BioJulia

Design and scope

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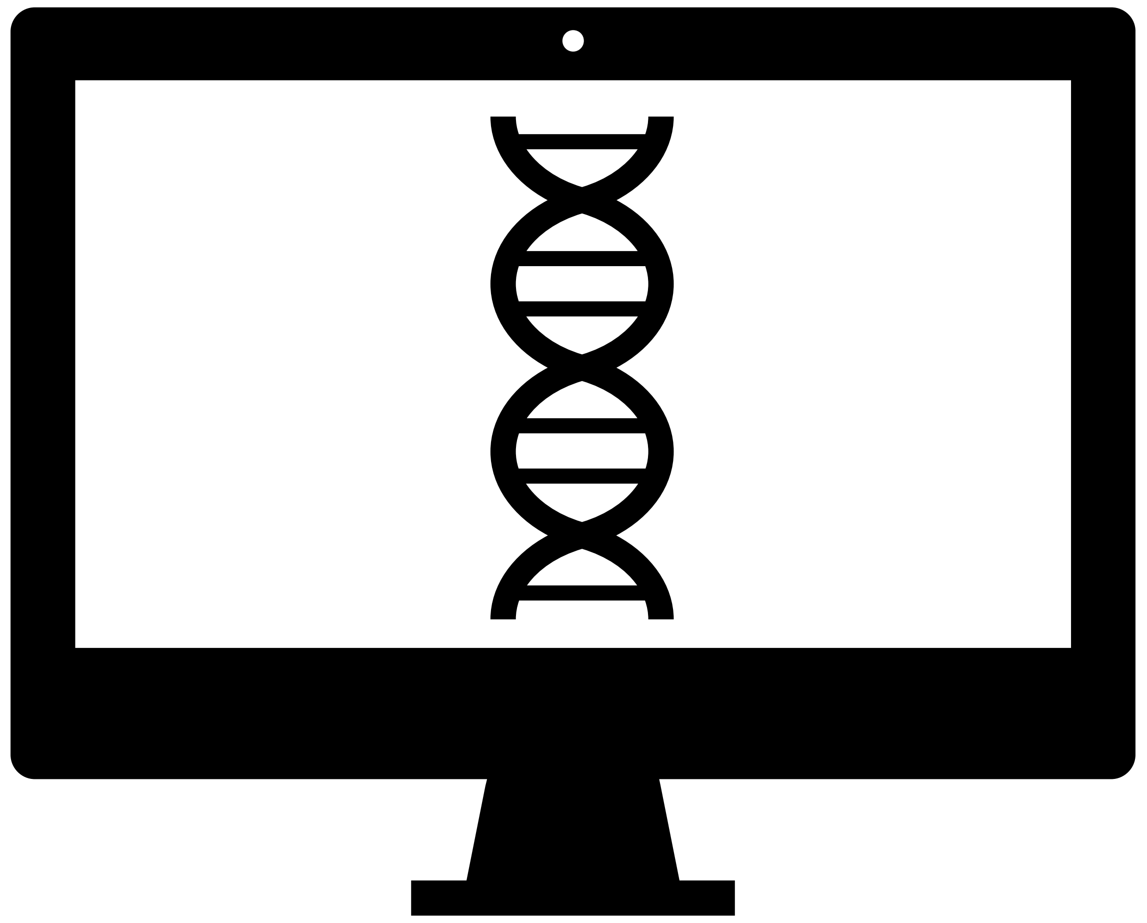
Overview

- What do bioinformaticians do?
- What would the ideal bioinformatics framework look like?
- The values of BioJulia
- BioSequences

Bioinformatics as a discipline

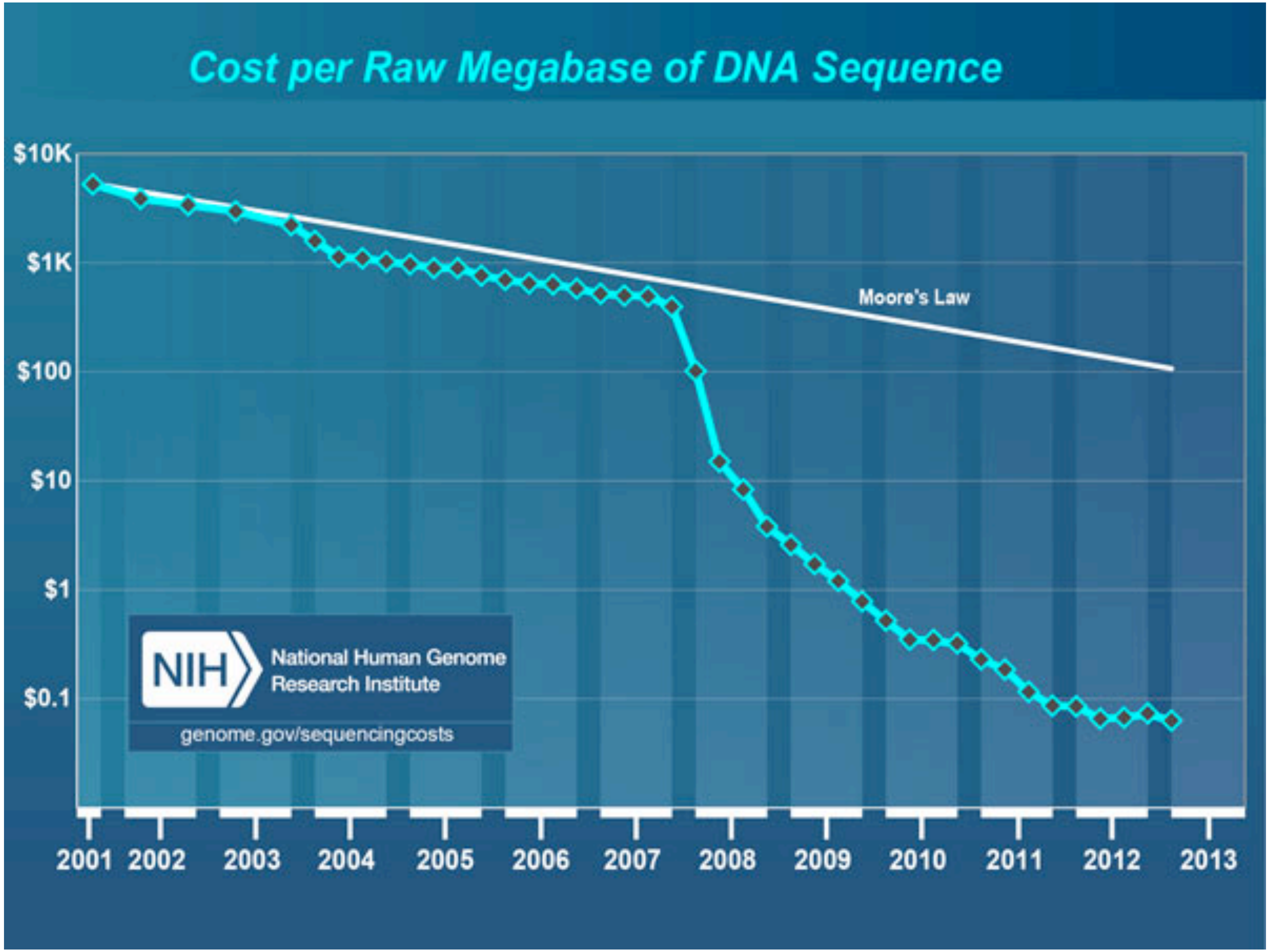


Field that develops methods and software tools for understanding biological data

Field that use and develop software for analysing biological sequences and molecules as digital data

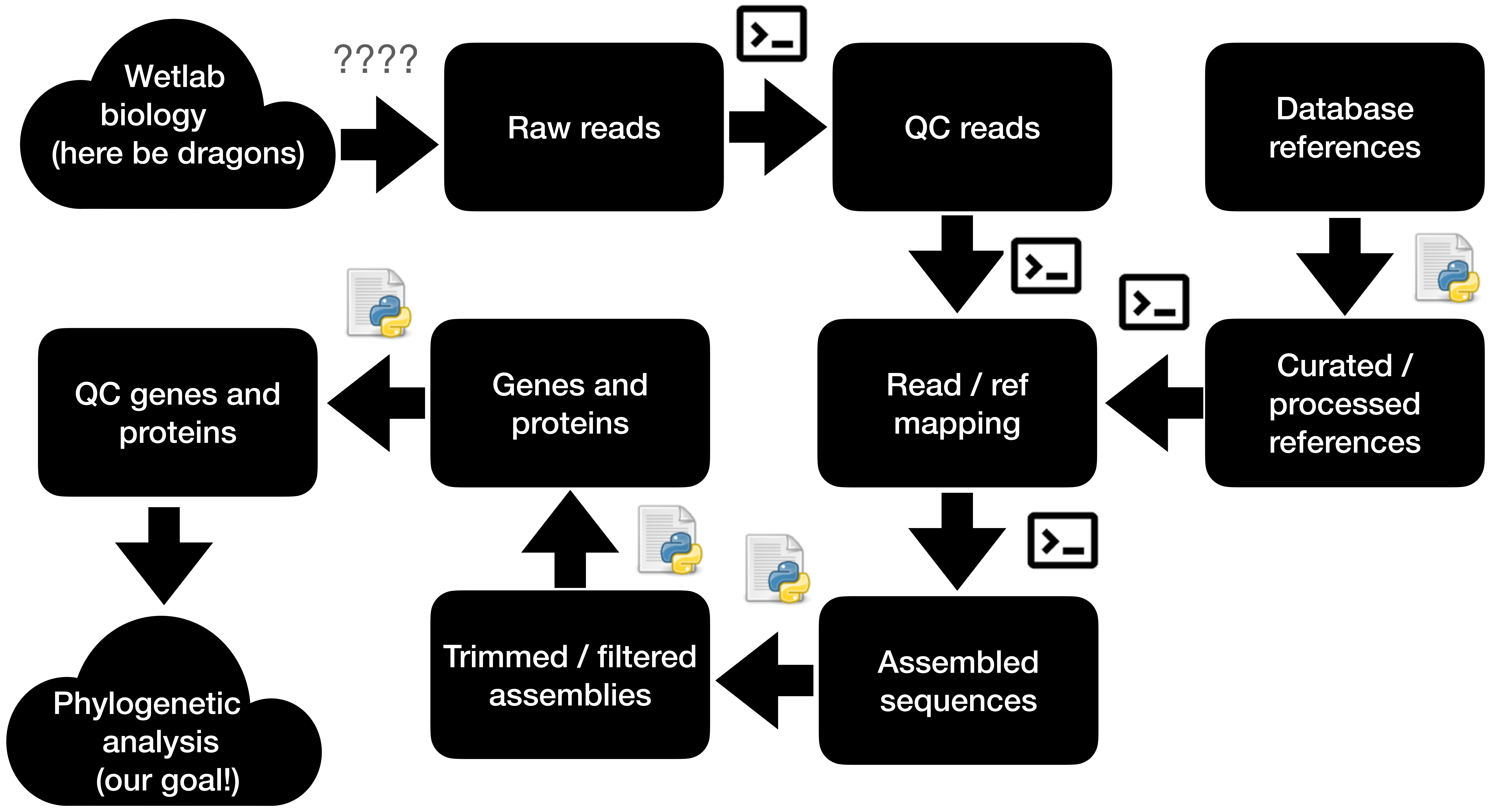
- Biological sequences *are* digital! No, really!
- This makes bioinformatics an "easy" field in some ways compared to most computational biology

The most abused graph in bioinformatics

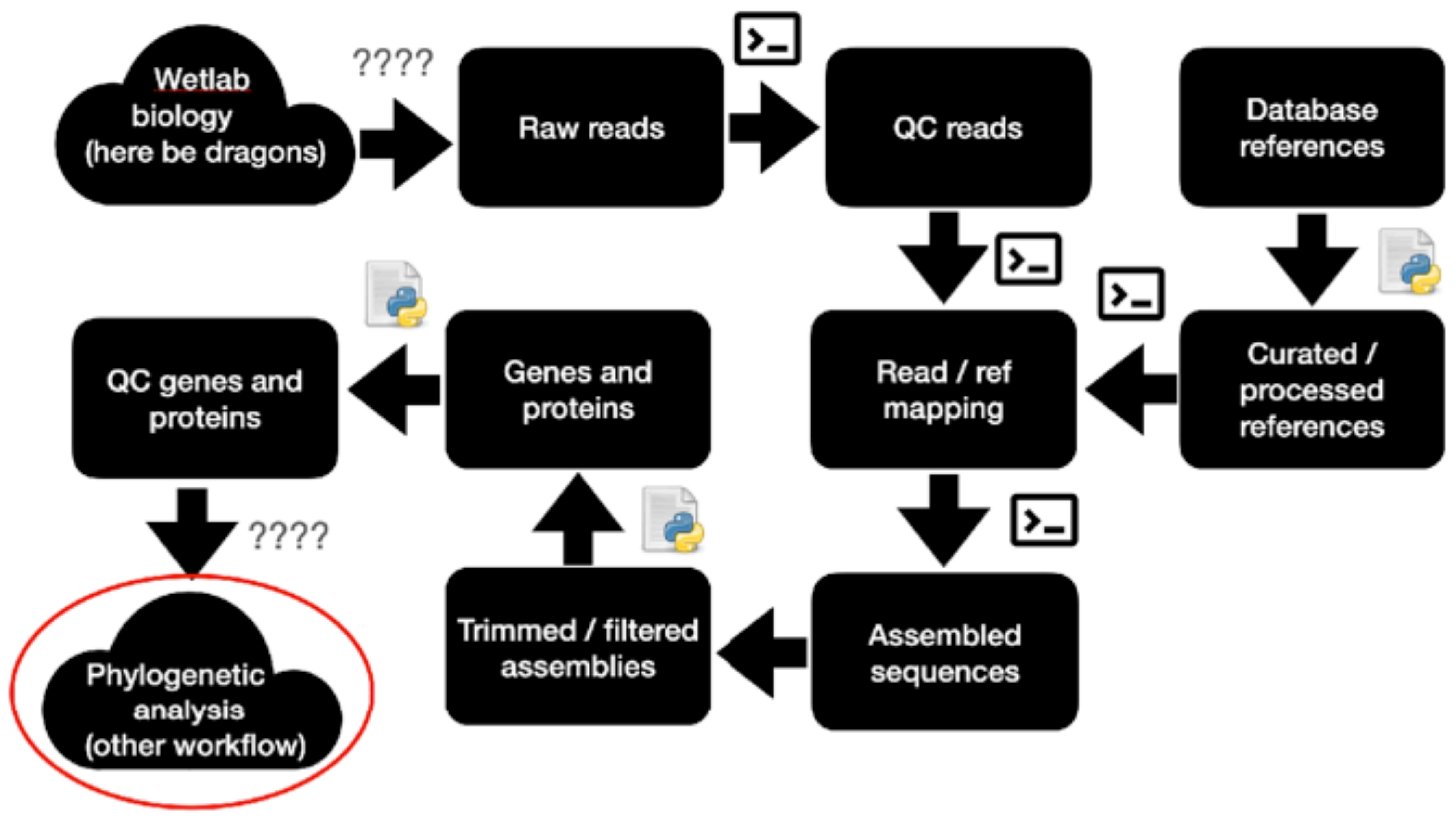


A similar story can be told about mass spectrometry, spatial data, and perhaps microfluidics?

Typical bioinformatics workflow


Wetlab biology (here be dragons)
????
Raw reads
QC reads
Database references
Curated / processed references
Read / ref mapping
Assembled sequences
Trimmed / filtered assemblies
Genes and proteins
QC genes and proteins
Phylogenetic analysis (our goal!)

Typical bioinformatics workflow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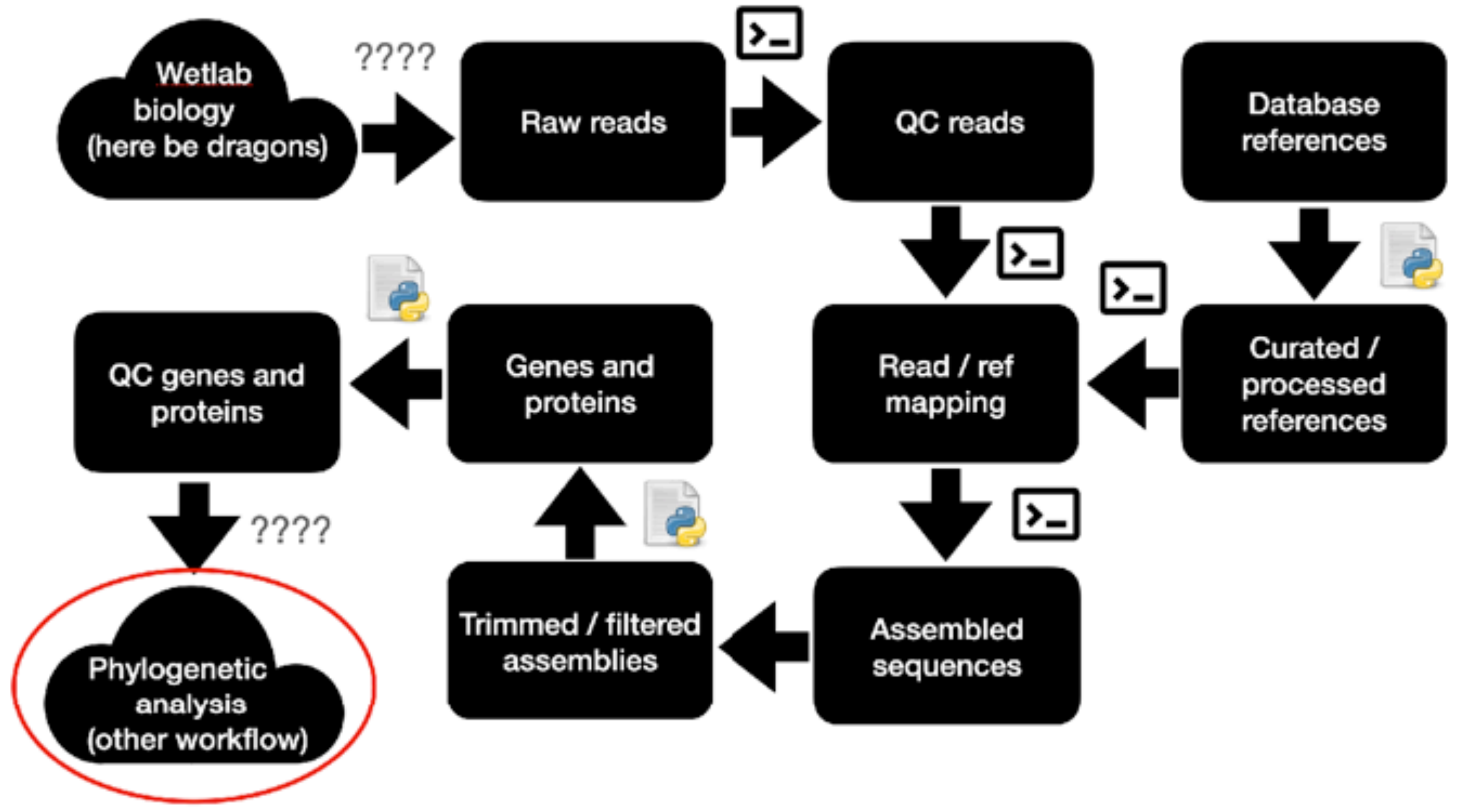
Phylogenetic analysis (other workflow)
Trimmed / filtered assemblies
Assembled sequences

- Workflows are mix of compiled tools and python/R scripts "duct taped" together
- Notoriously unreliable and brittle!
- Many tools *must* be in C/C++ because of the large amounts of data = speed!
- "Bag of tricks" approach all too common!
- But the large amounts of custom code and ever-changing requirements necessitates dynamic scripting languages

Typical bioinformatics workflow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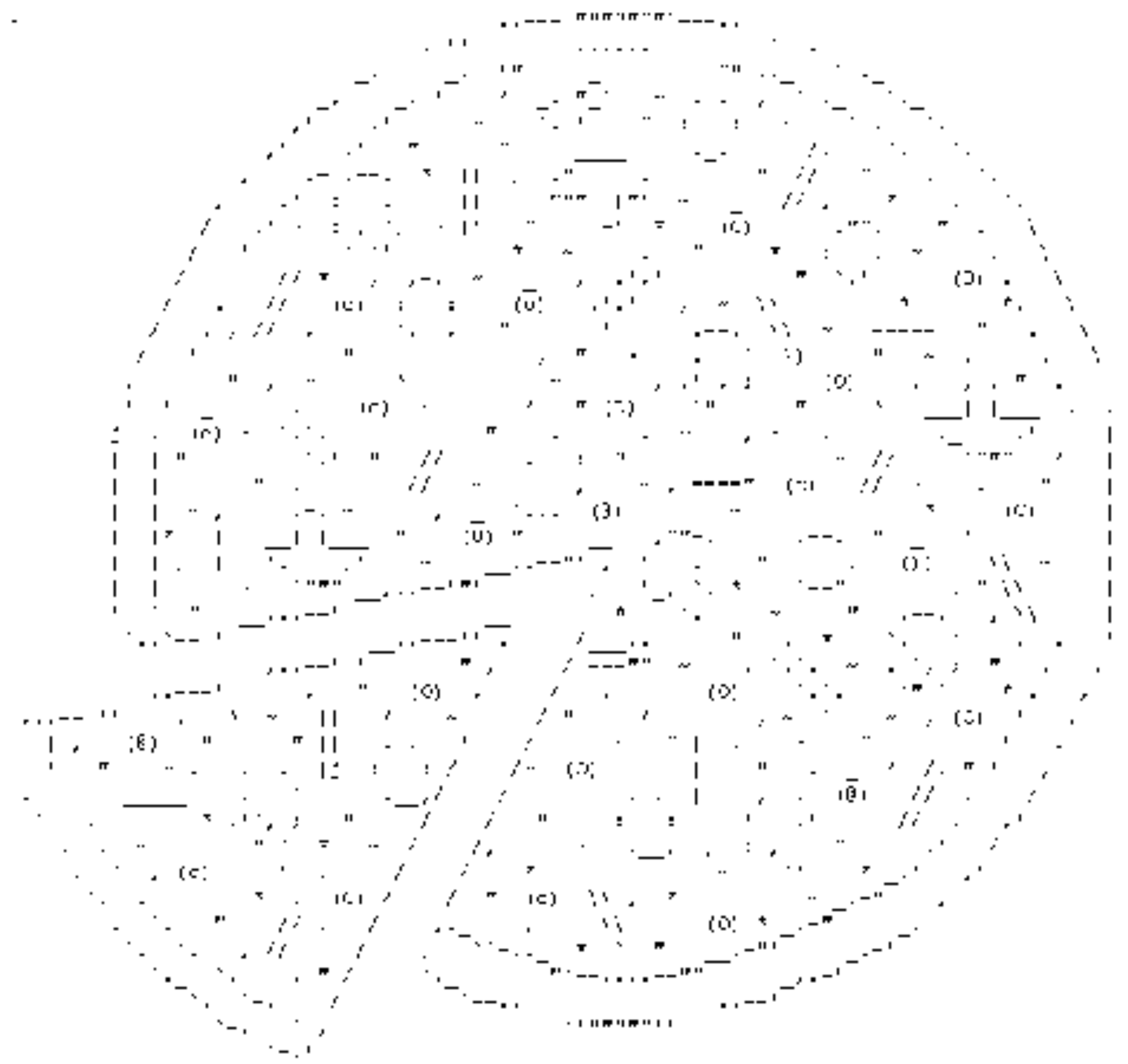
- Each step reads in data from disk, processes, and serialises back
- This leads to a *huge* amount of different file formats, and parsing problems
- Formats are often underspecified and ad-hoc
    - Perhaps deliberately?



Valid DNA sequence file, according to commonly used software

What would a solution look like?

- We need a *fast, dynamic* scripting language
- Rewrite ~~it~~ In Julia?
  - Completely unrealistic. Decades of tools, too much inherent knowledge
  - I can't even rewrite my own PhD project, realistically!
- Instead, improve the *glue language* aspect:
  - Fast enough to never need to use another language due to speed
  - Good at calling into shell and interface with other languages
  - Excellent at parsing
- We also need good "workflow management software", but that's another topic....

BioJulia

- Fast enough to not need C for speed
- Dynamic enough to not need Python for scripting
- Implement *basic, commonly used* bioinformatics algorithms
    - These are the "center of the wheel" of many analyses!
- Types and algorithms should be generic enough to be re-usable across a wide range of applications and tools: Don't re-invent the wheel too many times
- Have great file readers/writers
- Be the swiss army knife of bioinformatics
- Incremental replacement of Python/C with Julia.

BioSequences.jl

- Biological sequences *define* bioinformatics. Very important package!

bioseq is a `str`!"

`ACG"`

oseq is a `&[u8]`!"

`[` `65,67,71]`

- A biological sequence is a biological sequence!
- Internal representation is incidental. Abstract it away unless speed is necessary!
- "Parse, don't validate"

BioSequences.jl

```
BioSequence{Alphabet}
```

- `BioSequence`: Abstract type that all concrete biosequence types subtype
- This abstracts over its representation.

- `Alphabet`: Abstract type all alphabets subtype.
- An alphabet is the set of biological symbols that can be contained in the sequence
- Abstracts over "validation/parsing".
- The `Alphabet` + `BioSequence` type decide how data is encoded concretely.

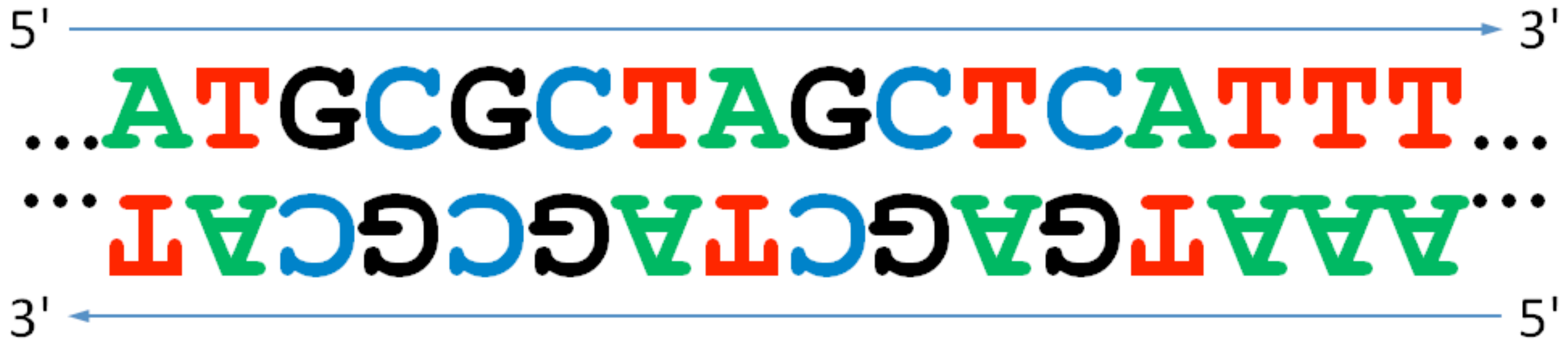
BioSequences.jl

`LongSequence{DNAAlphabet{4}}`

- `LongSequence`: Sequence is arbitrary length, mutable, stored on the heap in a Vector. Elements are encoded by bit-packing `UInt64`.
- You will use another kind of `BioSequence` next exercise!
- `DNAAlphabet{4}`: Sequence can contain `"-ACMGRSVTWYHKDBN"`. Each symbol is packed into 4 bits.
- Other pre-defined alphabets include `RNAAlphabet{2}` (`"ACGU"`) and `AminoAcidAlphabet` (`"ARNDCQEGHILKMFPSTWYVOUBJZX*-"`)

Things to do with biological sequences

Reverse complementation



Transcription and translation

```
DNA: TAG CTA GGC TAA ACA TAG

RNA: UGG CUA GGC UAC UCU UAG

AA :  W   L   G   Y   S  STOP
```

Questions?

Exercise 2