

# **CONTENIDO EN GC EN EL GENOMA HUMANO**

**Héctor Musto**

**Laboratorio Genómica Evolutiva**

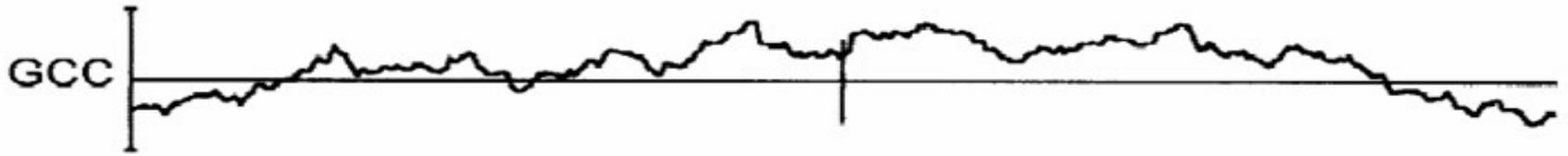
**Departamento Biología Celular y Molecular**

**Facultad de Ciencias**

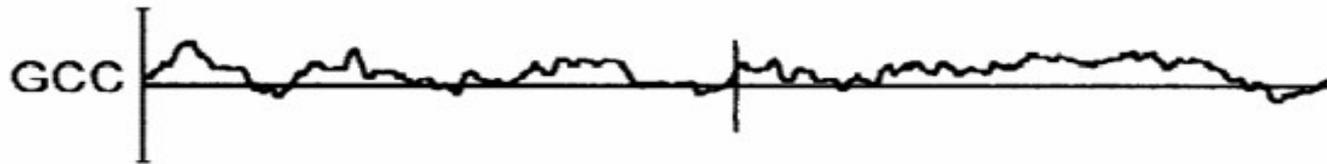
**Uruguay**

**hmusto@gmail.com**

# Variación en el contenido en GC% en procariontas



*E. coli*



*Synechocystis* sp.

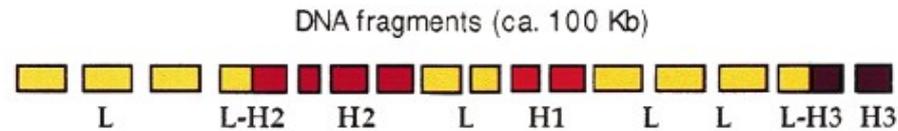


*M. jannaschii*

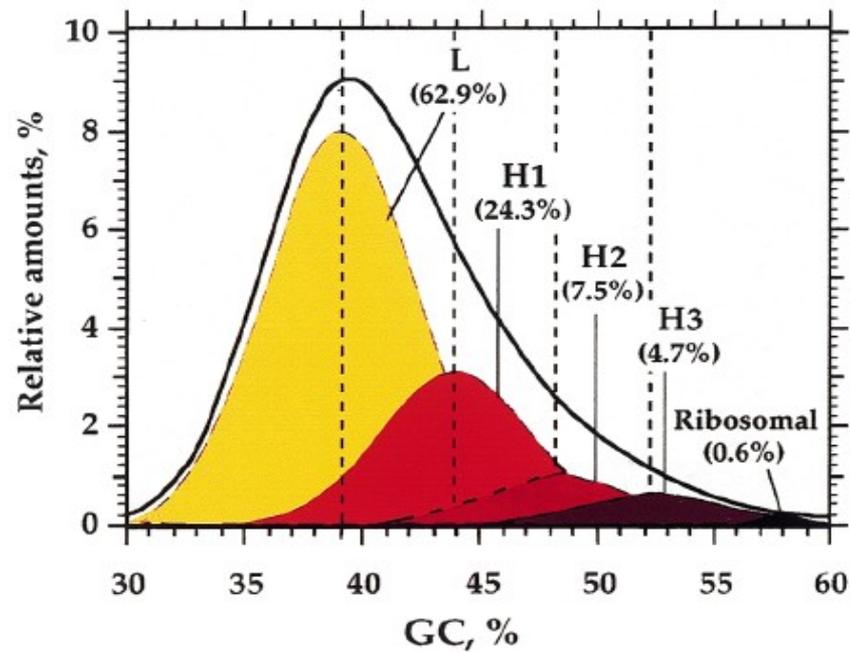
ISOCHORES >300 Kb

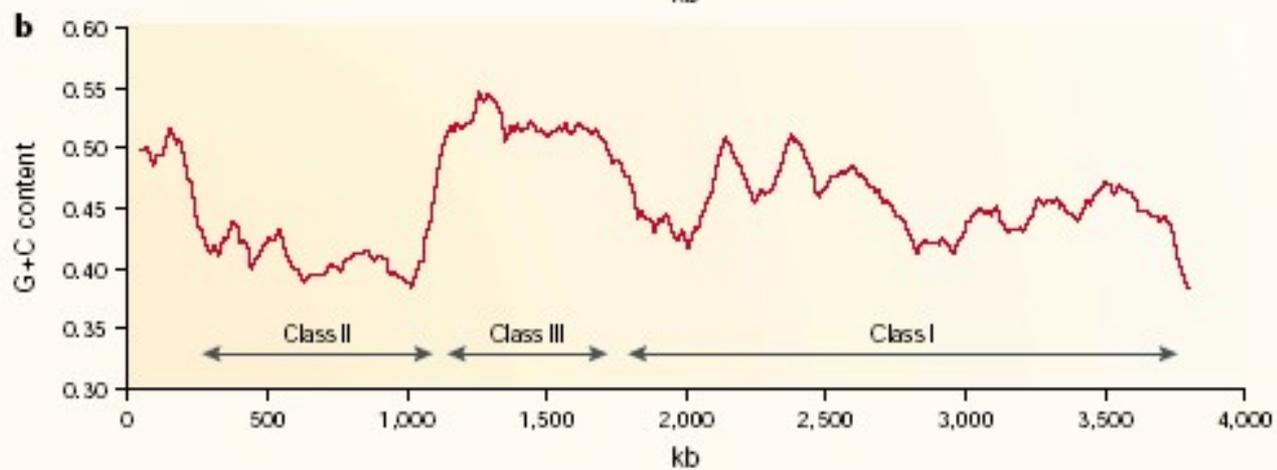
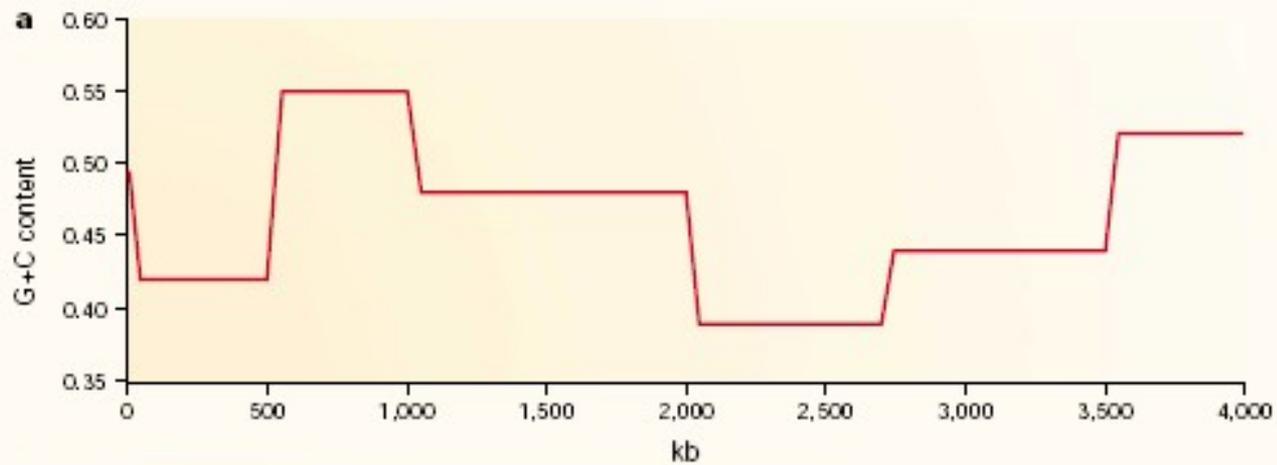


Degradation during DNA preparation

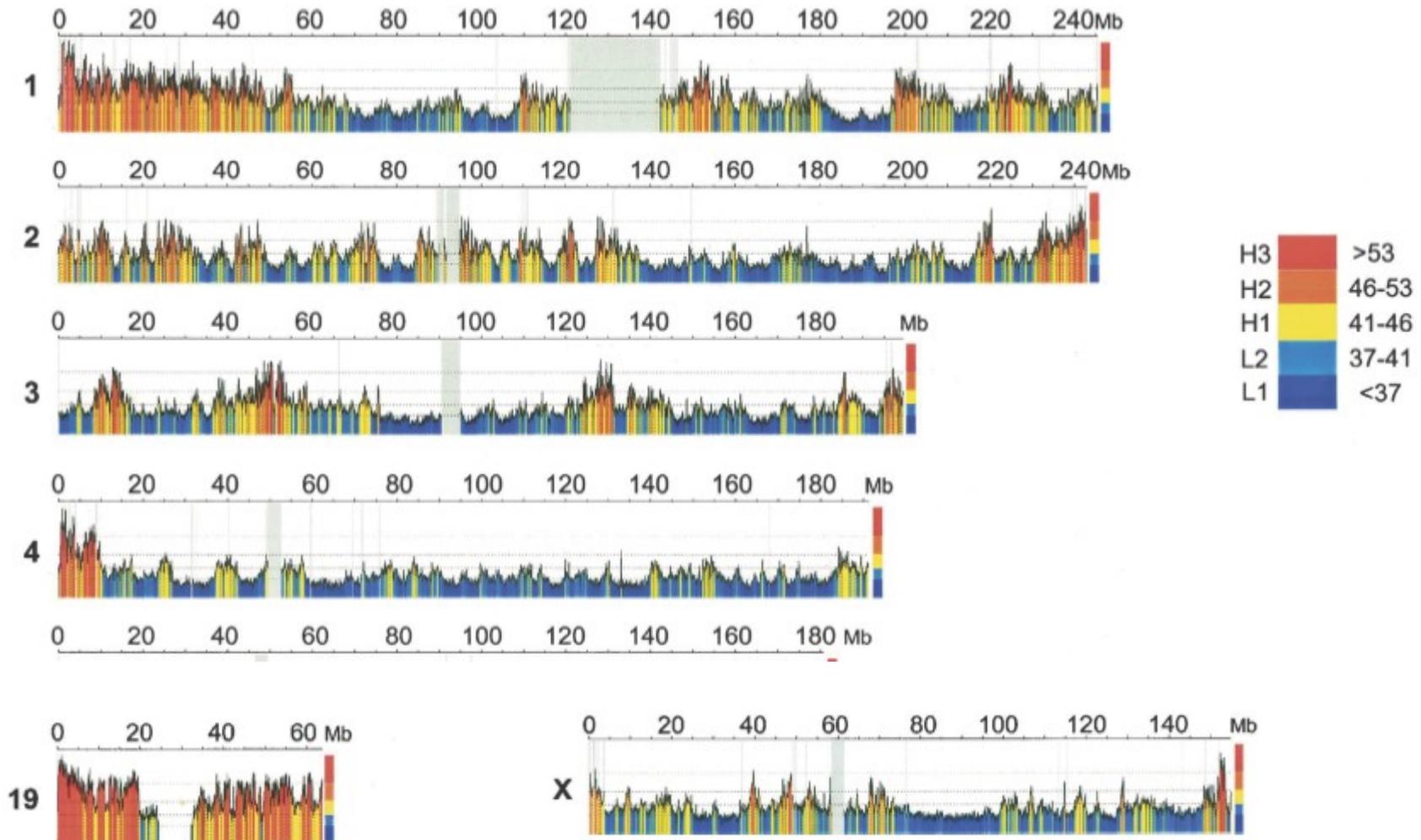


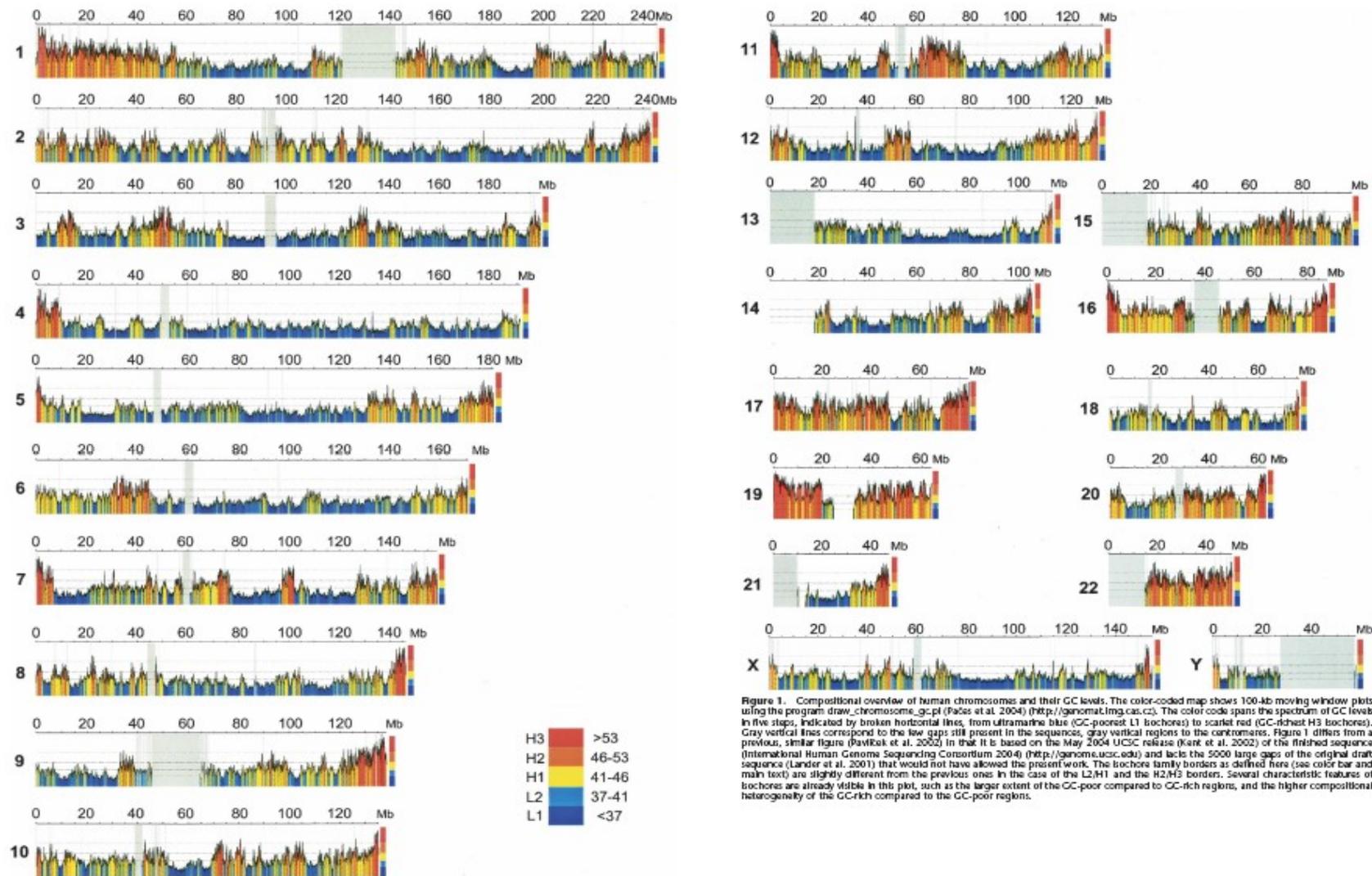
GC range 30-60%



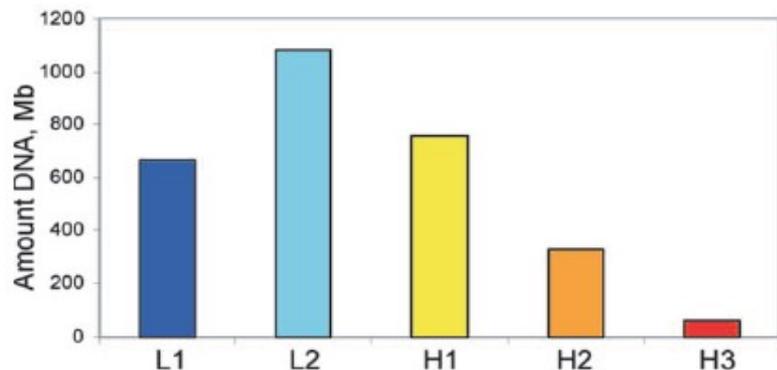


# Visualización de los isocoros por métodos computacionales

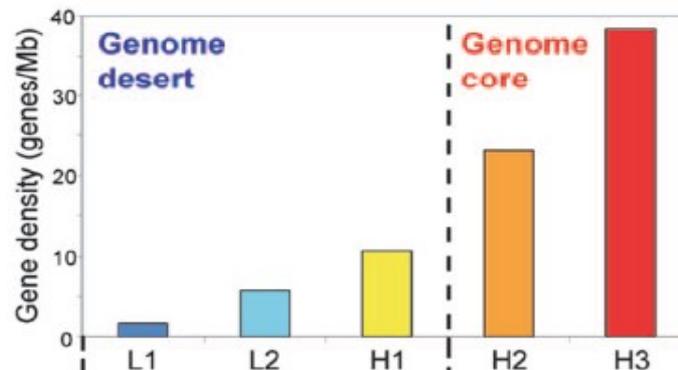




## Isochore families



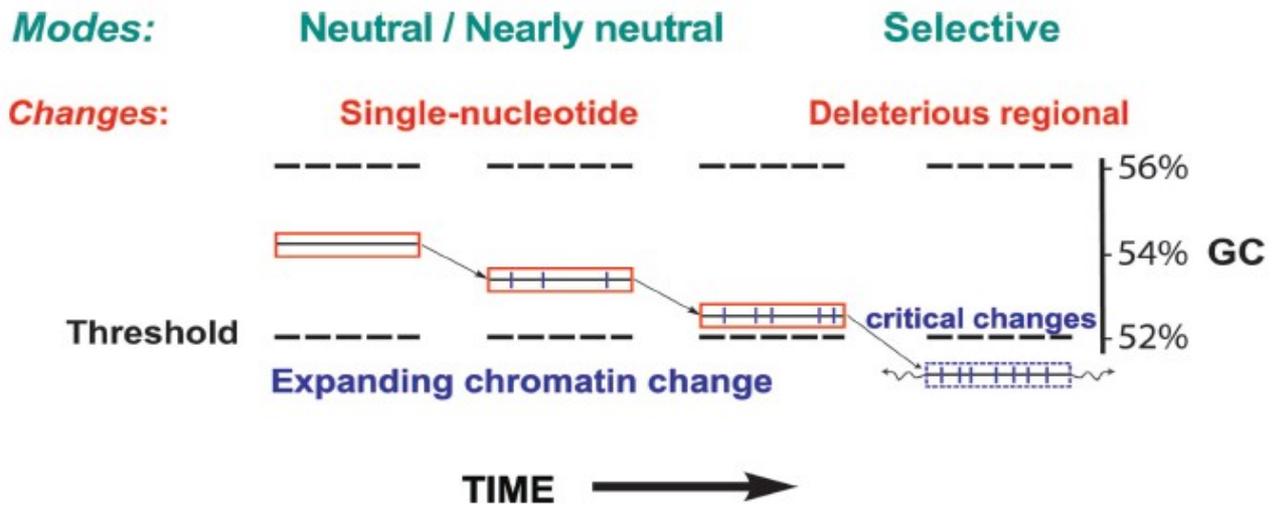
## Gene distribution



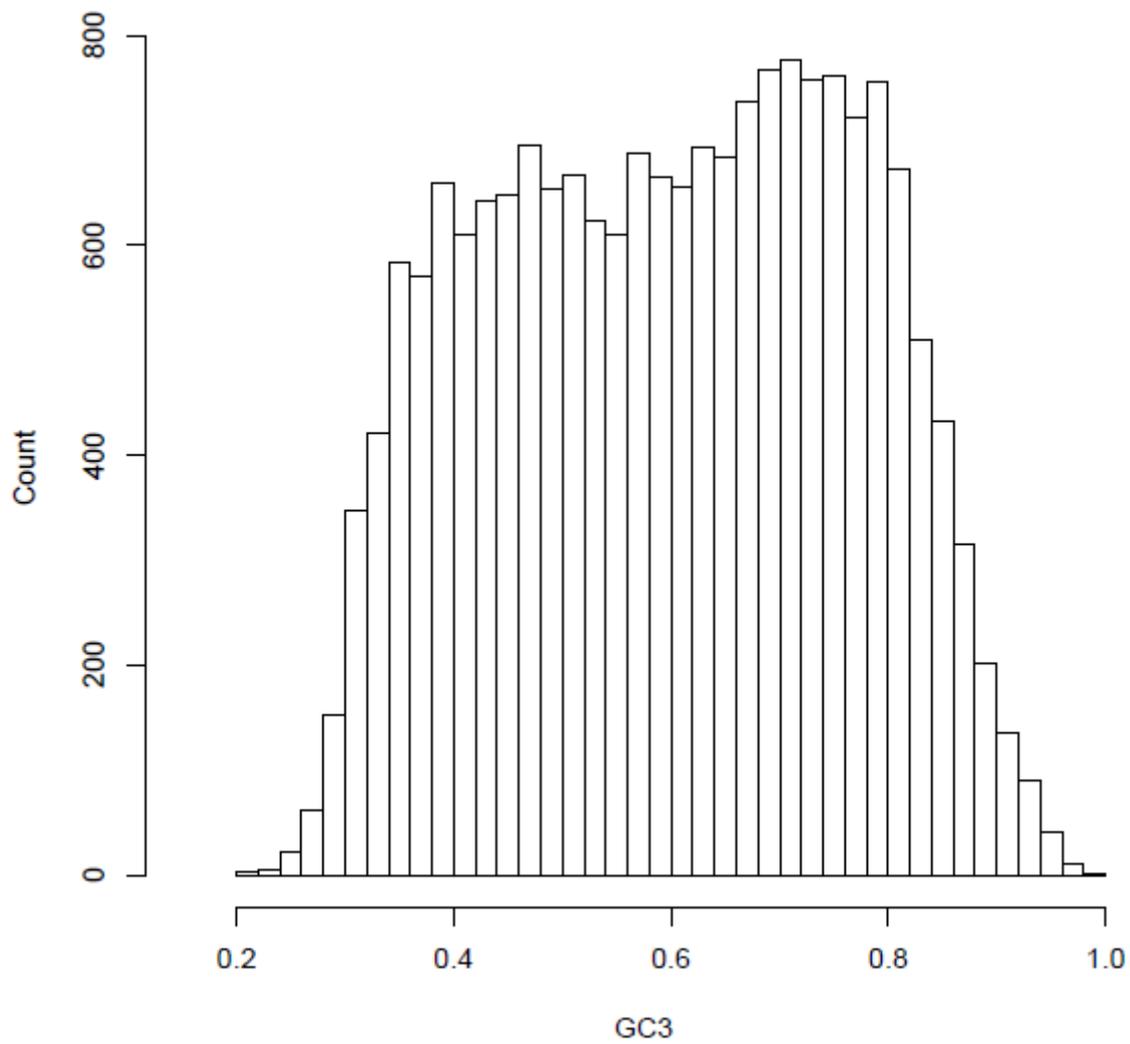
Increasing GC →

**Correlations of gene spaces with structures and functions**

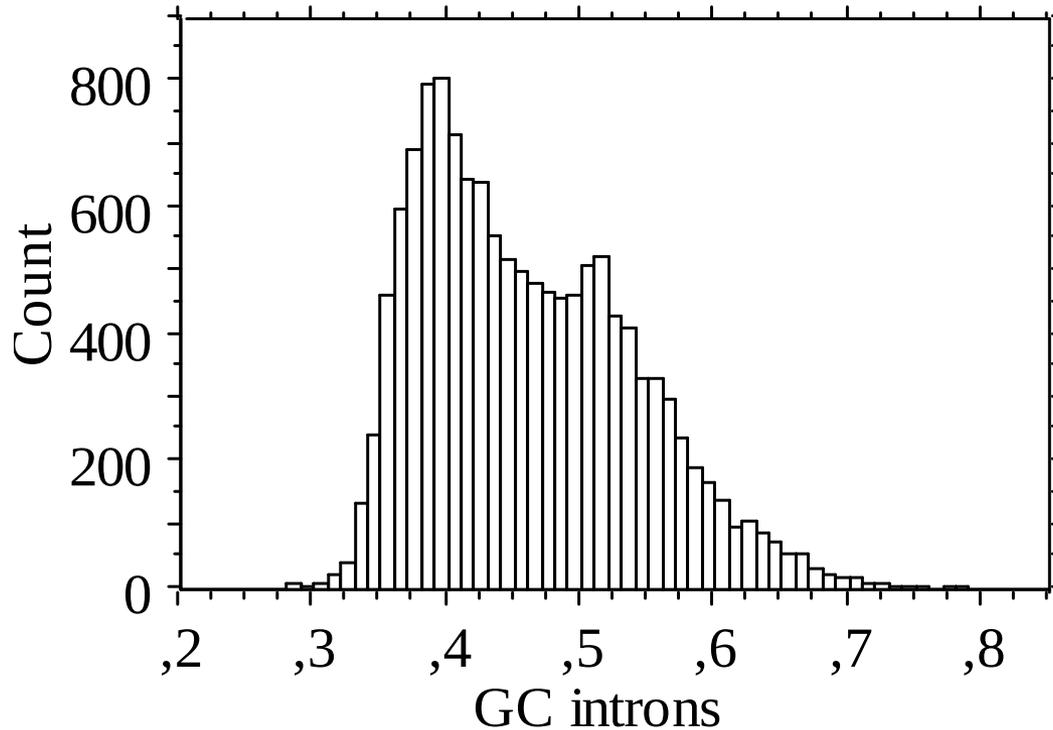
Intron, UTR size	Large	Small
Chromatin structure	Closed	Open
GC heterogeneity	Low	High
SINEs	Low	High
LINES	High	Low
Gene expression	Low	High
Replication timing	Late	Early
Recombination	Low	High



GC3 of CDS (All Chromosomes) n=19,046

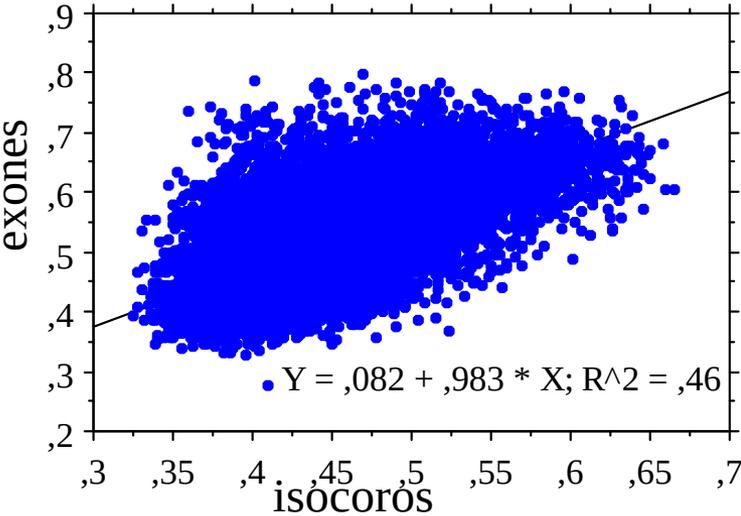


# Contenido en GC de los intrones humanos

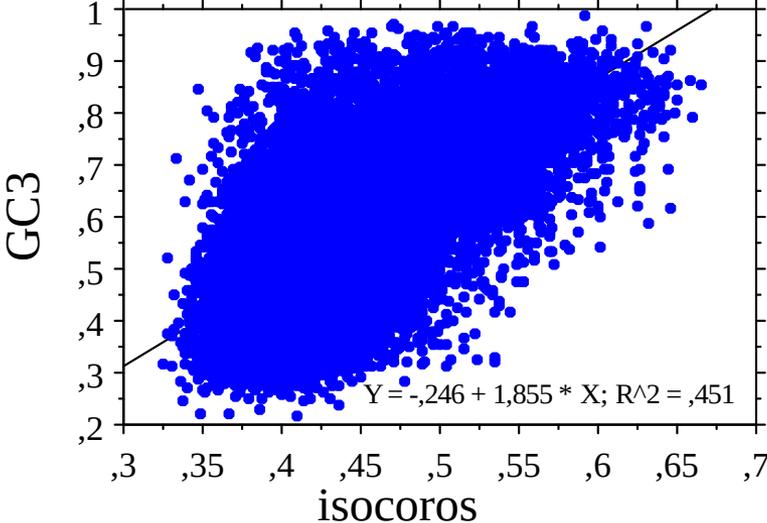


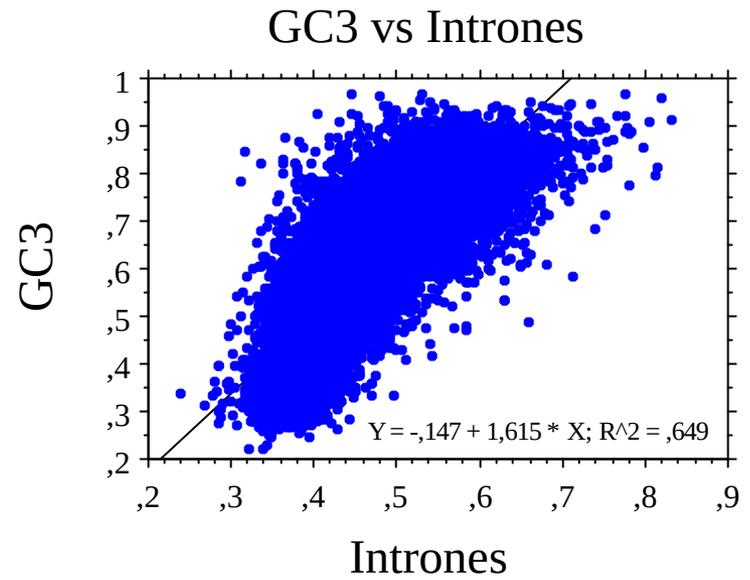
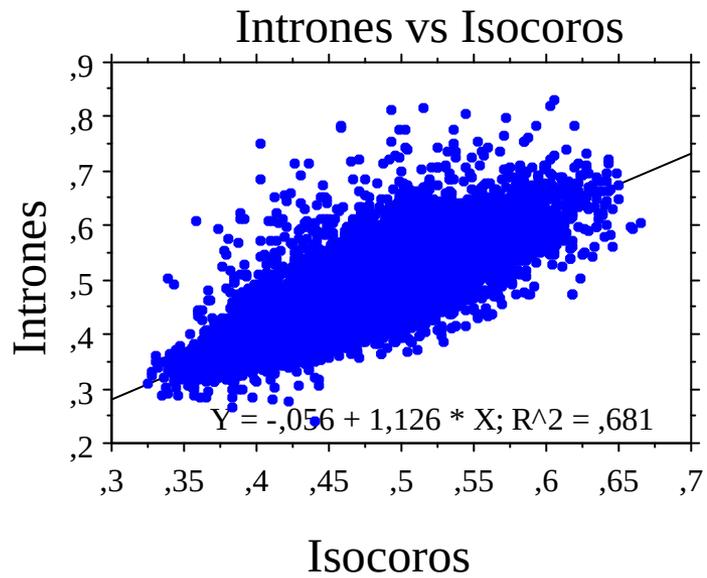
# CORRELACIONES COMPOSICIONALES

GC exones vs Isocoros

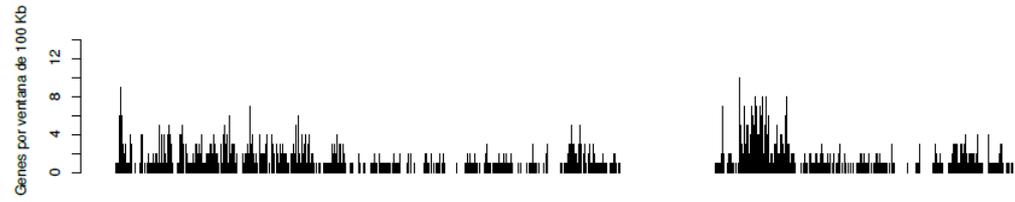
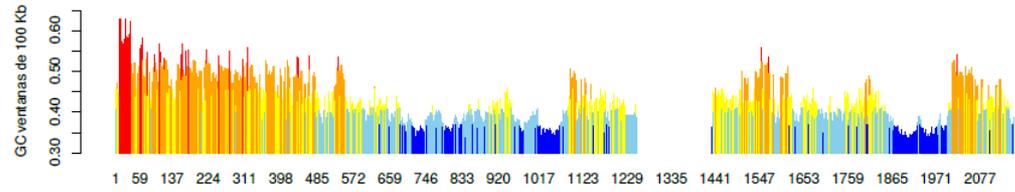


GC3 vs Isocoros

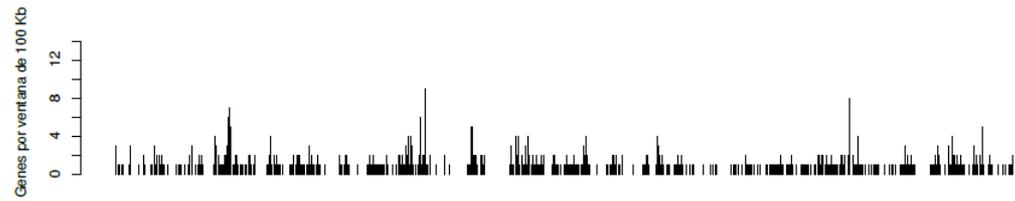
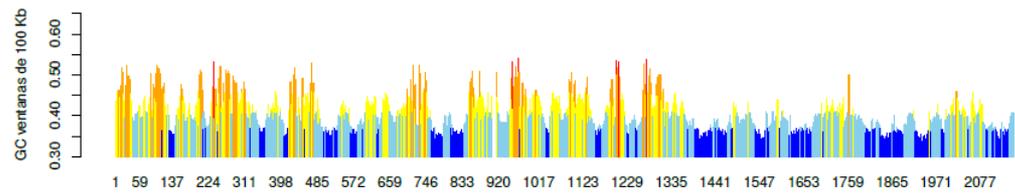




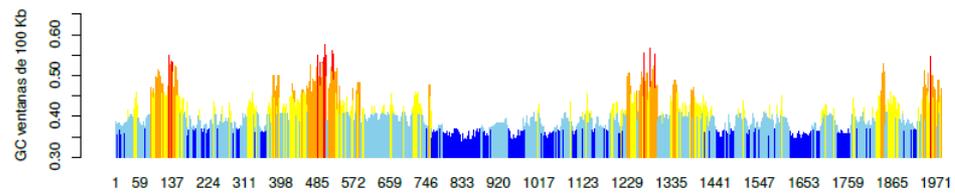
### Chr 1



### Chr 2

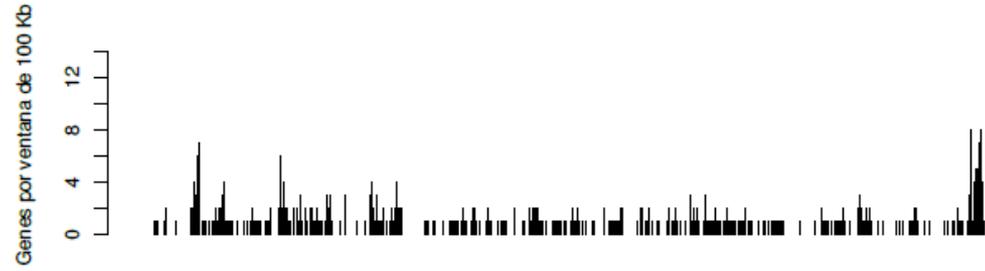
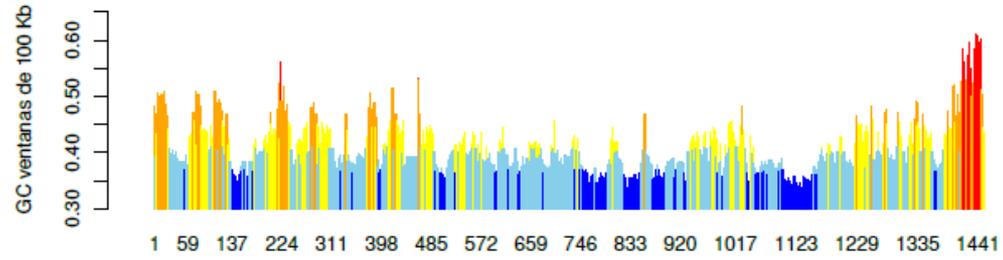


### Chr 3

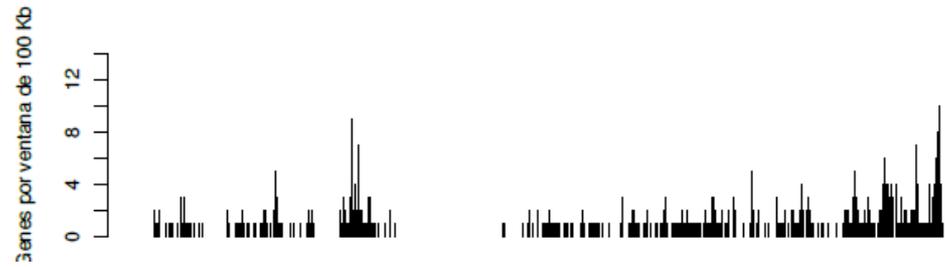
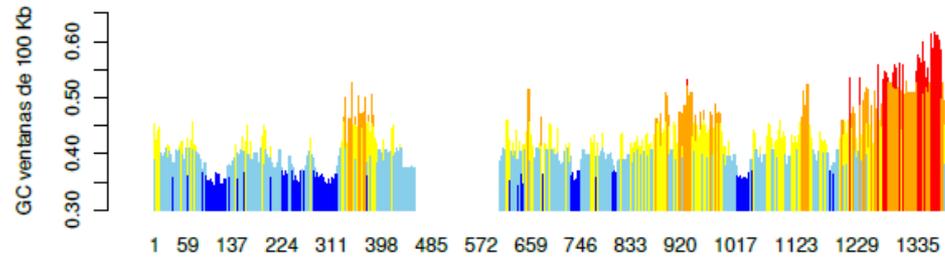


0 Kb

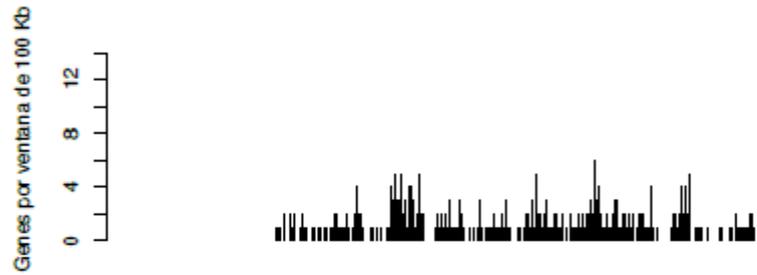
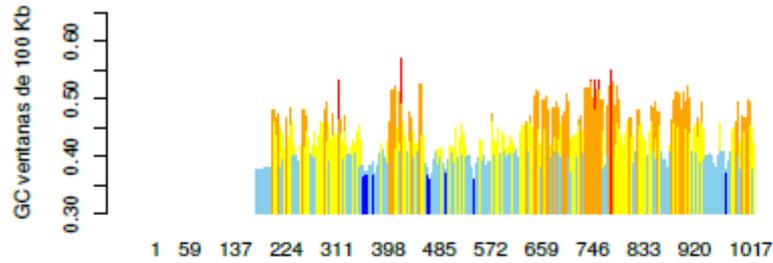
### Chr 8



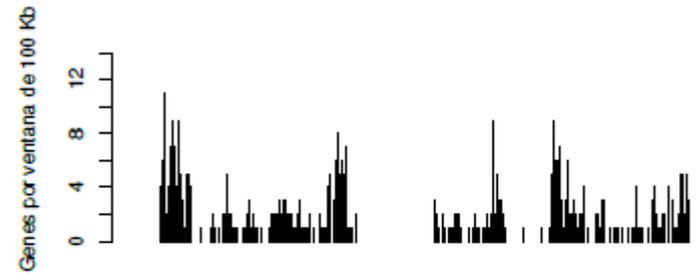
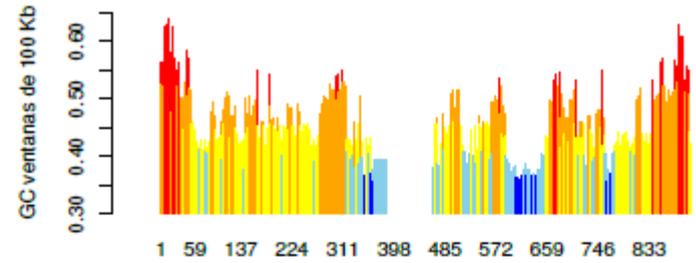
### Chr 9



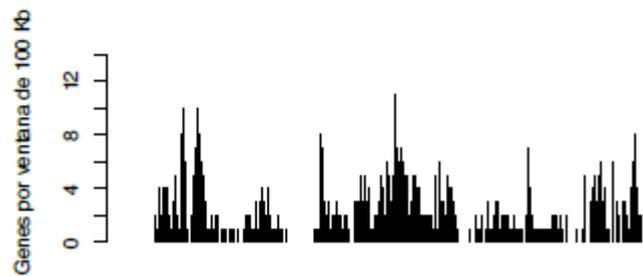
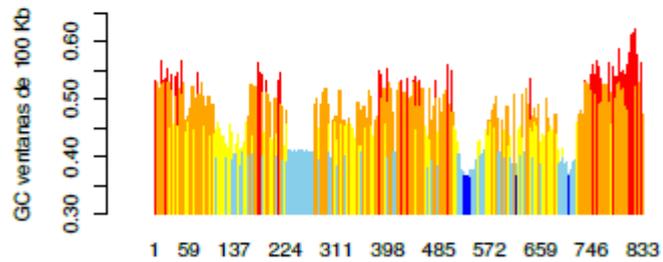
Chr 15



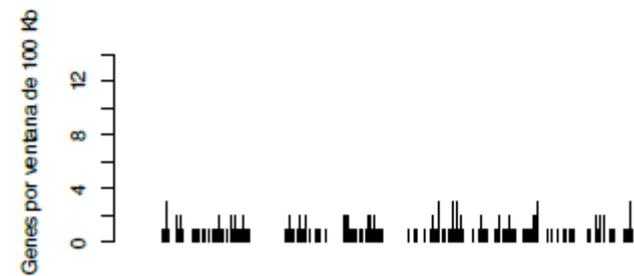
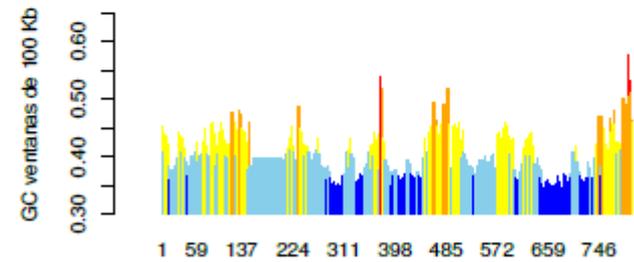
Chr 16



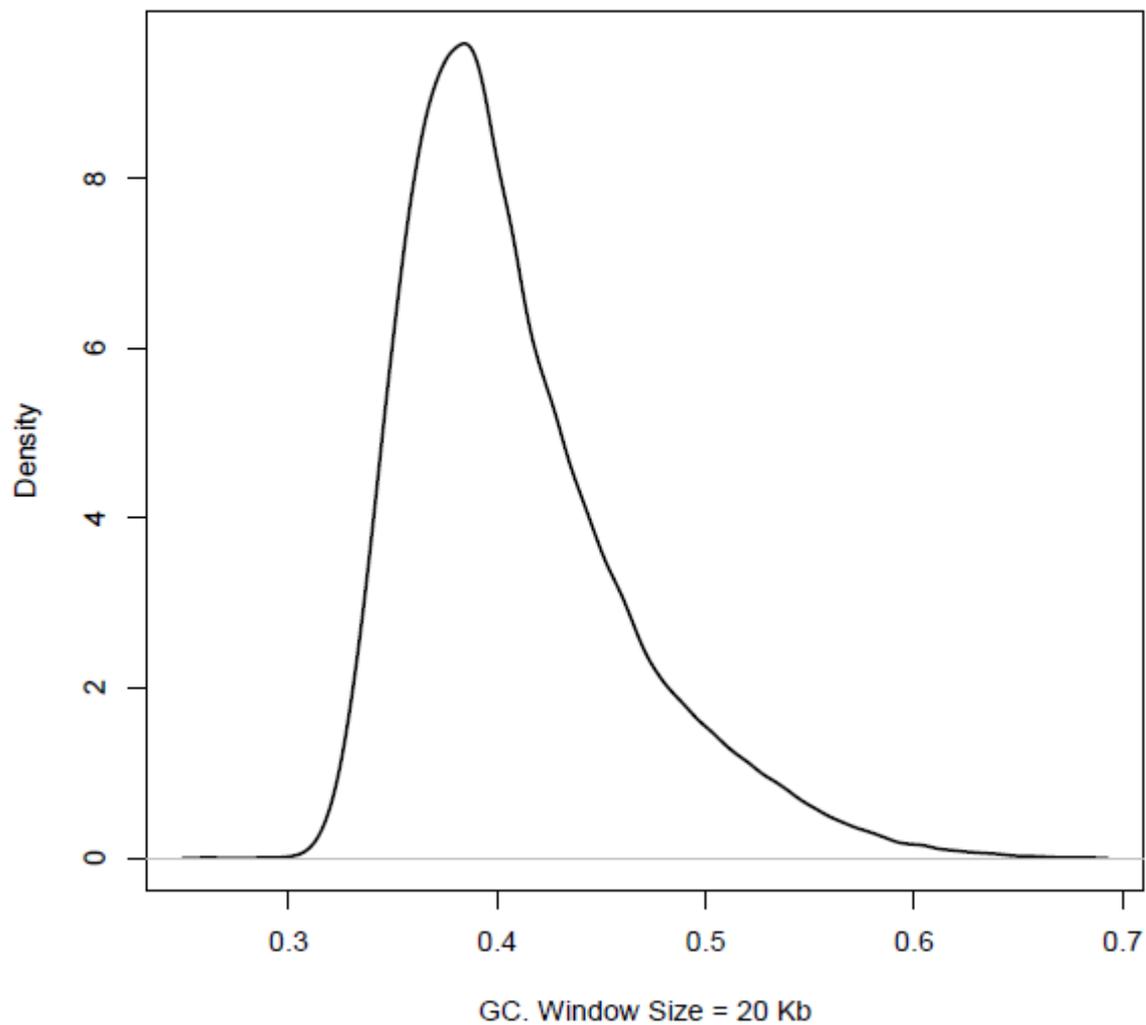
Chr 17



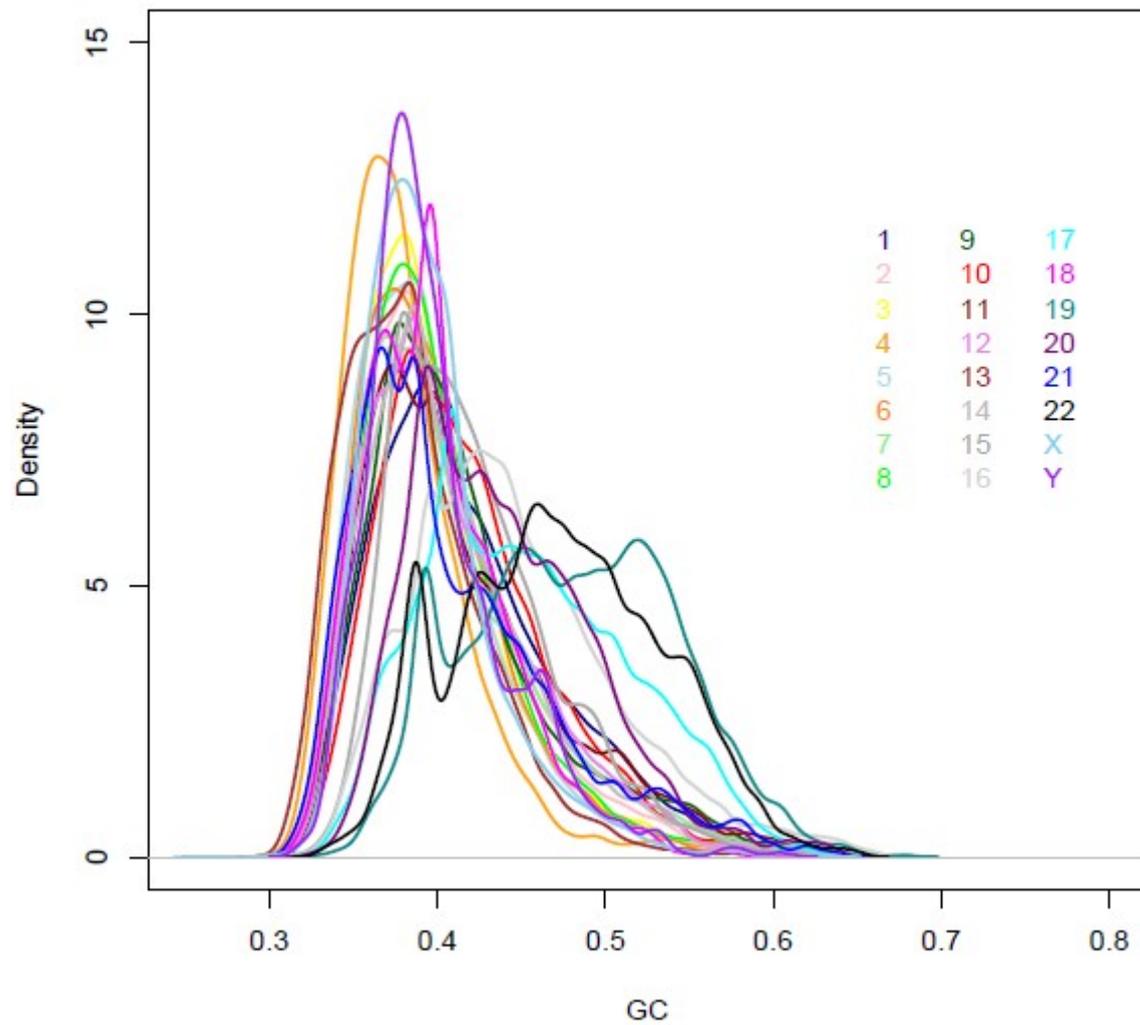
Chr 18



### Human Genome: GC distribution

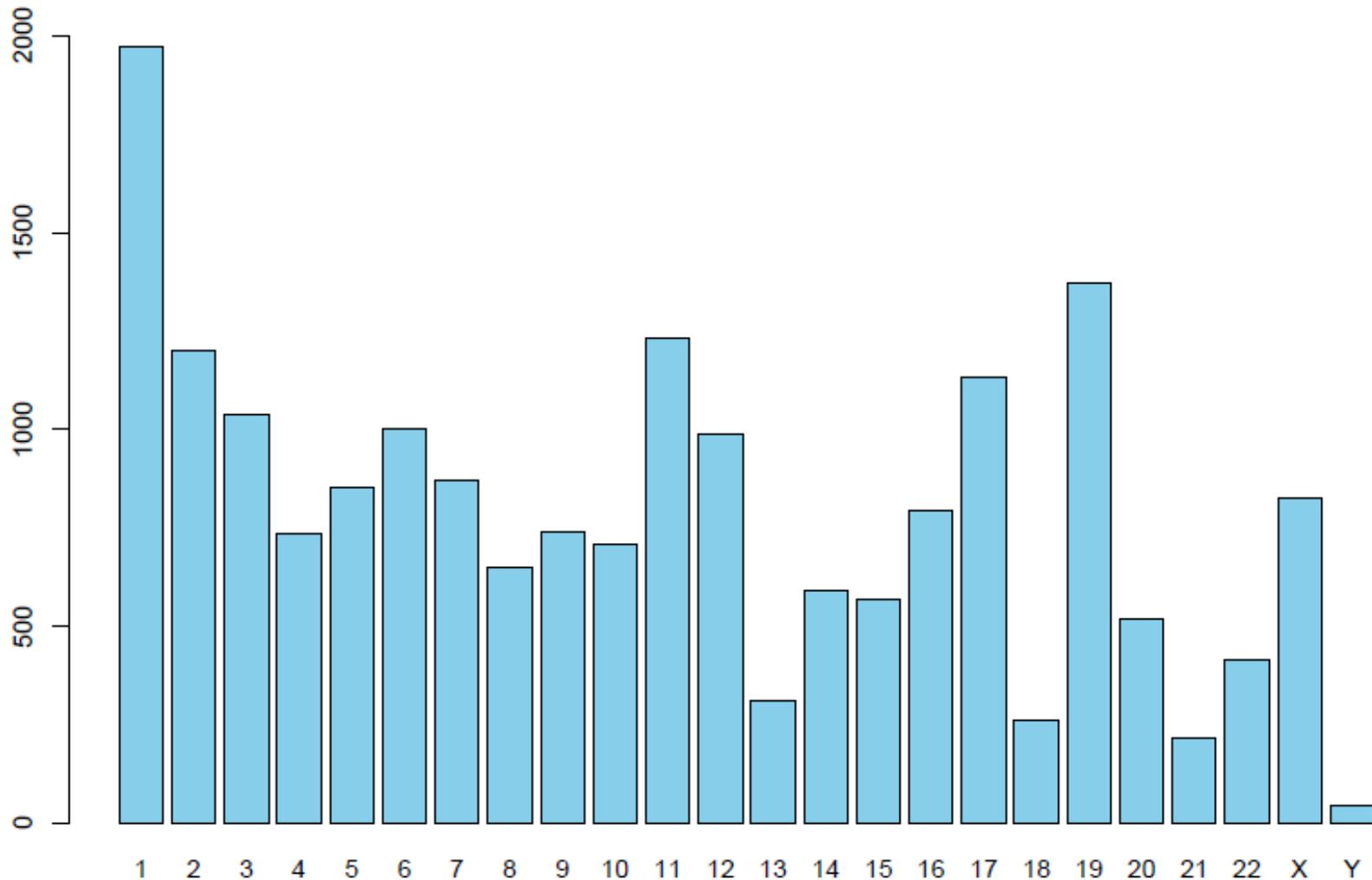


### All Chromosomes

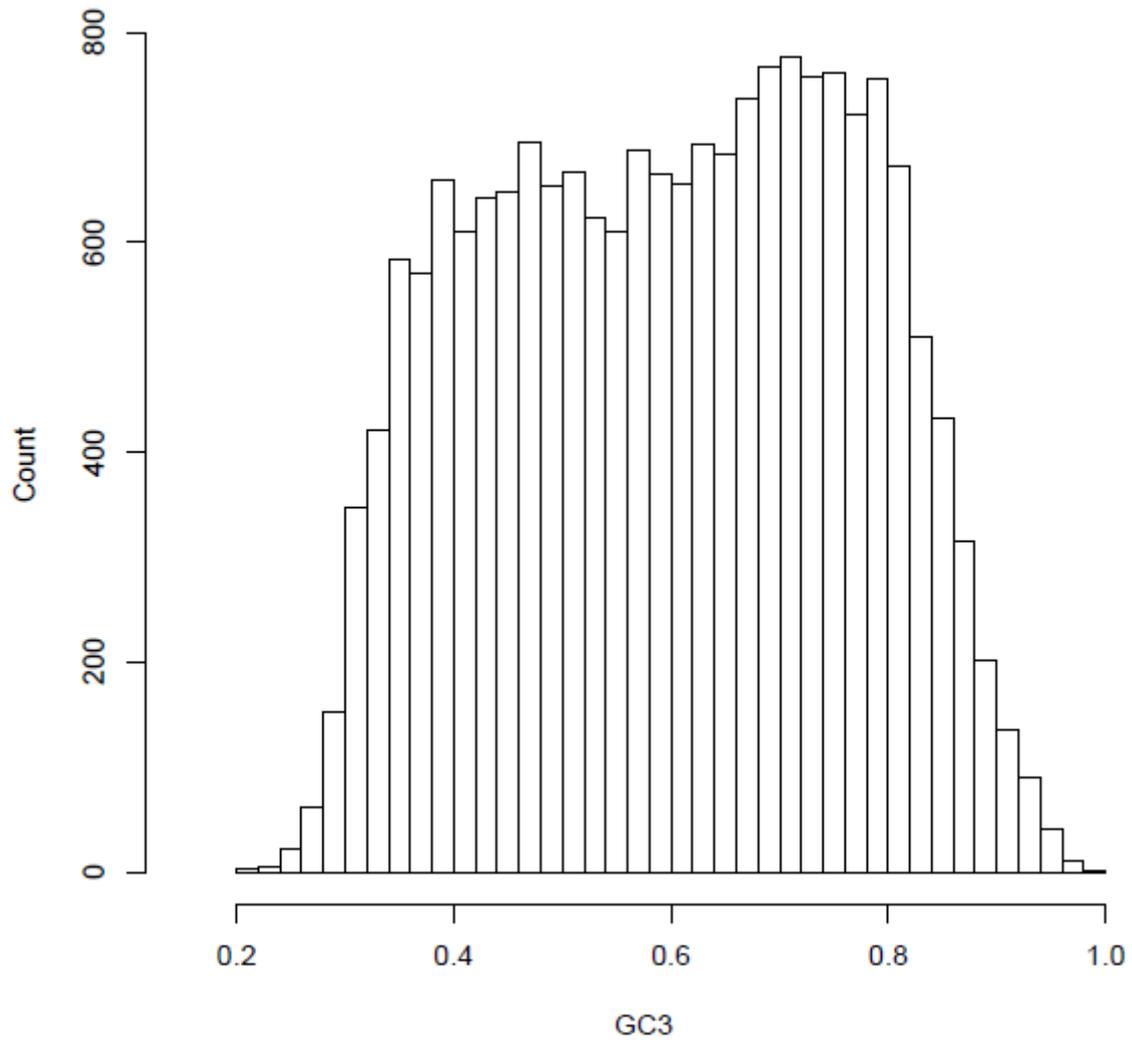


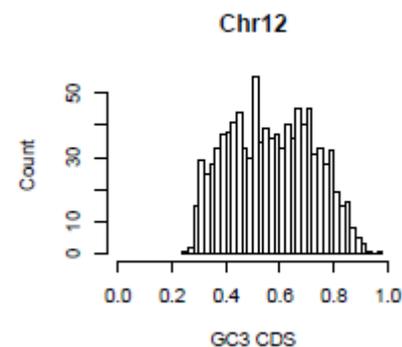
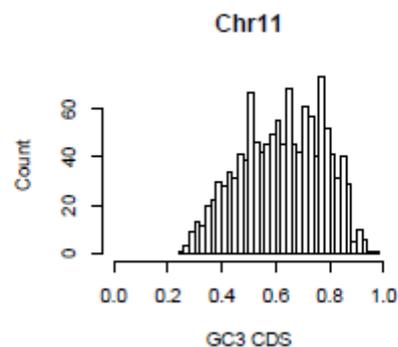
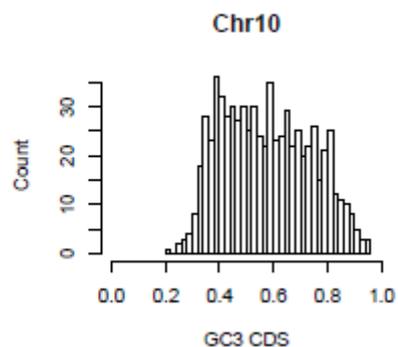
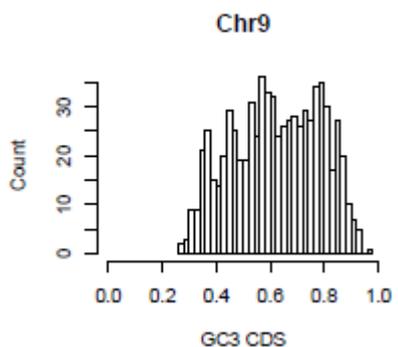
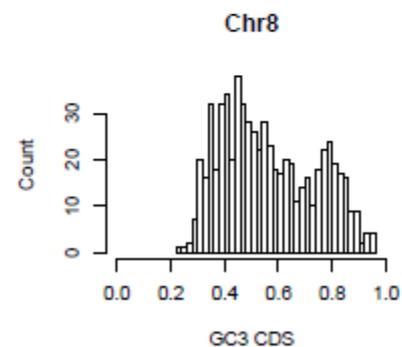
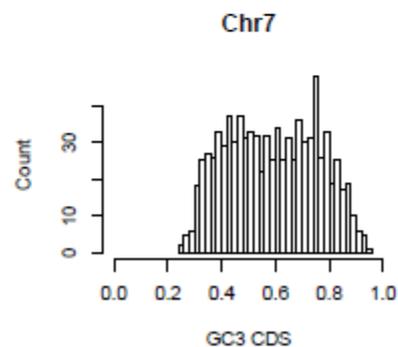
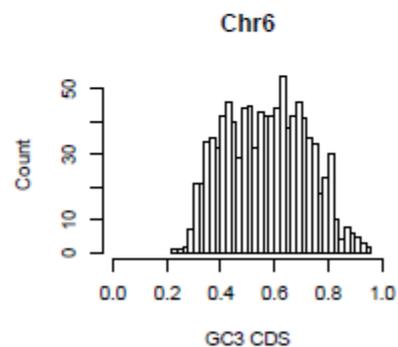
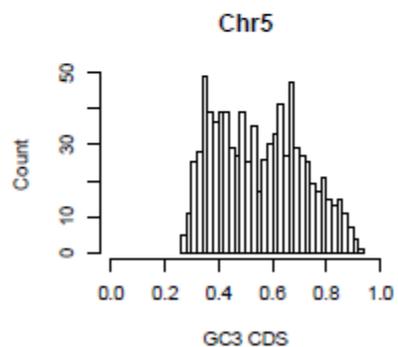
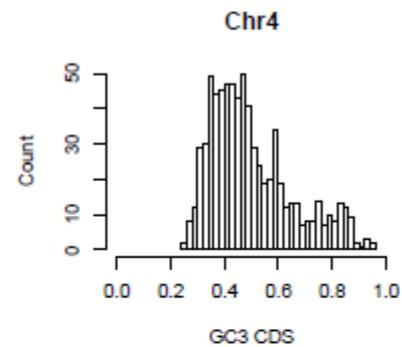
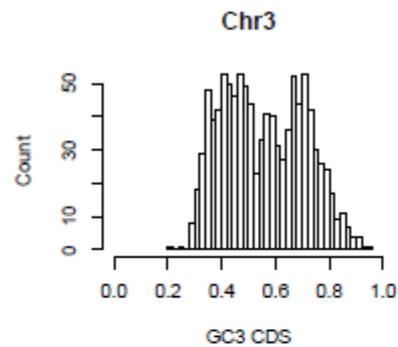
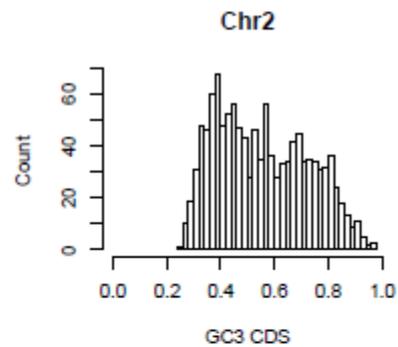
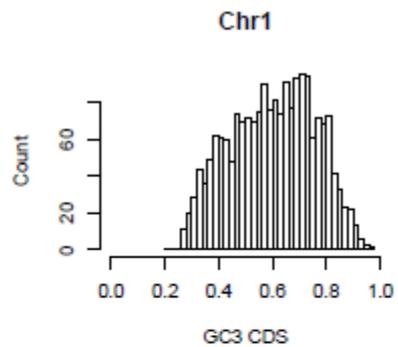
# Genes por cromosoma

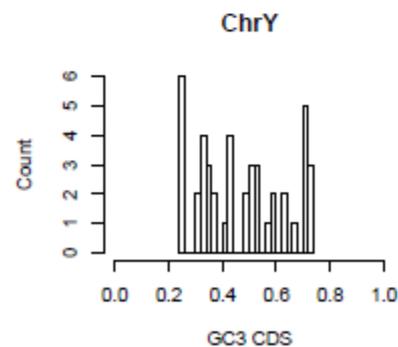
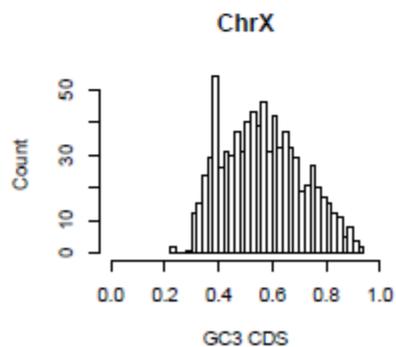
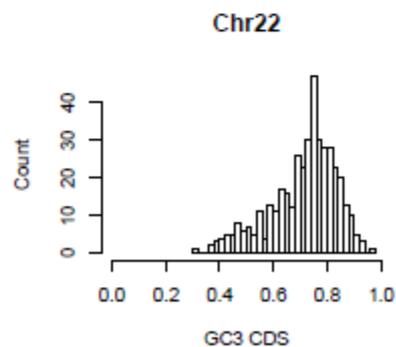
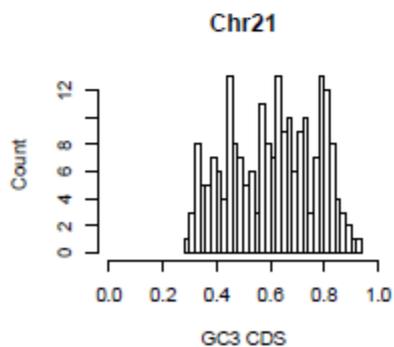
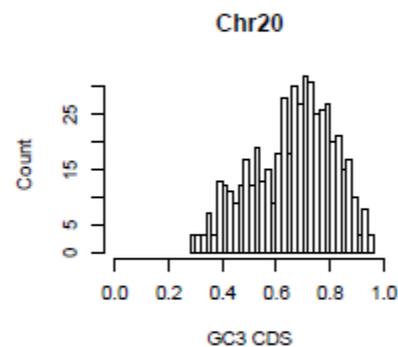
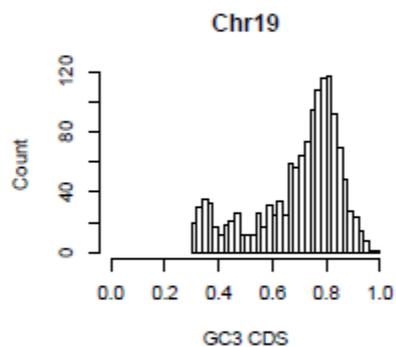
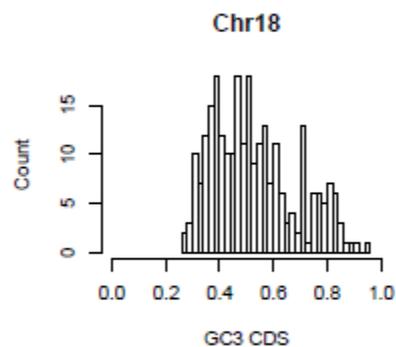
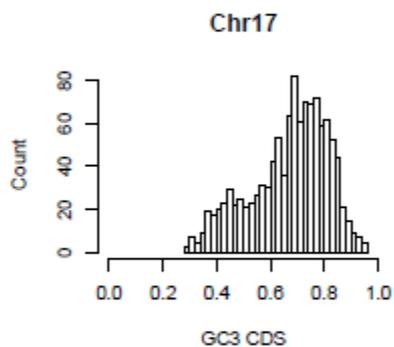
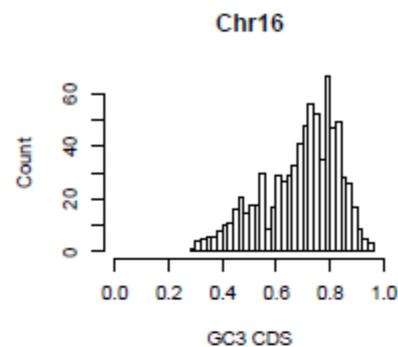
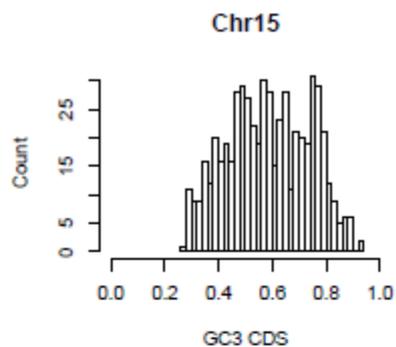
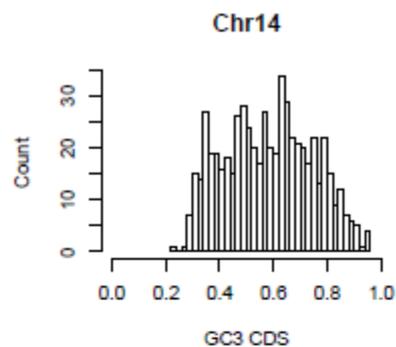
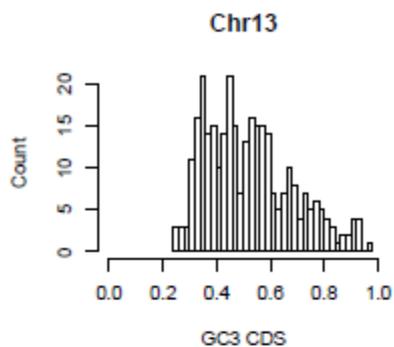
Genes per Chromosome, n=19,046



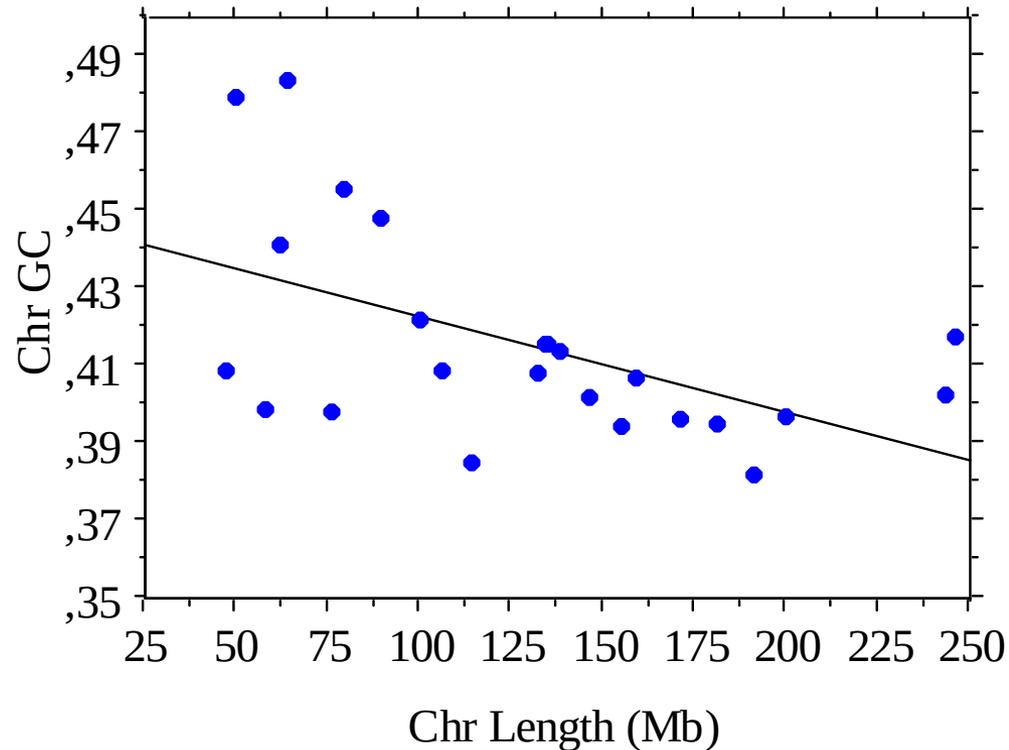
GC3 of CDS (All Chromosomes) n=19,046







# Correlación entre el contenido en GC de los cromosomas y la respectiva longitud

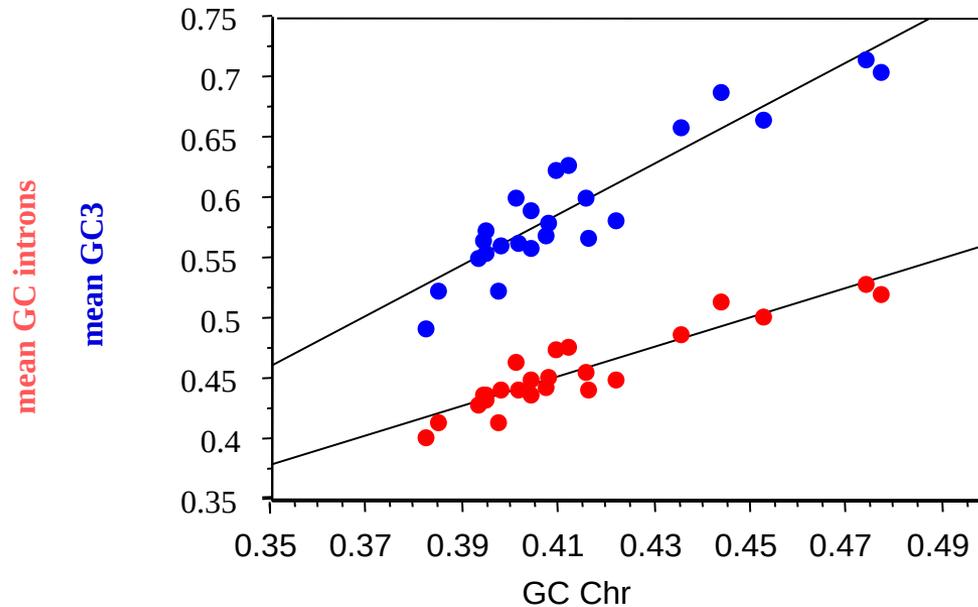


$$Y = ,447 - 2,462E-4 * X; R^2 = ,277; P = 0,008$$

# Correlaciones entre el GC de cada cromosoma y la media de GC3 y de los intrones

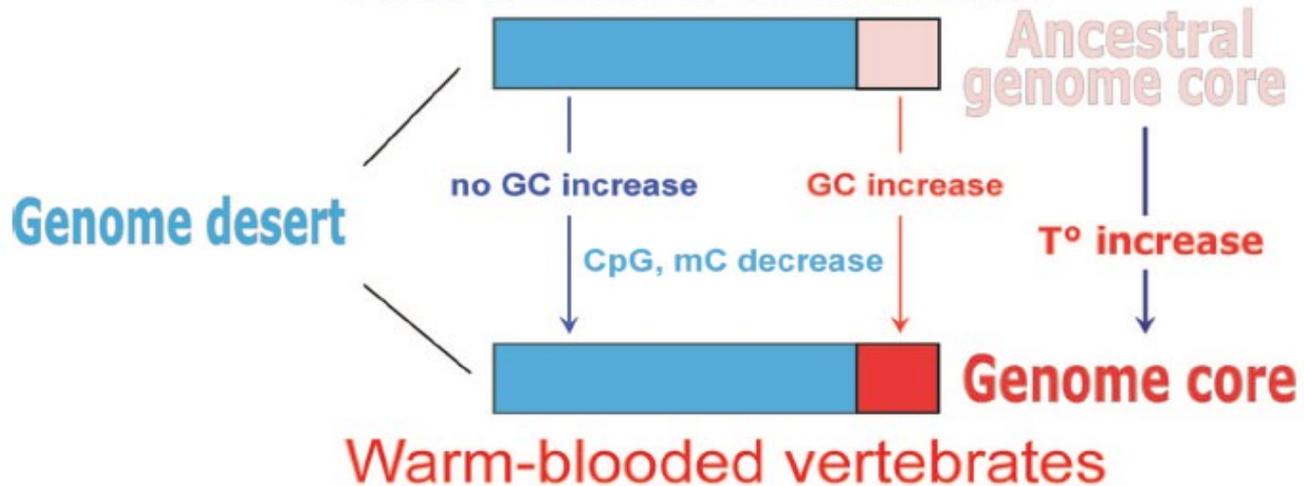
$$Y = -0.272 + 2.099 * X; R^2 = 0.849$$

$$Y = -0.049 + 1.224 * X; R^2 = 0.852$$

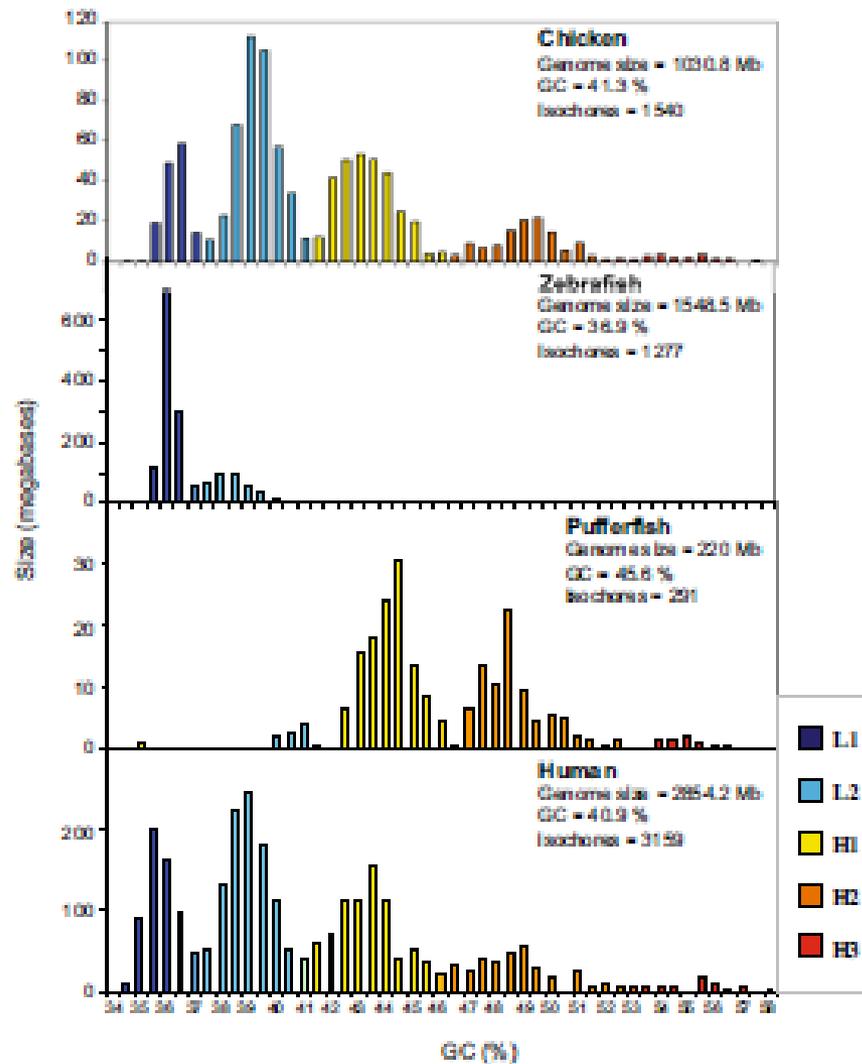


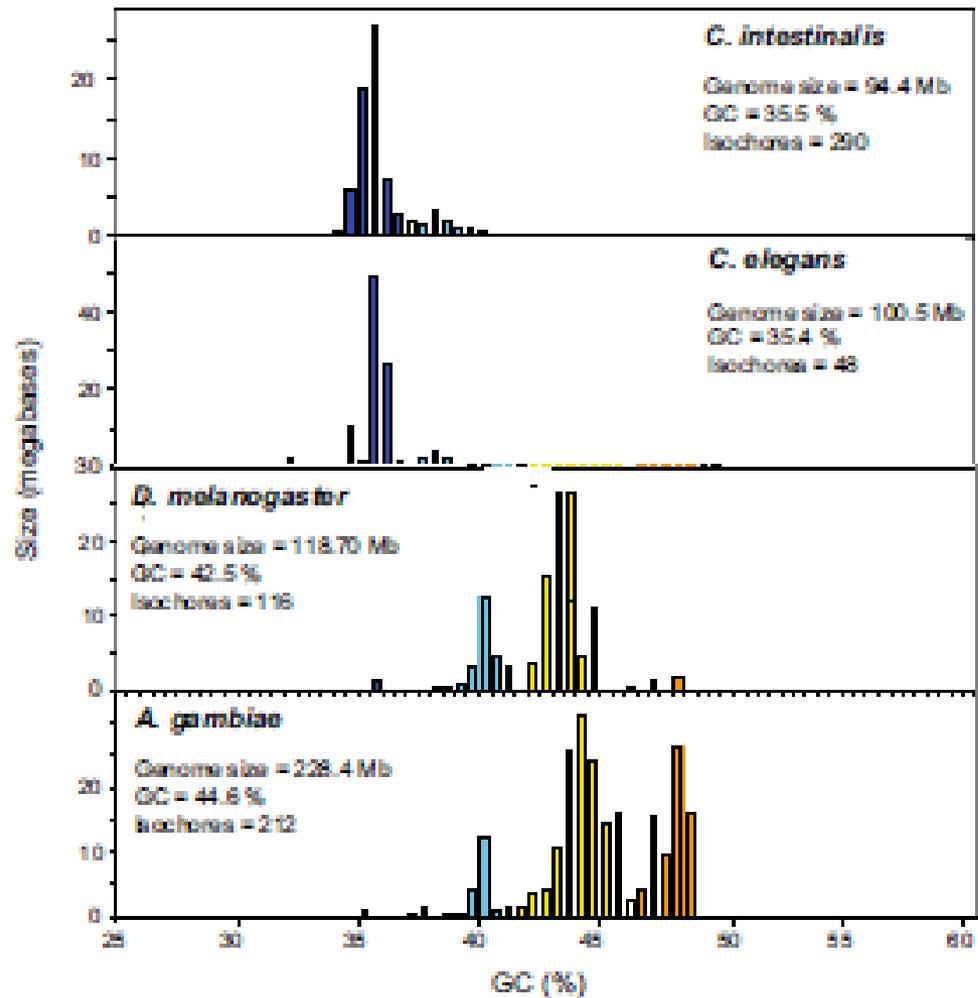
# Transitional mode

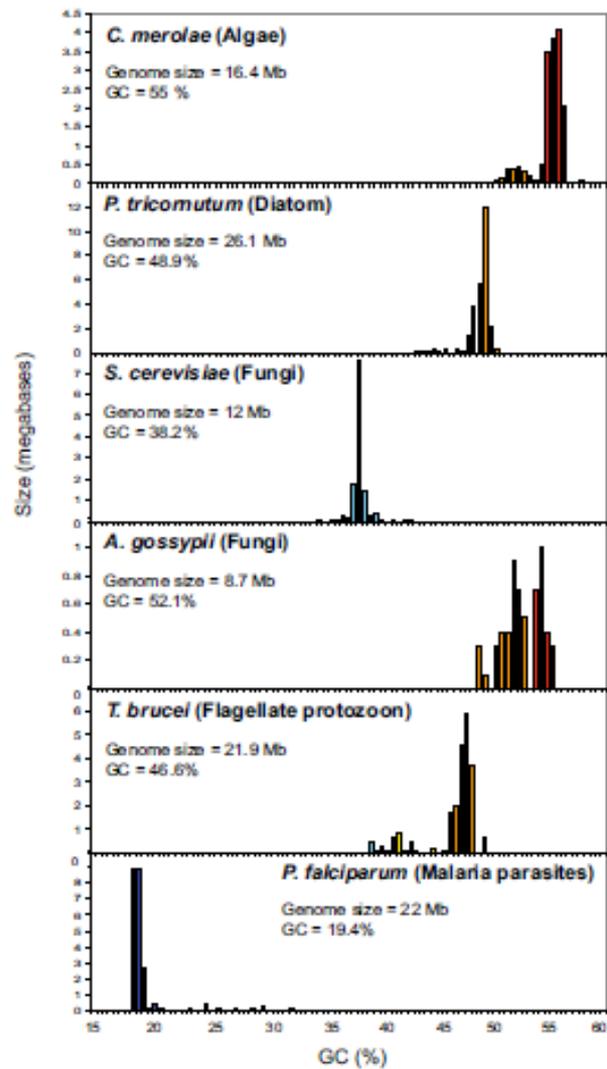
Cold-blooded vertebrates



Conservative mode

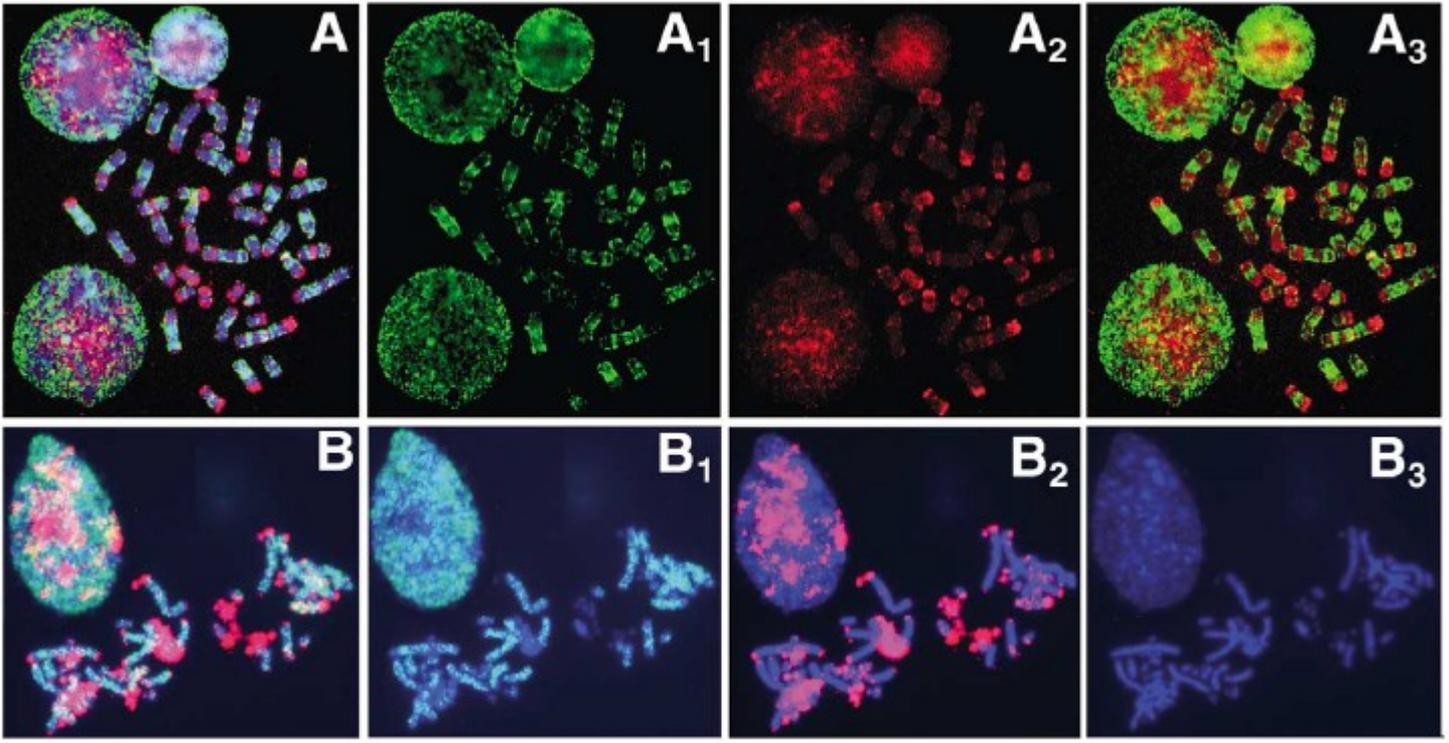




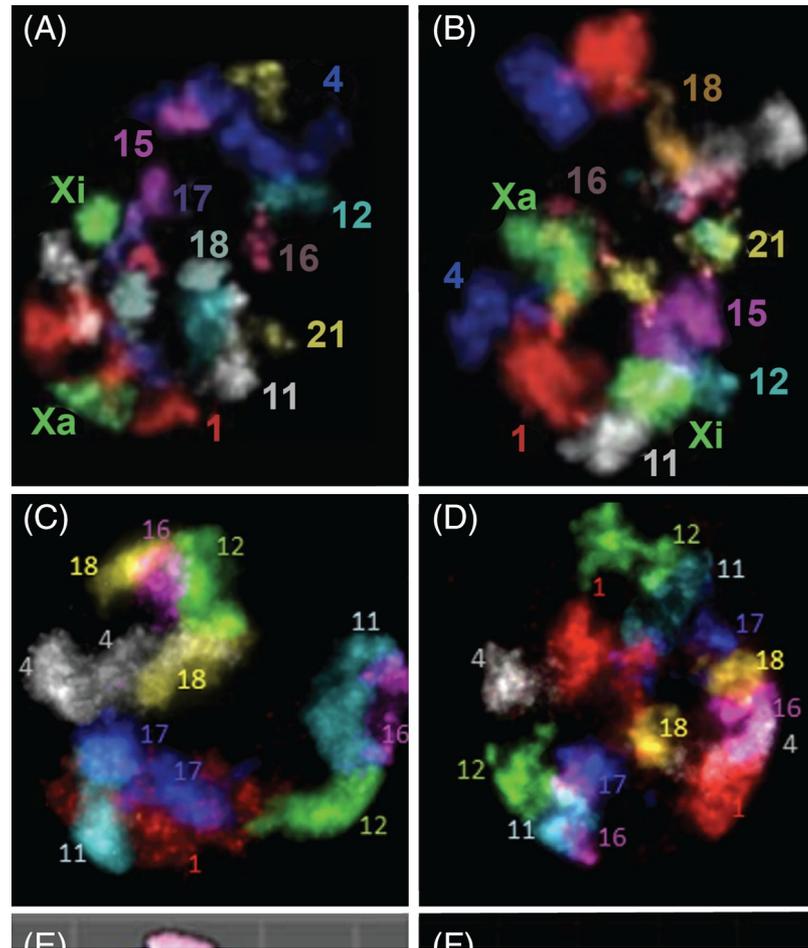


# Genoma Eucariota: estructura 3-D

El genoma de los mamíferos es un objeto de una dimensión (1-D) «metido» en un núcleo que es 400.000 veces más corto que el eje más largo de todo el ADN. Esta **restricción espacial** necesita una compactación jerárquica del genoma 1-D hasta la cromatina 3-D dentro del núcleo.



# Territorios cromosómicos en humano (FISH)



# Origen isocoros

- a) Teoría seleccionista: aumento de GC como adaptación a la mayor temperatura de mamíferos y aves.
- b) Teoría neutralista: biased gene conversion

# Origen isocoros

**Importante: el sesgo mutacional de los mamíferos, en todos los isocoros, es hacia AT... entonces???**

**Además, como vimos, tienen distintas propiedades funcionales y estructurales los isocoros ricos y pobres en GC... ¿Actúa la selección natural?**