* biological properties
  - low level data ⇒ high level properties

    (eg., sequence ⇒ function)

Bioinformatics: neighbour disciplines

- Computational biology
  - *Broader concept: includes computational ecology, physiology, neurology etc...*
- -omics:
  - *Genomics*
  - *Transcriptomics*
  - *Proteomics*
- Systems biology
  - *Putting it all together...*
  - *Building models, identify control & regulation*

Bioinformatics: prerequisites

- **Bio- side:**
  - Molecular biology
  - Cell biology
  - Genetics
  - Evolutionary theory
- **-informatics side:**
  - Computer science
  - Statistics
  - Theoretical physics

Molecular biology data...

- **DNA sequences**

```
>alpha-D
ATGCTGACCGACTCTGACAAGAAGCTGGTCCTGCAGGTGTGGGAGAAGGTGATCCGCCAC
CCAGACTGTGGAGCCGAGGCCCTGGAGAGGTGCGGGCTGAGCTTGGGGAAACCATGGGCA
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CACTGACCATCCCGCTCCCGCAGCTGTTCACCACCTACCCCCAGACCAAGACCTACTTCC
CCCACTTCGACTTGCACCATGGCTCCGACCAGGTCCGCAACCACGGCAAGAAGGTGTTGG
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GCGACCTGCATGCCTACAACCTGCGTGTCGACCCTGTCAACTTCAAGGCAGGCGGGGGAC
GGGGGTCAGGGGCCGGGGAGTTGGGGGCCAGGGACCTGGTTGGGGATCCGGGGCCATGCC
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TCCCTGTTCTATGTGGCCCTGGCTCTGTCTCATCTGTCCCCAACTGTCCCTGATTGCCTC
TGTCCCCCAGGTTGTTCATCACCTACCCCCAGACCAAGACCTACTTCCCCCACTTCGACC
TGTCACATGGCTCCGCTCAGATCAAGGGGCACGGCAAGAAGGTGGCGGAGGCACTGGTTG
AGGCTGCCAACCACATCGATGACATCGCTGGTGCCCTCTCCAAGCTGAGCGACCTCCACG
CCCAAAAGCTCCGTGTGGACCCCGTCAACTTCAAAGTGAGCATCTGGGAAGGGGTGACCA
GTCTGGCTCCCCTCCTGCACACACCTCTGGCTACCCCCTCACCTCACCCCCCTTGCTCACC
ATCTCCTTTTGCCTTTCAGCTGCTGGGTCACTGCTTCCTGGTGGTCGTGGCCGTCCACTT
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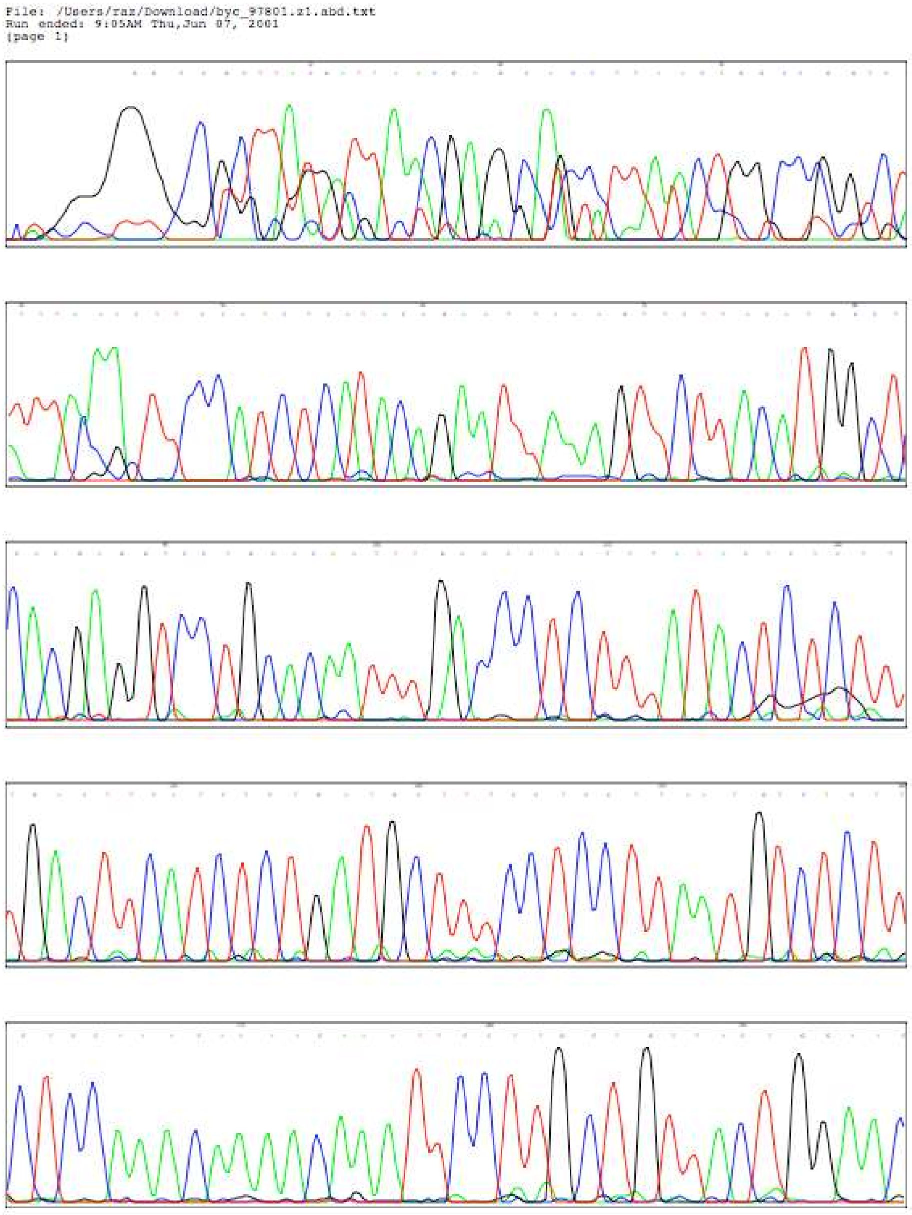

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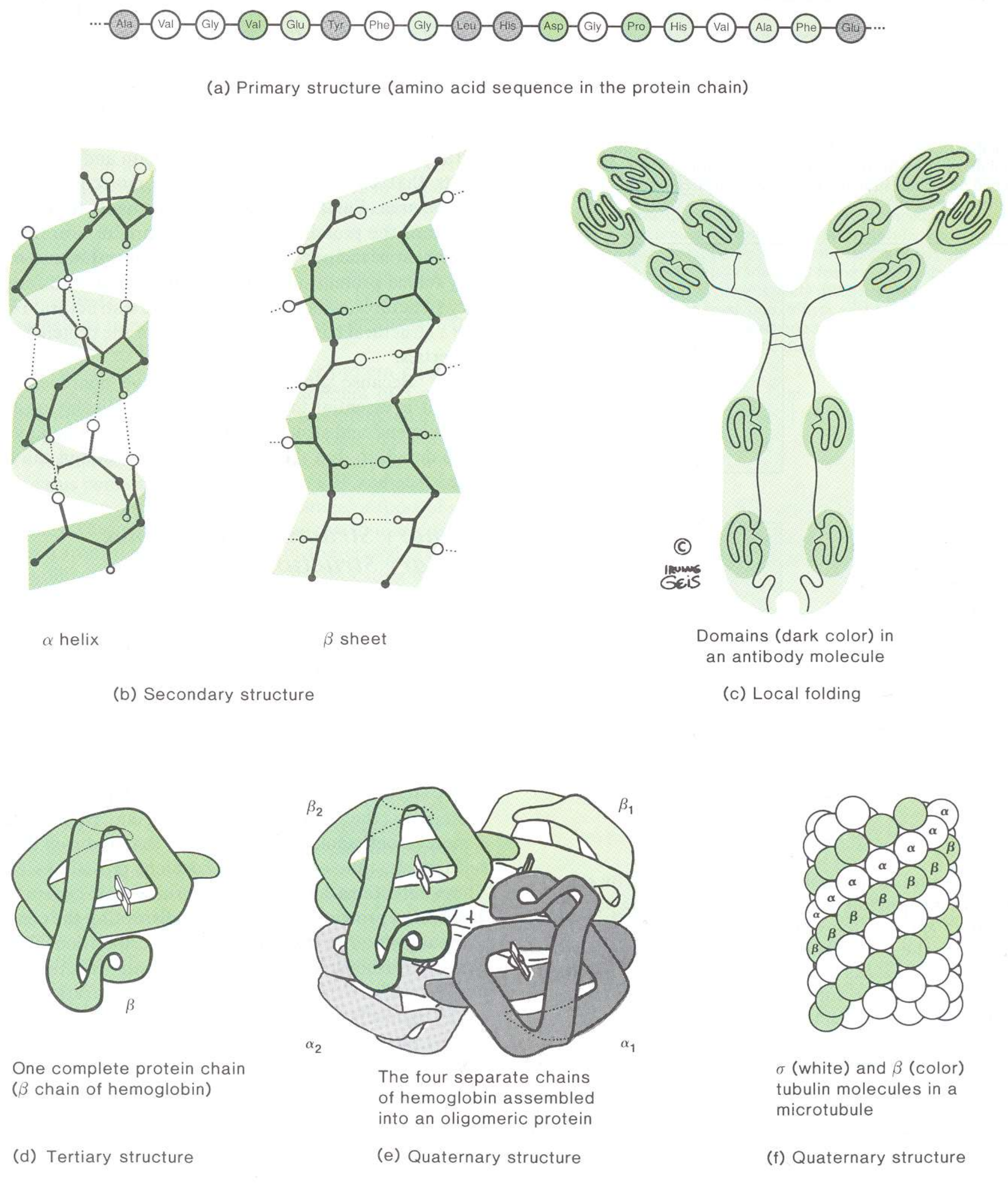
Molecular biology data...

- Amino acid sequences
- Protein structure:
  - X-ray crystallography
  - NMR



(a) Primary structure (amino acid sequence in the protein chain)

α helix

β sheet

(b) Secondary structure

Domains (dark color) in an antibody molecule

(c) Local folding

One complete protein chain (β chain of hemoglobin)

(d) Tertiary structure

The four separate chains of hemoglobin assembled into an oligomeric protein

(e) Quaternary structure

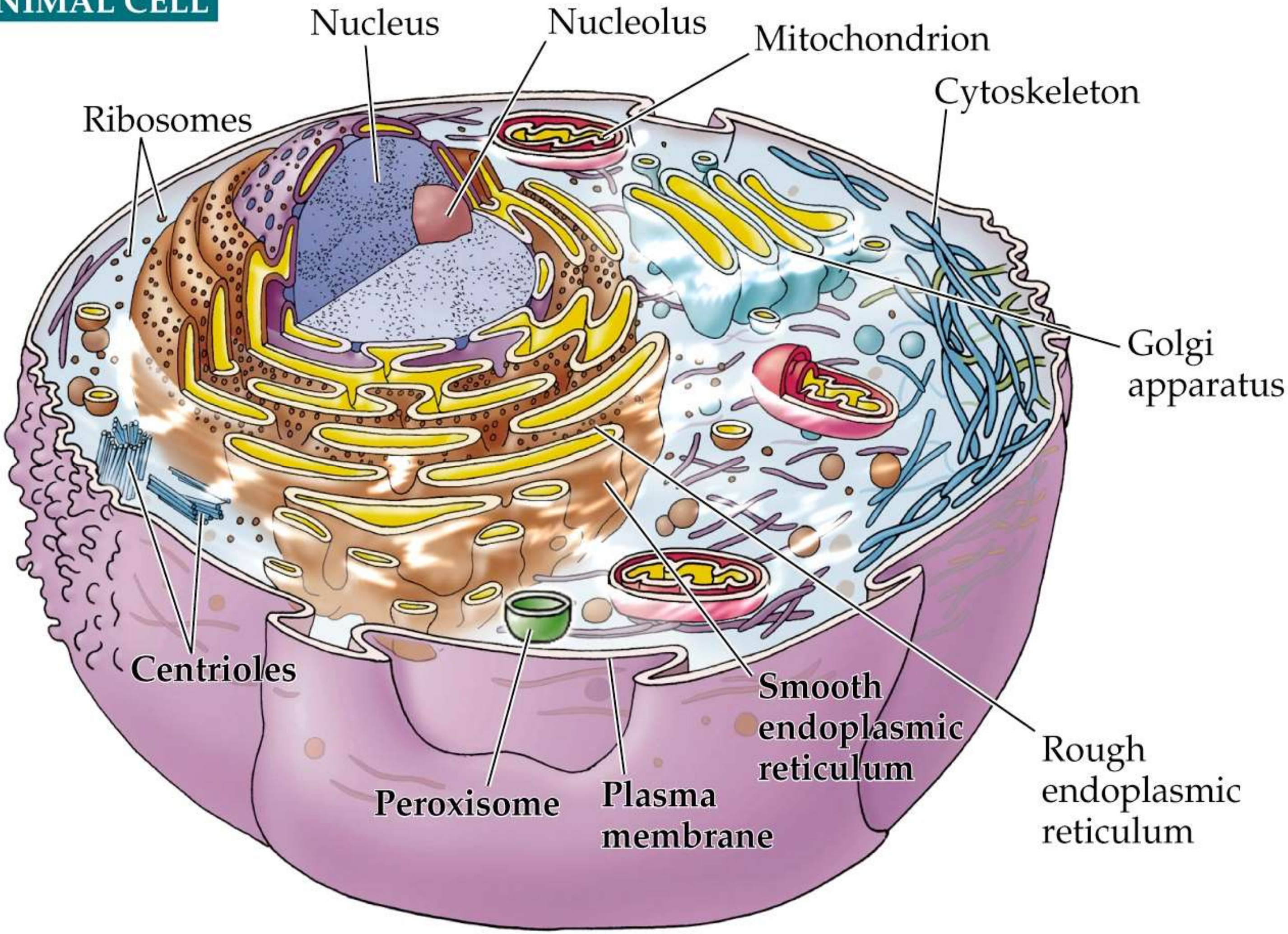
σ (white) and β (color) tubulin molecules in a microtubule

(f) Quaternary structure

Cell biology & proteomics data...

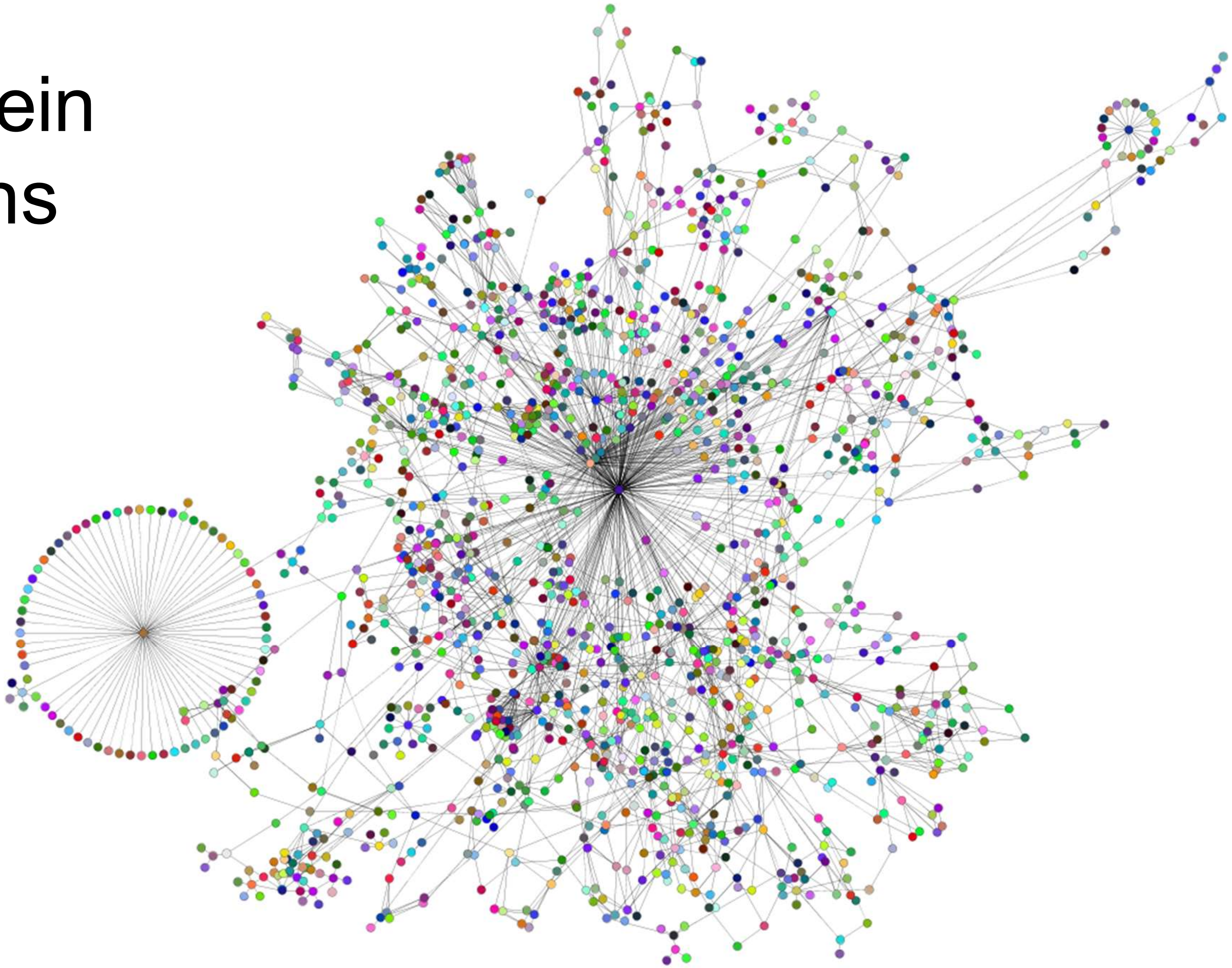
- Subcellular localization

AN ANIMAL CELL

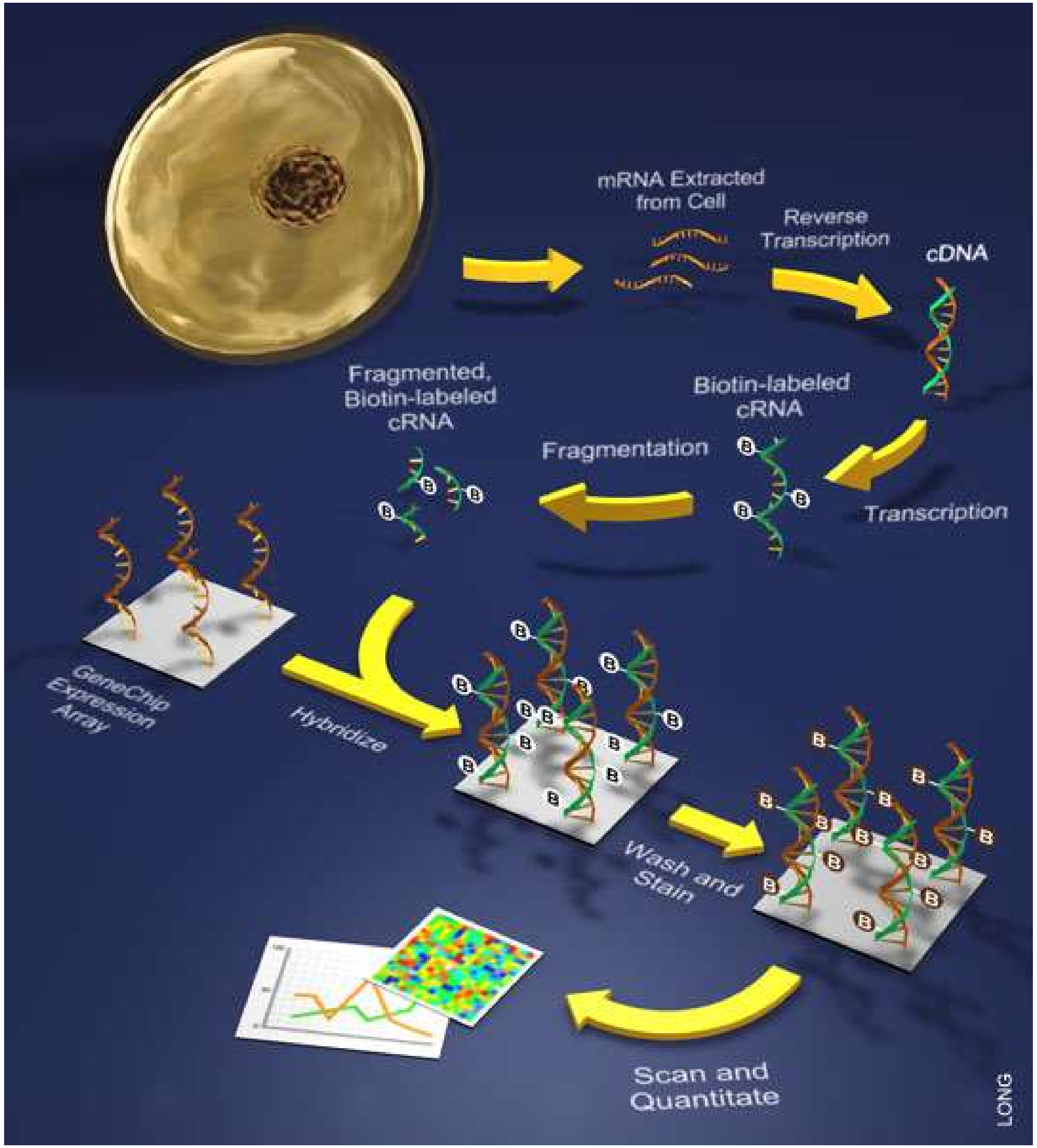


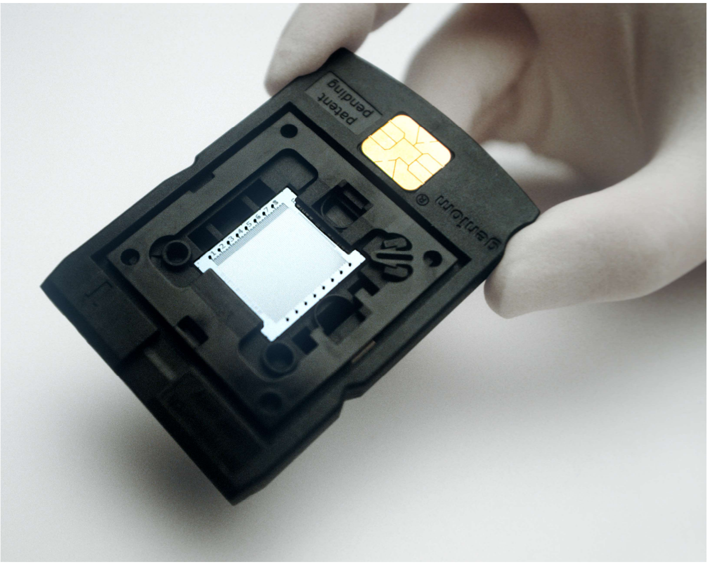
Cell biology & proteomics data...

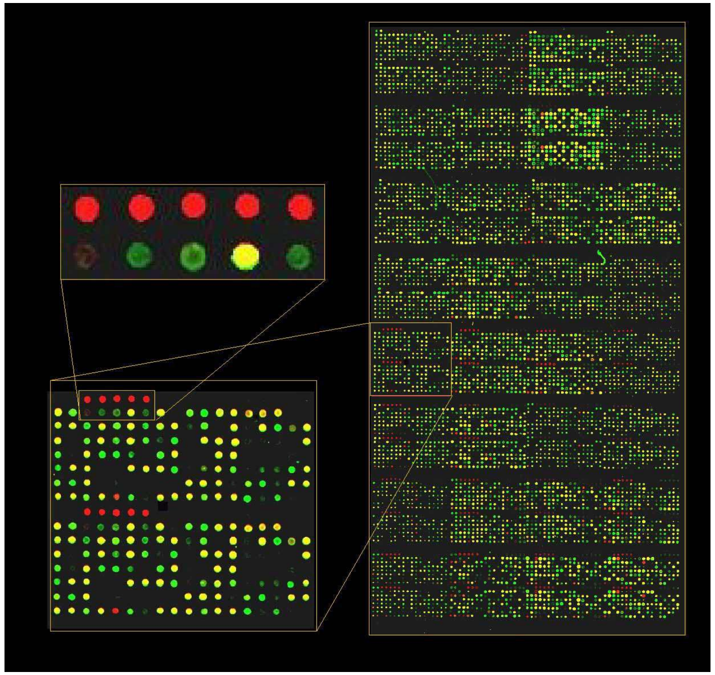
protein-protein
interactions



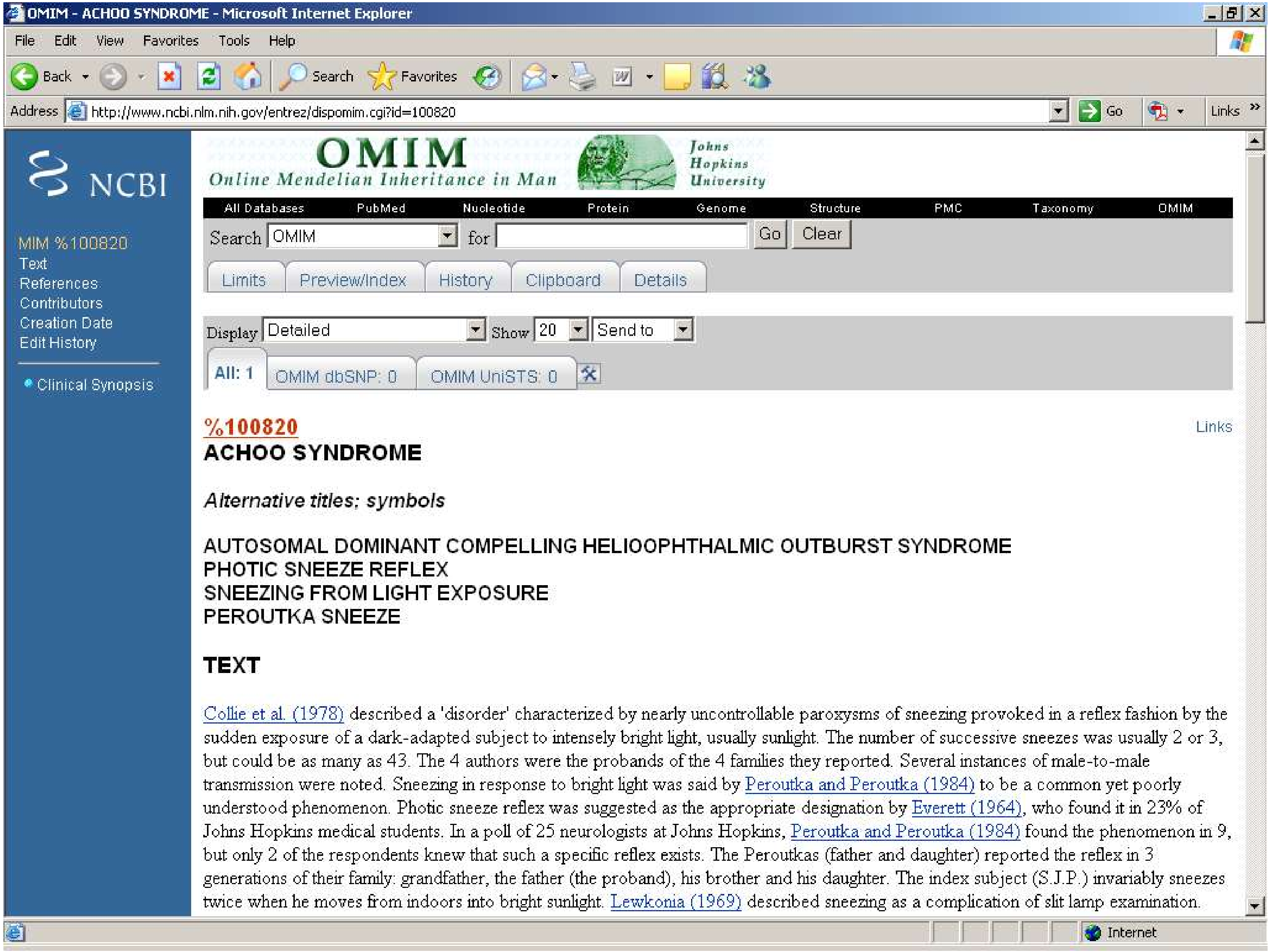
Transcriptomics: DNA microarray technology







Phenotype data: human diseases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ACHOO SYNDROME

Alternative titles; symbols

AUTOSOMAL DOMINANT COMPELLING HELIOOPHTHALMIC OUTBURST SYNDROME
PHOTIC SNEEZE REFLEX
SNEEZING FROM LIGHT EXPOSURE
PEROUTKA SNEEZE

TEXT

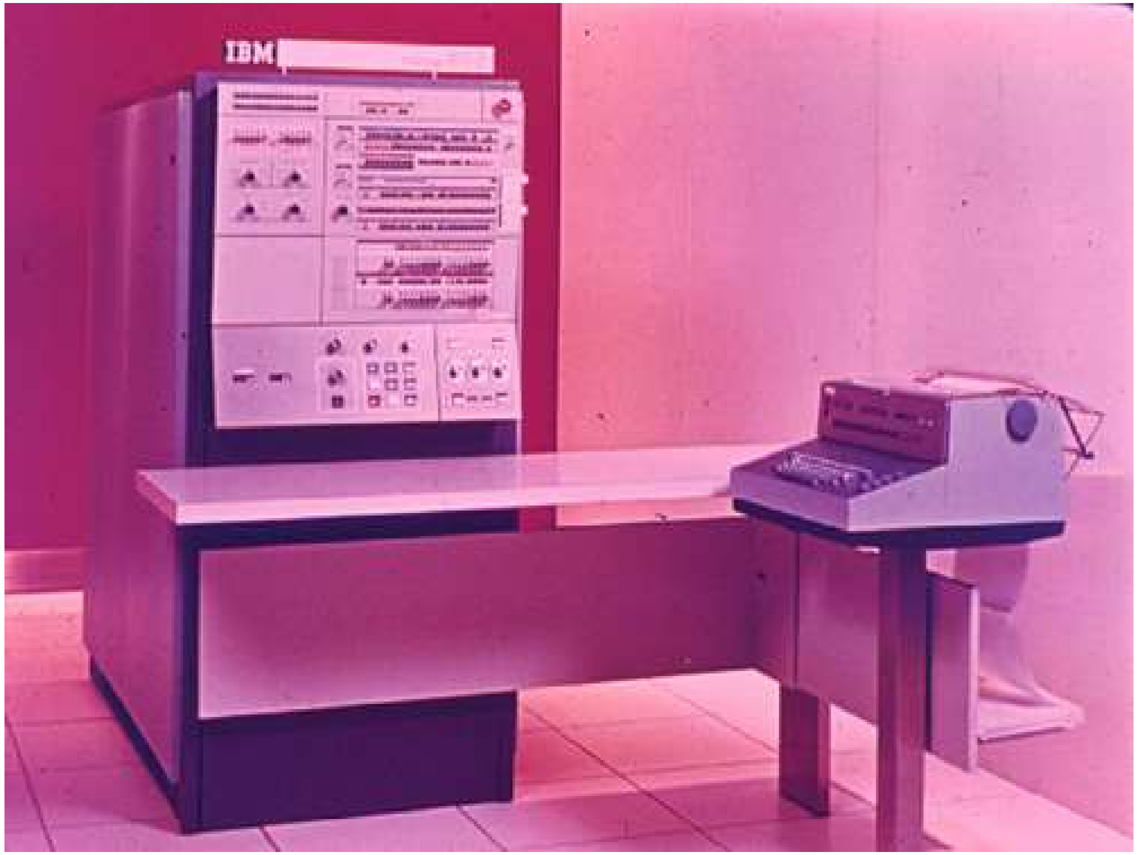
Collie et al. (1978) described a 'disorder' characterized by nearly uncontrollable paroxysms of sneezing provoked in a reflex fashion by the sudden exposure of a dark-adapted subject to intensely bright light, usually sunlight. The number of successive sneezes was usually 2 or 3, but could be as many as 43. The 4 authors were the probands of the 4 families they reported. Several instances of male-to-male transmission were noted. Sneezing in response to bright light was said by Peroutka and Peroutka (1984) to be a common yet poorly understood phenomenon. Photic sneeze reflex was suggested as the appropriate designation by Everett (1964), who found it in 23% of Johns Hopkins medical students. In a poll of 25 neurologists at Johns Hopkins, Peroutka and Peroutka (1984) found the phenomenon in 9, but only 2 of the respondents knew that such a specific reflex exists. The Peroutkas (father and daughter) reported the reflex in 3 generations of their family: grandfather, the father (the proband), his brother and his daughter. The index subject (S.J.P.) invariably sneezes twice when he moves from indoors into bright sunlight. Lewkonia (1969) described sneezing as a complication of slit lamp examination.

Prediction methods

- Homology / Alignment
- Simple pattern (“word”) recognition
- Statistical methods
  - Weight matrices: calculate amino acid *probabilities*
  - *Other examples:* Regression, variance analysis, clustering
- Machine learning
  - Like statistical methods, but parameters are estimated by iterative *training* rather than direct calculation
  - *Examples:* Neural Networks (**NN**), Hidden Markov Models (**HMM**), Support Vector Machines (**SVM**)
- Combinations

The computer

- *Everything* can be reduced to bits (0 or 1)



https://en.wikipedia.org/wiki/IBM_System/360

Digital information

- A byte = 8 bits

0 1 0 0 0 0 0 1

Can be interpreted as

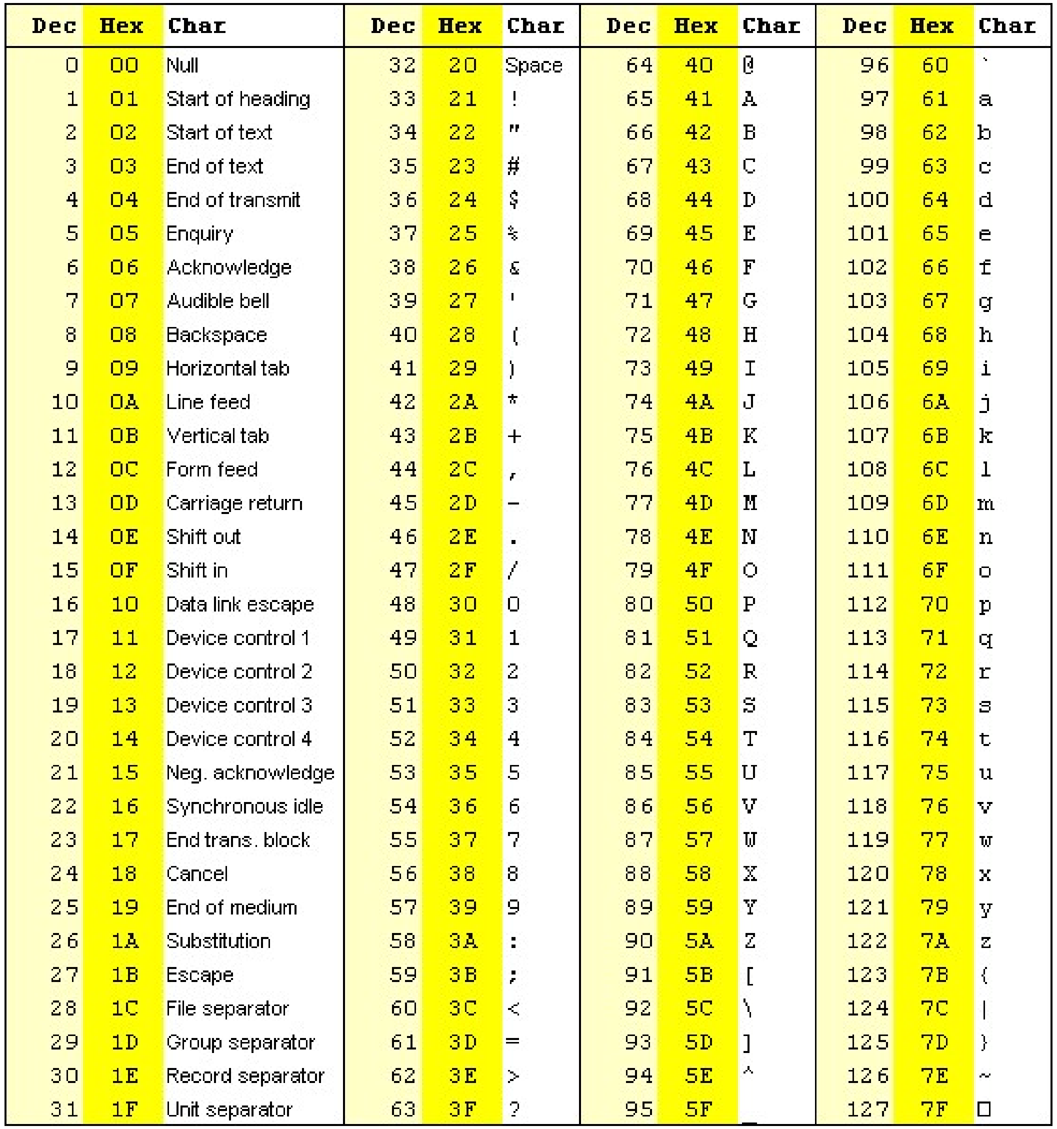
- The number 65
- The letter ”A”
- Part of a machine code instruction
- Part of a colour specification
- Part of a sound encoding
- …

Text files

A text file is a file where every byte is interpreted as a character

Examples

Plain text	.txt
Program settings	.ini
C source code	.c
Python script	.py
$\mathrm{T_EX}$ source	.tex
Web page source	.html
Sequences	.fasta

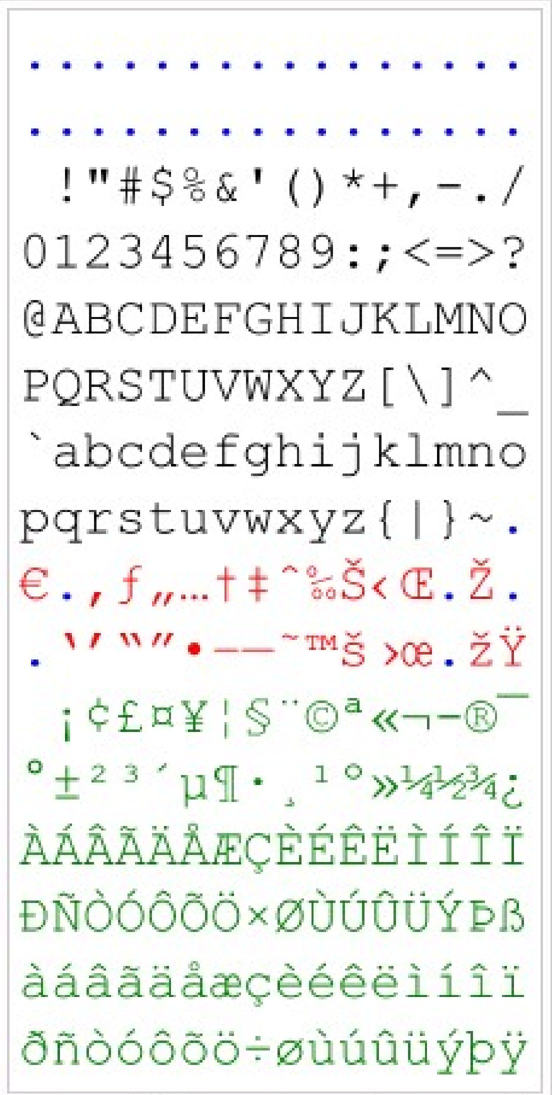


Dec	Hex	Char	Dec	Hex	Char	Dec	Hex	Char	Dec	Hex	Char
0	00	Null	32	20	Space	64	40	@	96	60	`
1	01	Start of heading	33	21	!	65	41	A	97	61	a
2	02	Start of text	34	22	"	66	42	B	98	62	b
3	03	End of text	35	23	#	67	43	C	99	63	c
4	04	End of transmit	36	24	$	68	44	D	100	64	d
5	05	Enquiry	37	25	%	69	45	E	101	65	e
6	06	Acknowledge	38	26	&	70	46	F	102	66	f
7	07	Audible bell	39	27	'	71	47	G	103	67	g
8	08	Backspace	40	28	(	72	48	H	104	68	h
9	09	Horizontal tab	41	29	)	73	49	I	105	69	i
10	0A	Line feed	42	2A	*	74	4A	J	106	6A	j
11	0B	Vertical tab	43	2B	+	75	4B	K	107	6B	k
12	0C	Form feed	44	2C	,	76	4C	L	108	6C	l
13	0D	Carriage return	45	2D	-	77	4D	M	109	6D	m
14	0E	Shift out	46	2E	.	78	4E	N	110	6E	n
15	0F	Shift in	47	2F	/	79	4F	O	111	6F	o
16	10	Data link escape	48	30	0	80	50	P	112	70	p
17	11	Device control 1	49	31	1	81	51	Q	113	71	q
18	12	Device control 2	50	32	2	82	52	R	114	72	r
19	13	Device control 3	51	33	3	83	53	S	115	73	s
20	14	Device control 4	52	34	4	84	54	T	116	74	t
21	15	Neg. acknowledge	53	35	5	85	55	U	117	75	u
22	16	Synchronous idle	54	36	6	86	56	V	118	76	v
23	17	End trans. block	55	37	7	87	57	W	119	77	w
24	18	Cancel	56	38	8	88	58	X	120	78	x
25	19	End of medium	57	39	9	89	59	Y	121	79	y
26	1A	Substitution	58	3A	:	90	5A	Z	122	7A	z
27	1B	Escape	59	3B	;	91	5B	[	123	7B	{
28	1C	File separator	60	3C	<	92	5C	\	124	7C	|
29	1D	Group separator	61	3D	=	93	5D	]	125	7D	}
30	1E	Record separator	62	3E	>	94	5E	^	126	7E	~
31	1F	Unit separator	63	3F	?	95	5F	_	127	7F	□

The ASCII table

Extended character sets

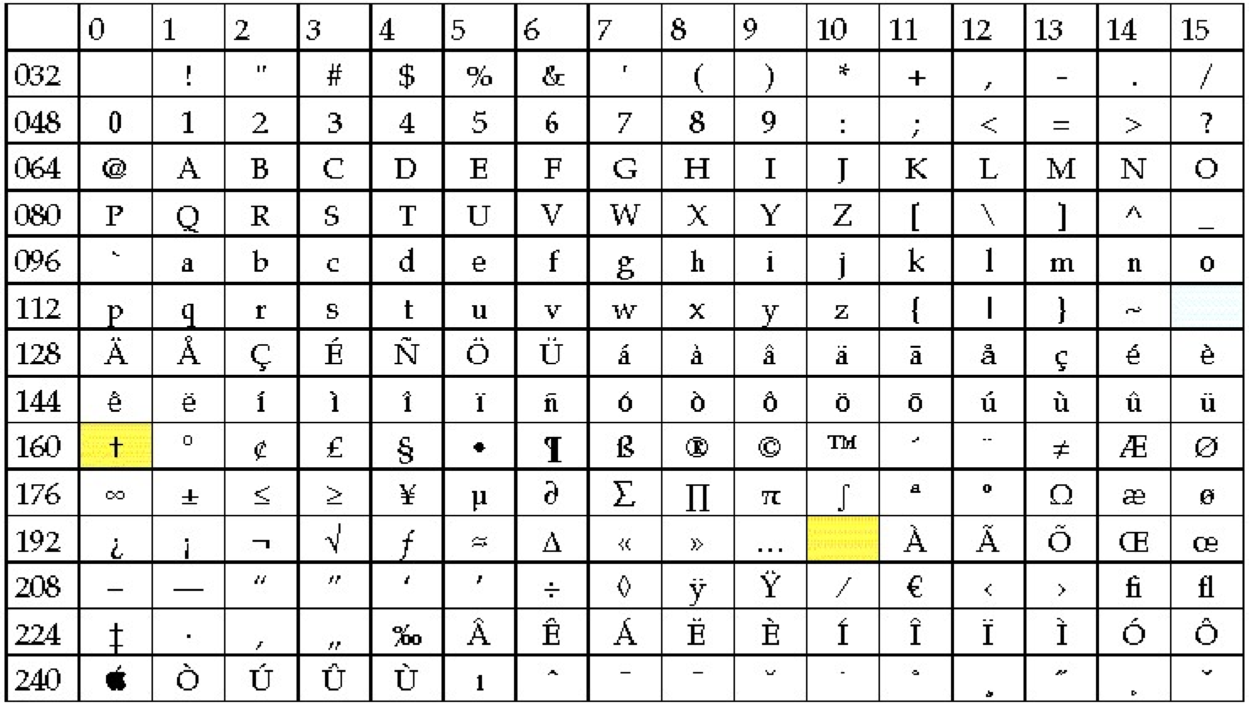
The are *many* ways to interpret characters with values above 127. Here, you see two of them.



```
................
................
 !"#$%&'()*+,-./
0123456789:;<=>?
@ABCDEFGHIJKLMNO
PQRSTUVWXYZ[\]^_
`abcdefghijklmno
pqrstuvwxyz{|}~.
€.‚ƒ„…†‡ˆ‰Š‹Œ.Ž.
.‘’“”•–—˜™š›œ.žŸ
 ¡¢£¤¥¦§¨©ª«¬-®¯
°±²³´µ¶·¸¹º»¼½¾¿
ÀÁÂÃÄÅÆÇÈÉÊËÌÍÎÏ
ÐÑÒÓÔÕÖ×ØÙÚÛÜÝÞß
àáâãäåæçèéêëìíîï
ðñòóôõö÷øùúûüýþÿ
```

Windows-1252, sometimes called incorrectly "ANSI". Blue dots indicate unused or control characters

"Mac OS Roman" Encoding:



	0	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15
032		!	"	#	$	%	&	'	(	)	*	+	,	-	.	/
048	0	1	2	3	4	5	6	7	8	9	:	;	<	=	>	?
064	@	A	B	C	D	E	F	G	H	I	J	K	L	M	N	O
080	P	Q	R	S	T	U	V	W	X	Y	Z	[	\	]	^	_
096	`	a	b	c	d	e	f	g	h	i	j	k	l	m	n	o
112	p	q	r	s	t	u	v	w	x	y	z	{	|	}	~	
128	Ä	Å	Ç	É	Ñ	Ö	Ü	á	à	â	ä	ã	å	ç	é	è
144	ê	ë	í	ì	î	ï	ñ	ó	ò	ô	ö	õ	ú	ù	û	ü
160	†	°	¢	£	§	•	¶	ß	®	©	™	´	¨	≠	Æ	Ø
176	∞	±	≤	≥	¥	µ	∂	Σ	∏	π	∫	ª	º	Ω	æ	ø
192	¿	¡	¬	√	ƒ	≈	Δ	«	»	…		À	Ã	Õ	Œ	œ
208	–	—	“	”	‘	’	÷	◊	ÿ	Ÿ	⁄	€	‹	›	ﬁ	ﬂ
224	‡	·	‚	„	‰	Â	Ê	Á	Ë	È	Í	Î	Ï	Ì	Ó	Ô
240		Ò	Ú	Û	Ù	ı	ˆ	˜	¯	˘	˙	˚	¸	˝	˛	ˇ

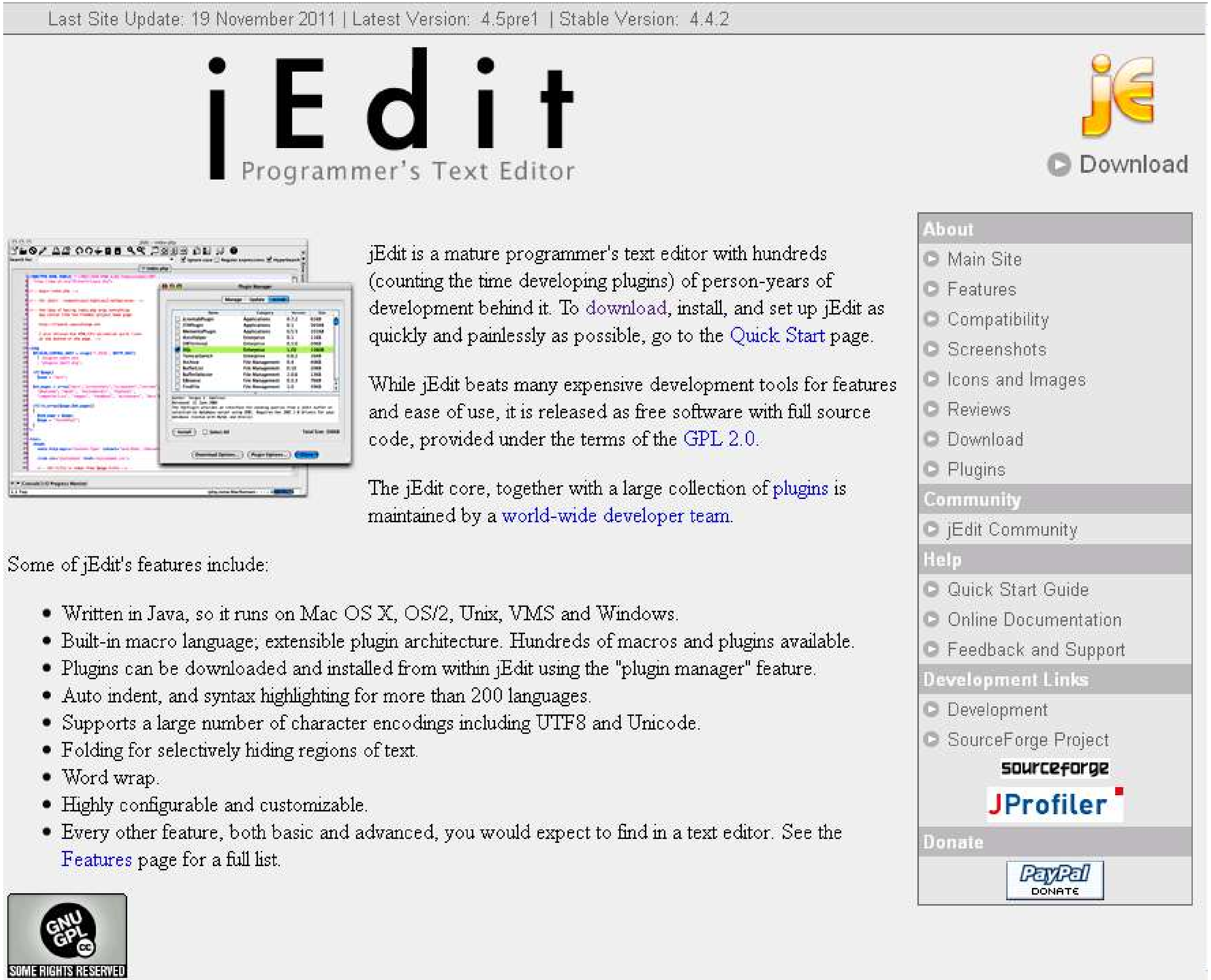
Text files—line endings

- UNIX standard (including Mac OS X):
  - 10 — LF ("Line feed" char).
- Old Mac (System 9 and before):
  - 13 — CR ("Carriage Return" char).
- DOS/Windows:
  - 13, 10 — both CR and LF.

A good text editor can handle all three systems.

jEdit

www.jedit.org



jEdit is a mature programmer's text editor with hundreds (counting the time developing plugins) of person-years of development behind it. To download, install, and set up jEdit as quickly and painlessly as possible, go to the Quick Start page.

While jEdit beats many expensive development tools for features and ease of use, it is released as free software with full source code, provided under the terms of the GPL 2.0.

The jEdit core, together with a large collection of plugins is maintained by a world-wide developer team.

Some of jEdit's features include:

- Written in Java, so it runs on Mac OS X, OS/2, Unix, VMS and Windows.
- Built-in macro language; extensible plugin architecture. Hundreds of macros and plugins available.
- Plugins can be downloaded and installed from within jEdit using the "plugin manager" feature.
- Auto indent, and syntax highlighting for more than 200 languages.
- Supports a large number of character encodings including UTF8 and Unicode.
- Folding for selectively hiding regions of text.
- Word wrap.
- Highly configurable and customizable.
- Every other feature, both basic and advanced, you would expect to find in a text editor. See the Features page for a full list.