

Genome sequencing and assembly

The story of four-year-old Georgia Walburn-Green and her family.

<http://www.itv.com/news/2016-01-12/georgias-story-gives-hope-to-many-families-after-dna-sequencing-breakthrough/>



The story of four-year-old Georgia Walburn-Green and her family.

Born seemingly healthy, her parents Amanda and Matt told ITV News they only had 20 minutes of “worry-free cuddling” with their brand new baby before doctors drew the curtains around the bed to tell them something was not quite right - they could see that Georgia’s head was bigger than normal.

News on ITV on 12-jan-2016

Since then, Georgia has had a range of problems with her eyes, kidneys and brain.

Her physical and mental development has been delayed. Despite years of tests, though, doctors couldn't pinpoint what was wrong with this otherwise lively, smiling girl.

That changed a few weeks ago, when Amanda and Matt got a phone call from scientists at Great Ormond Street Hospital.

Georgia's condition, they were told, was due to a mutation in a gene called Kdn5b.

Fragment assembly of DNA

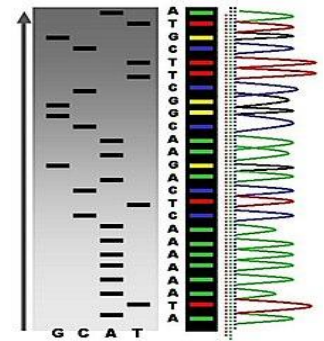
- Biological background
- Models
- Algorithms
- Heuristics

Fragment assembly of DNA

- Fred Sanger developed the first technique for sequencing DNA in 1977.
- DNA is replicated in the presence of chemically altered versions of the A, C, G, and T bases.
- These bases stop the replication process when they are incorporated into the growing strand of DNA, resulting in varying lengths of short DNA.
- These short DNA strands are ordered by size, and by reading the end letters from the shortest to the longest piece, the whole sequence of the original DNA is revealed.

Fragment assembly of DNA

- [Automated method of DNA sequencing](#), built upon the chemistry of PCR and the sequencing process developed by Frederick Sanger in 1977.
- https://scholar.google.co.in/scholar?q=Sanger+method+of+DNA+sequencing&btnG=&hl=en&as_sdt=0%2C5
- <http://smcg.ccg.unam.mx/enp-unam/03-EstructuraDelGenoma/animaciones/secuencia.swf>
- <https://www.dnalc.org/view/15479-Sanger-method-of-DNA-sequencing-3D-animation-with-narration.html>
- <http://www.yourgenome.org/video/sanger-dna-sequencing>



Limitations to sequencing

- You must have a primer of known sequence to initiate PCR
- Only about 1000nts can be sequenced in a single reaction

Limitations to sequencing

- Human Genome sequencing ?