Evolution of chromosomes and genomes

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Chromosome rearrangements

- Inversion
- Translocation
- Transposition
- Tetraploidy

Additional question: when these occur they are rare in a population: how do they spread?
Chironomus banded chromosomes

Fig. 4.1. The four polytene elements from a salivary gland nucleus of Chironomus thummi, showing the banding. Small arrows indicate the approximate positions of the centromeres. The shortest chromosome has its longer arm largely heterochromatic and the other three elements have short heterochromatic regions at the tips. From Bauer (1935).
Paracentric and pericentric inversions

A Paracentric Inversion

A B C D E F G H I J

A B C D E F G H I J

A B F E D C G H I J

A B F E D C G H I J

A B F E D C G H I J

A B C E F G H I J

A B C I J

E F G H D

A B C E F G H I J

A B C I J

E F G H D
Pairing in a paracentric inversion heterozygote
Crossover in a paracentric inversion heterozygote
These two chromosomes did not cross over
This one did. How will it segregate?
This one did too. How will it segregate?
Pairing in a pericentric inversion heterozygote
Crossover in a pericentric inversion heterozygote
These two products didn’t undergo crossing-over
One that did. Is anything deleted? Duplicated?
The other one that did. Anything deleted, duplicated?
A translocation

Before

Breaks

Rearrangement

After
Segregation of translocation heterozygote

at first division of meiosis metaphase

A pair of translocated chromosomes
pairs with a pair of untranslocated chromosomes
Adjacent segregation in translocations

... Leads to deletions and duplications.
Alternate segregation in translocations

... Leads to euploid (normal gene complement) gametes
Banding pattern changes

in two species of Chironomus midges.
Polytene Drosophila chromosomes
FIGURE 5.2

Phylogenetic relationships of the gene arrangements in the third chromosomes of Drosophila pseudoobscura, D. persimilis, and D. miranda.
Chromosome phylogeny of Hawaiian Drosophila
Formation of the Hawaiian islands
Geology of the Hawaiian islands

A chain of seamounts leads northwest back across the Pacific as far as the Kamchatka peninsula of Siberia, an estimated 85 million years.
Inferred migration events on the Hawaiian islands

![Diagram showing inferred migration events on the Hawaiian islands. The diagram illustrates the movement of species from one island to another, with arrows indicating the direction of migration. Each island is labeled with a number, and the number of species found on each island is given in parentheses. The width of the arrows is proportional to the number of proposed founders.](image-url)
When a tetraploid mates with a diploid

A tetraploid

Gametes

Triploid offspring

An ordinary diploid
The gametes are aneuploid

- A tetraploid
- An ordinary diploid

Gametes

Triploid offspring

Gametes
And so are the offspring

a tetraploid

an ordinary diploid

gametes

triploid offspring

gametes

the resulting aneuploid offspring
Polyploid evolution in Clarkia

C. Williamsonii
Fort Miller Clarkia

C. prostrata – 26
C. purpurea ssp. purpurea – 26
C. purpurea ssp. quadrirugulosa
C. affinis – 26
C. davyi – 17
C. tenella – 17
C. speciosa ssp. speciosa – 9
C. nitens – 9
C. imbricata – 8

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The fern *Ophioglossum*, high number champion

Highest number is in this genus, about 1260 chromosomes, or 630 pairs.
The ant *Myrmecia*, low chromosome number champion

Myrmecia pilosula, the “Jack-jumper ant”

Fig. 1. Chromosomes from prepupal cerebral ganglia. (A) Worker prometaphase chromosomes. Identical C-banding provides evidence for homology of the two chromosomes. (B) Male prometaphase chromosome. Chromosomes consistently display a large centromeric C-band on the short arm and a smaller centromeric C-band on the long arm. Most of the short-arm C-band is not immediately adjacent to the centromere, though a very small portion of the short-arm C-band is centromeric. Arrows indicate position of centromere.
The (temporary?) death of junk DNA

An integrated encyclopedia of DNA elements in the human genome

The ENCODE Project Consortium

Affiliations | Contributions | Corresponding author

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Abstract

Abstract | Introduction | ENCODE data production and initial analyses | ENCODE data integration with known genomic features | Genome-wide integration | Insights into human genomic variation | Rare variants, individual genomes and somatic variants | Common variants associated with disease | Concluding remarks | Methods | References | Acknowledgements | Author information | Supplementary information | Comments

The human genome encodes the blueprint of life, but the function of the vast majority of its nearly three billion bases is unknown. The Encyclopedia of DNA Elements (ENCODE) project has systematically mapped regions of transcription, transcription factor association, chromatin structure and histone modification. These data enabled us to assign biochemical functions for 80% of the genome, in particular outside of the well-studied protein-coding regions. Many discovered candidate regulatory elements are physically associated with one another and with expressed genes, providing new insights into the mechanisms of gene regulation. The newly identified elements also show a statistical correspondence to sequence variants linked to human disease, and can thereby guide interpretation of this variation. Overall, the project provides new insights into the organization and regulation of our genes and genome, and is an expansive resource of functional annotations for biomedical research.
## Pros and cons

<table>
<thead>
<tr>
<th>Pro</th>
<th>Con</th>
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<tbody>
<tr>
<td>1. Much of the genome is transcribed.</td>
<td>1. The Onion Test: do onions really need $5 \times$ as much DNA as we do?</td>
</tr>
<tr>
<td>2. Transcription factor binding sites are found throughout the genome.</td>
<td>2. Mutational load would be excessive if the whole genome needs to be kept in its current state</td>
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<tr>
<td>3. Just think of all the grants we can apply for to work out the functions of all that DNA!</td>
<td>3. Most of the genome isn’t conserved evolutionarily.</td>
</tr>
<tr>
<td>4. Annotatable “selfish” DNA elements make up about half of the human genome.</td>
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Distribution of chromosome numbers in mammals

[Histogram showing the distribution of chromosome numbers in mammals, with different categories like Monotremes, Marsupials, and Eutheria.]
Karyotypes of Drosophila species

Fig. 11.3. Male karyotypes of some members of the subgenus Sophophora of Drosophila. Several different karyotypes have been reported for montium and takahashii. Based on various authorities.
Karyotypes of gymnosperms

Fig. 4.12  Karyotypes of various genera of gymnosperms. (a) *Pinus*, showing the symmetrical karyotype characteristic of the families Pinaceae, Cupressaceae, and most genera of Taxodiaceae. (b), (c), Moderately asymmetrical karyotypes of *Amentotaxus argyrotanis* (Taxaceae) and *Stangeria paradoxa* (Cycadaceae). (d), (e), (f), Strongly asymmetrical karyotypes of *Podocarpus nivalis* (Podocarpaceae), *Ginkgo biloba* (Ginkgoaceae), and *Welwitschia mirabilis* (Welwitschiaceae). ((a) from Bowden,²¹ (b) from Chuang and Hu;²⁰ (c) from Marchant;¹⁵⁸ (d) from Hair and Beuzenberg;²⁰ (e) from Lee;¹³¹ (f) from Khoshoo and Ahuja.¹²²)
Human and mouse genomes compared

The color code identifies the Mouse chromosome numbers

chr 1
chr 2
chr 3
chr 4
chr 5
chr 6
chr 7
chr 8
chr 9
chr 10
chr 11
chr 12
chr 13
chr 14
chr 15
chr 16
chr 17
chr 18
chr 19
chr 20
chr 21
chr 22

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Human and chimp karyotypes compared