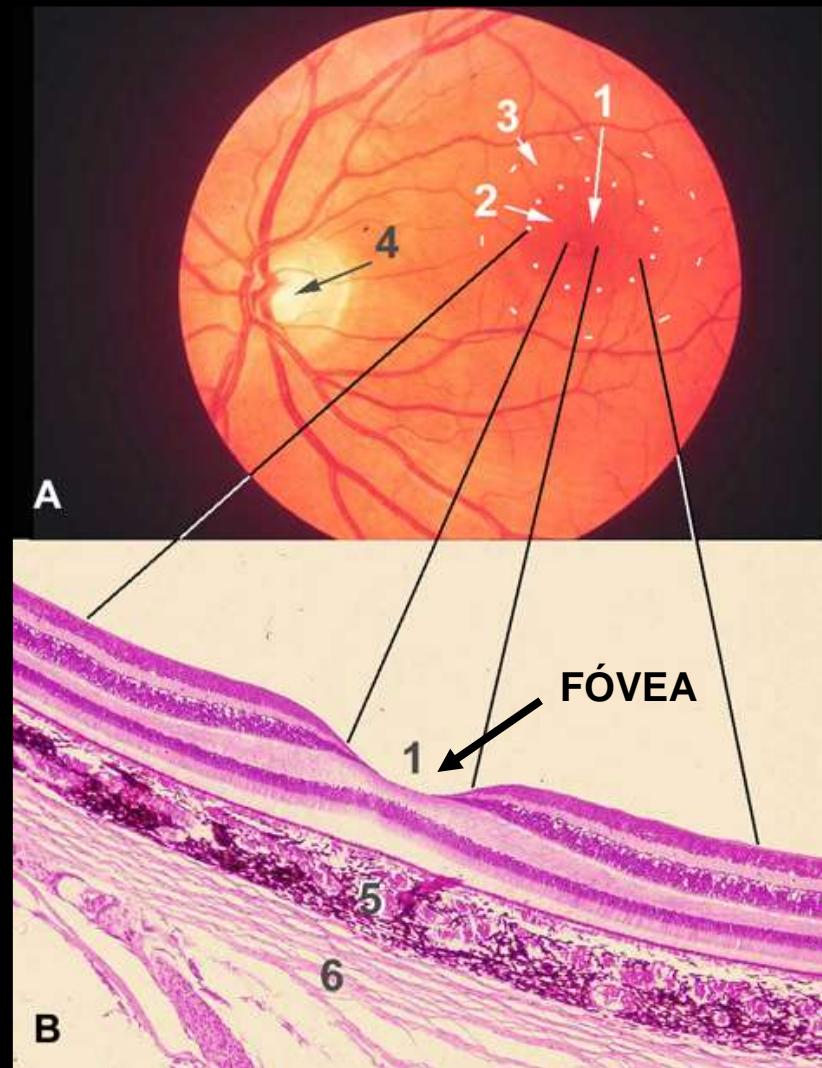
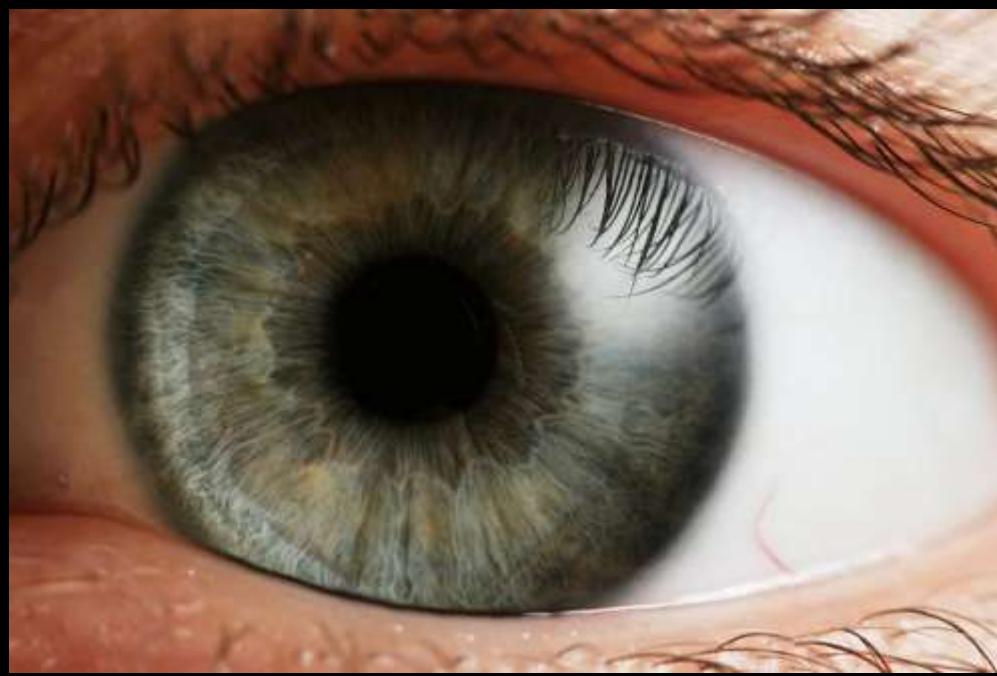


F B K G M S E H U Q T A P D R N Z X D O H C J Q V S Y

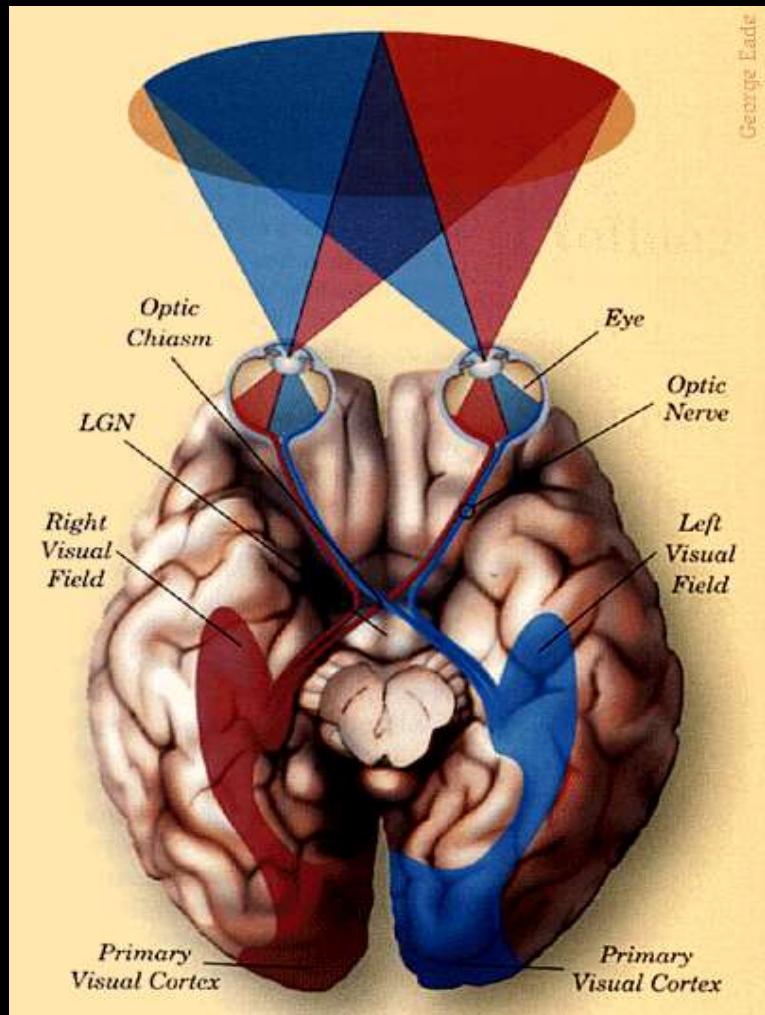
C M G Q W B F C P D L O Y

P R H





Albinismo







Pepe

Elena



Bea



Fatou



Ablaye



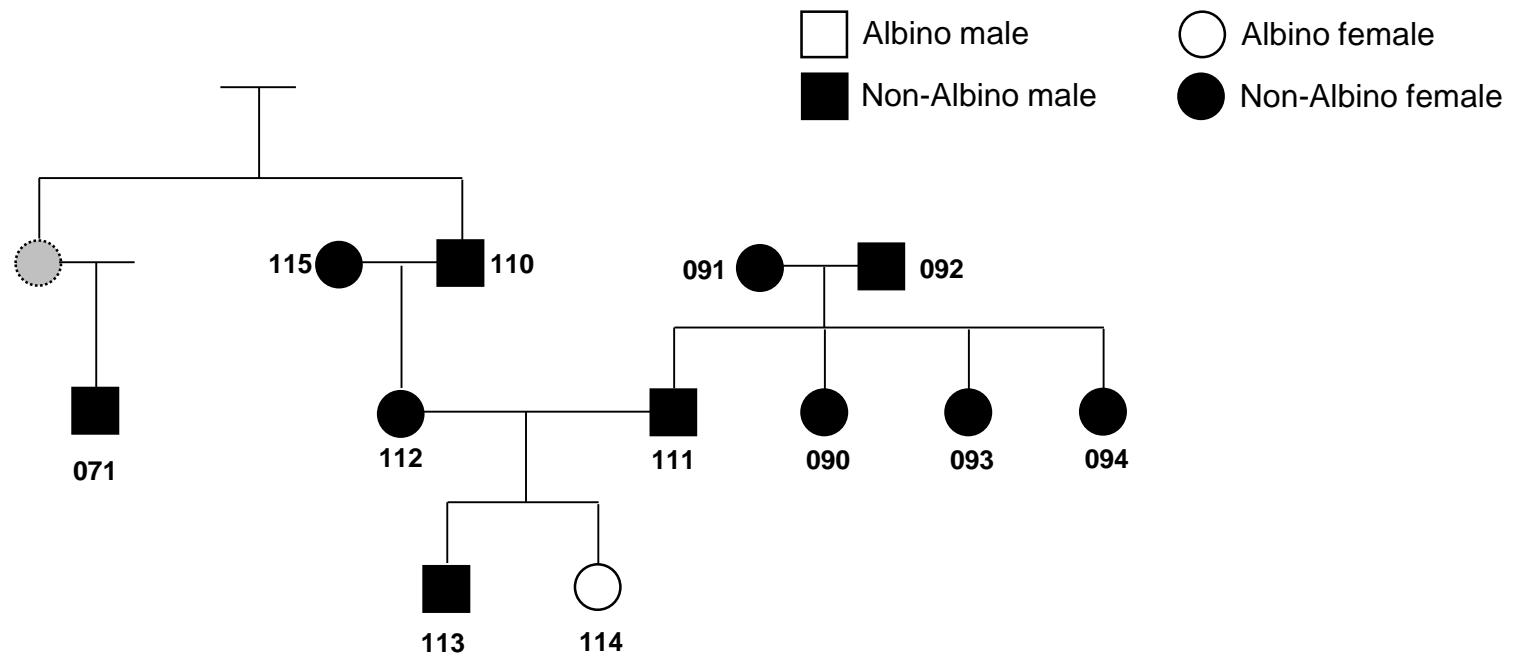
Ana



Luis Carlos

<i>TYR</i>	OCA1
<i>OCA2</i>	OCA2
<i>TYRP1</i>	OCA3
<i>SLC45A2</i>	OCA4
<i>4q24</i>	OCA5
<i>SLC24A5</i>	OCA6
<i>C10orf11</i>	OCA7
<i>GPR143</i>	OA1
<i>SLC38A8</i>	FHONDA
<i>LYST</i>	CHS1
<i>HPS1</i>	HPS1
<i>AP3B1</i>	HPS2
<i>HPS3</i>	HPS3
<i>HPS4</i>	HPS4
<i>HPS5</i>	HPS5
<i>HPS6</i>	HPS6
<i>DTNBP1</i>	HPS7
<i>BLOC1S3</i>	HPS8
<i>BLOC1S6</i>	HPS9
<i>AP3D1</i>	HPS10

20 tipos de albinismo



ALBINOCHIP

Diagnóstico Genético de albinismo

Un proyecto de investigación

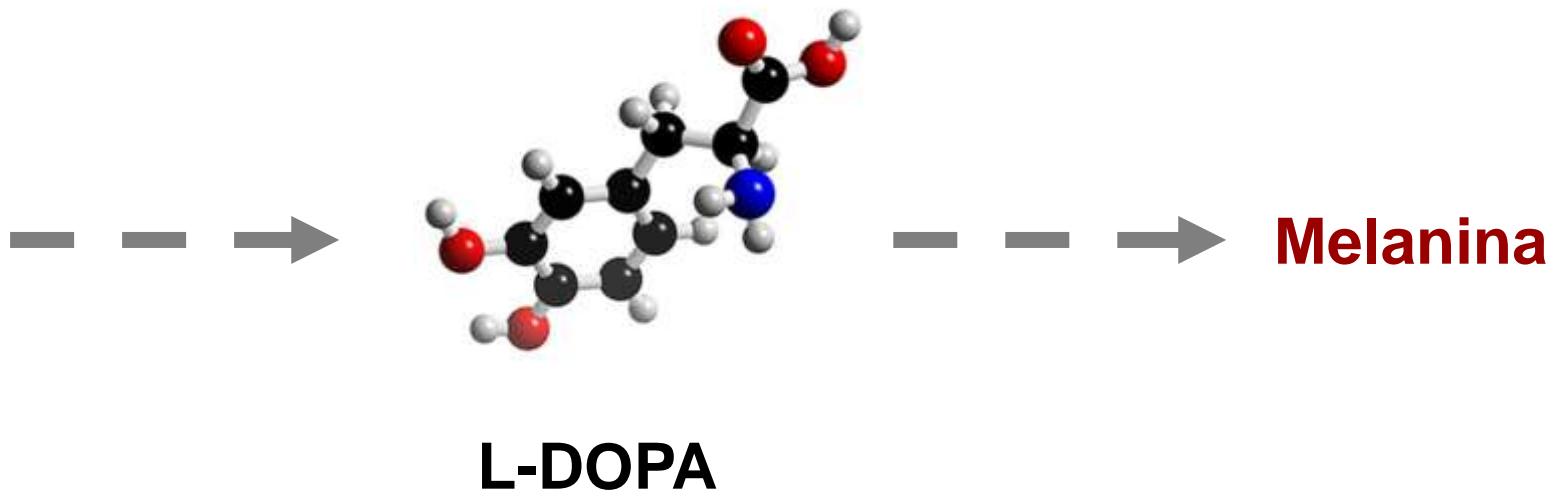


Modelos animales de albinismo





Modelos animales de albinismo



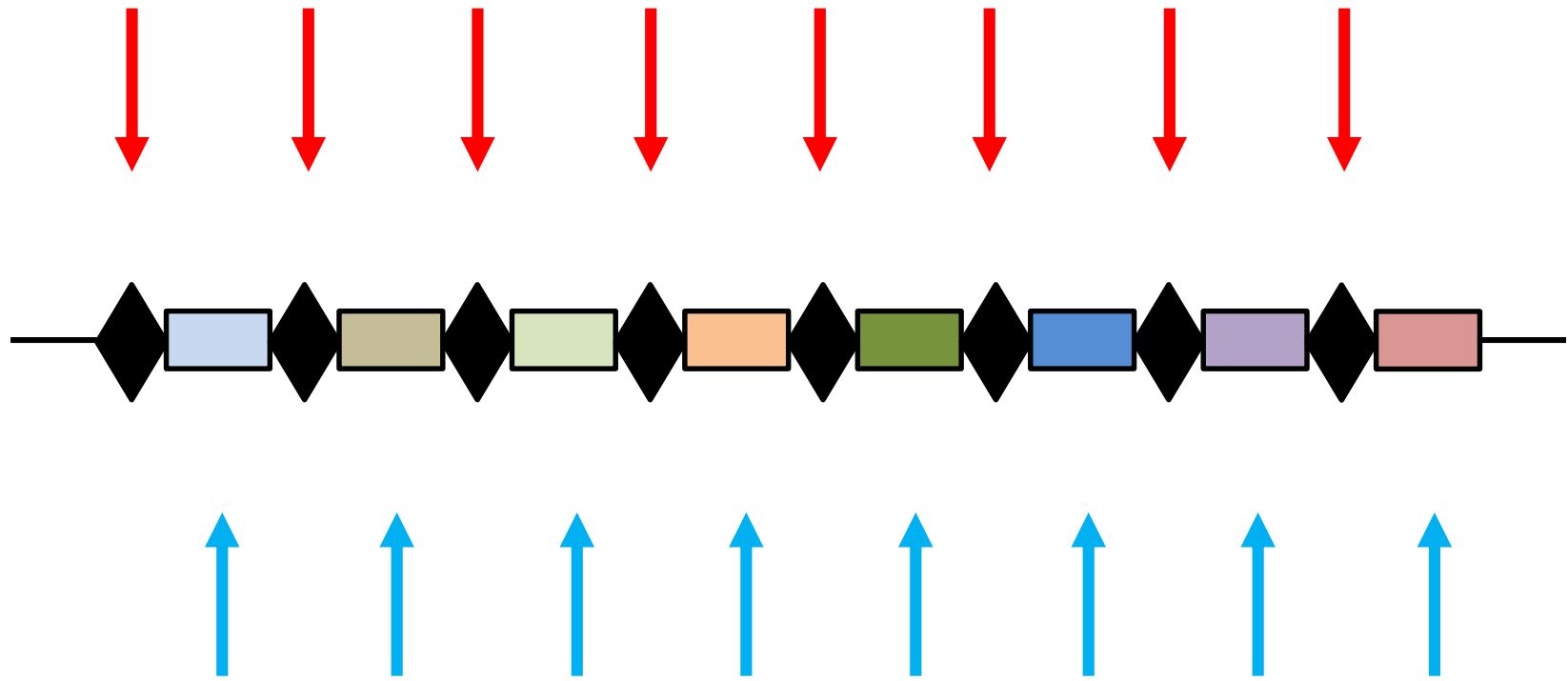




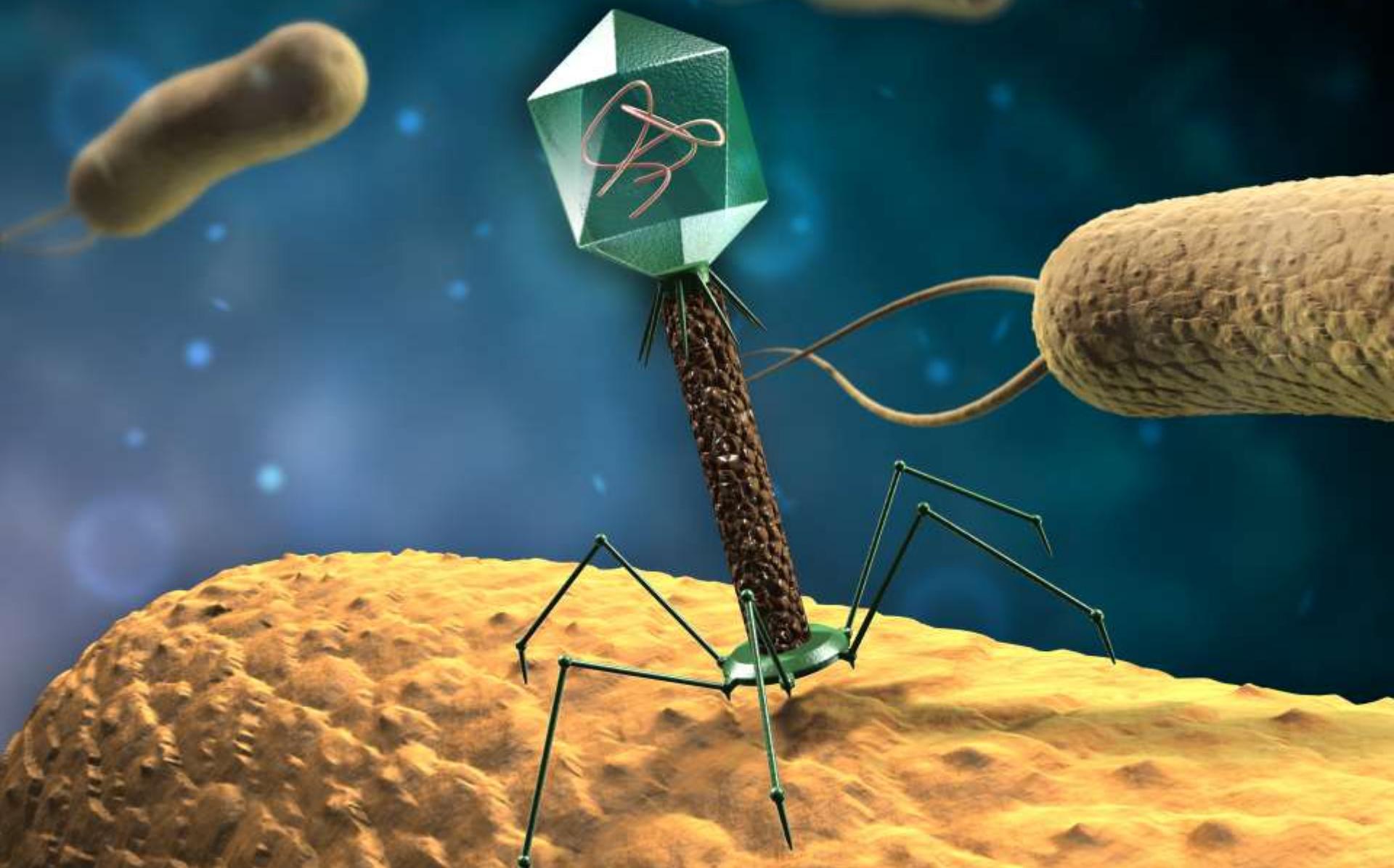
Francisco Juan Martínez Mojica



Salinas de Santa Pola (Alicante)



CRISPR





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Philippe Horvath
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Vilnius University, Lithuania



Feng Zhang
BROAD-MIT, Cambridge, MA, USA



George Church
Harvard Med School, Boston, MA, USA

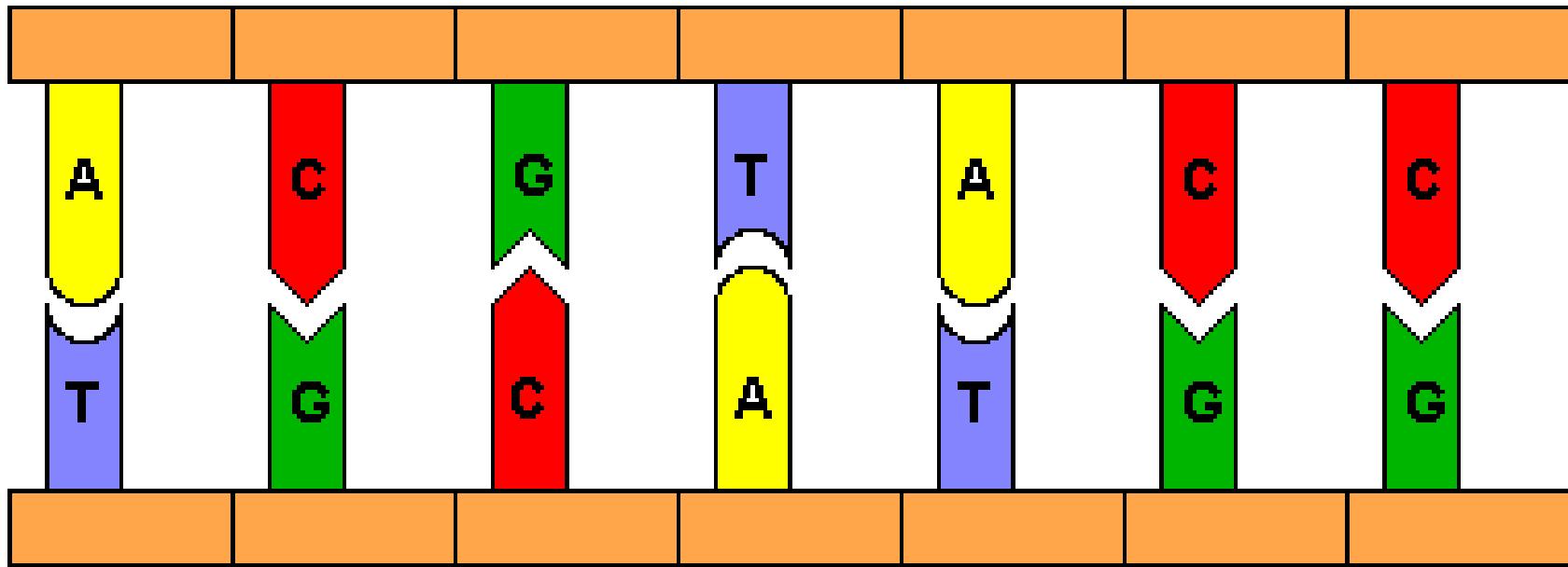


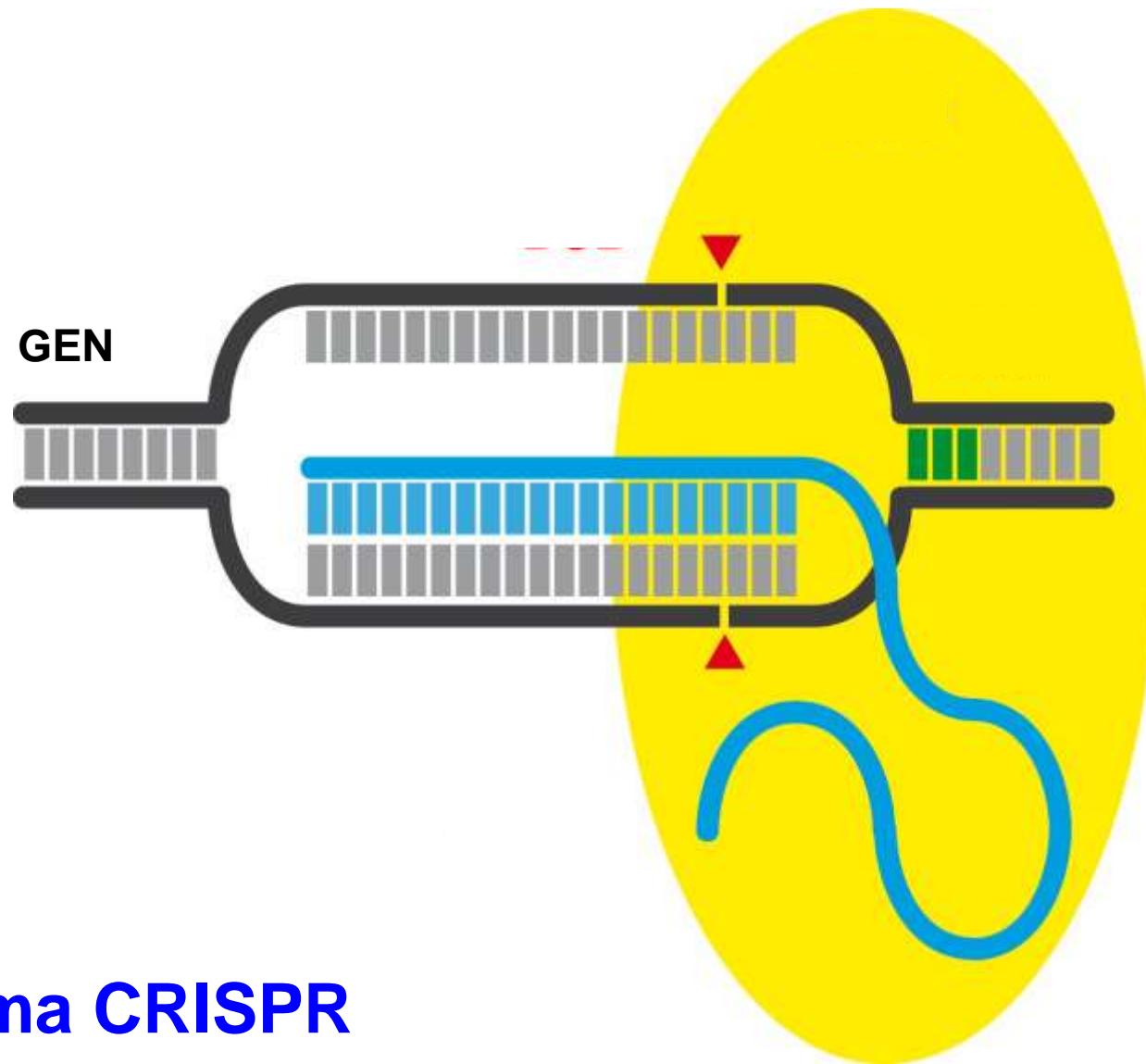
Rudolf Jaenisch
Whitehead Inst, Cambridge, MA, USA



J. Keith Joung
Mass Gen Hosp, Charlestown, MA, USA

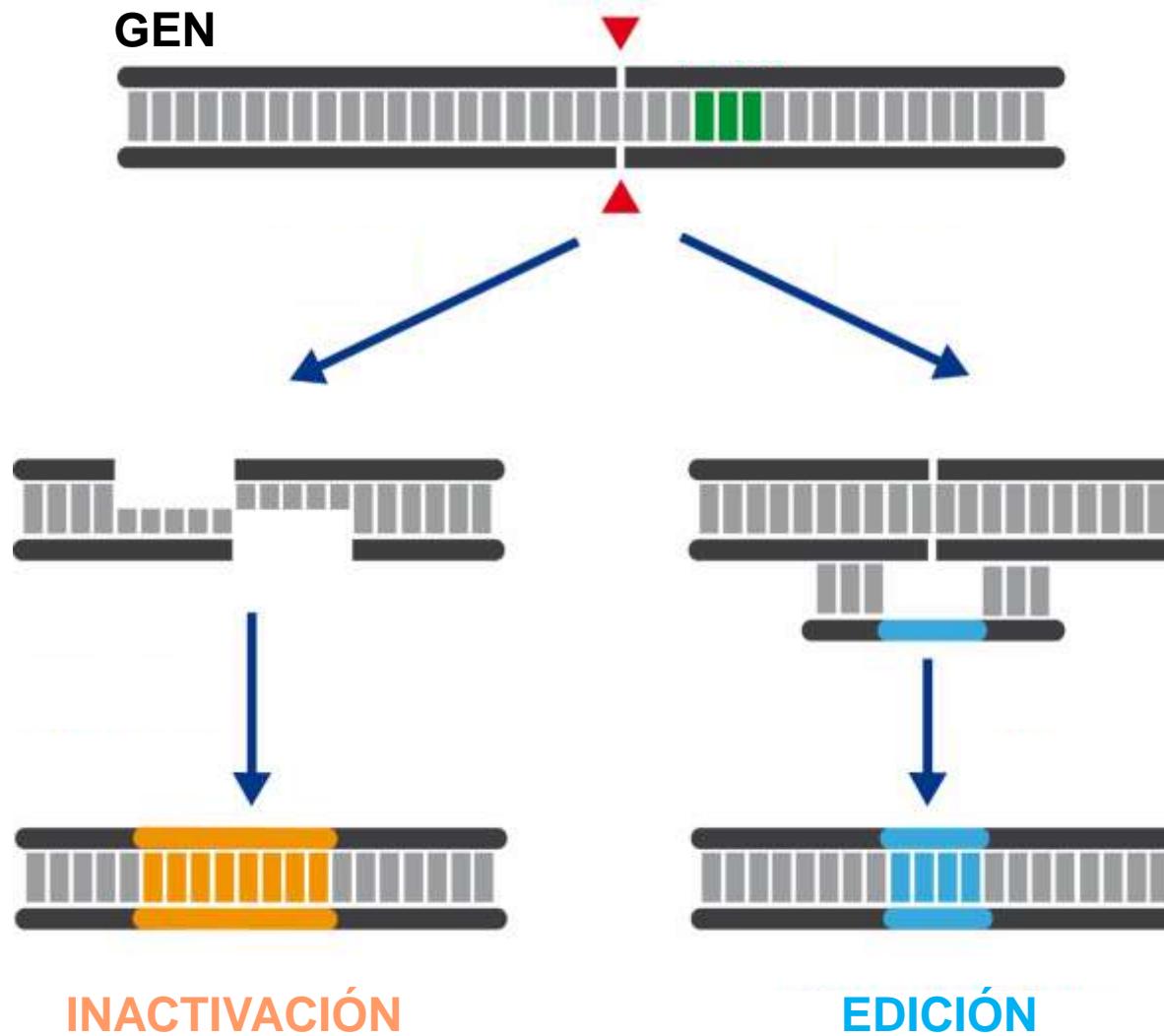






El sistema CRISPR

¿Cómo funcionan las CRISPR?



Con CRISPR podemos...

- **Mutar** un gen
- **Corregir** una mutación de un gen
- **Editar** un gen, cambiar una o varias letras
- **Eliminar** un trozo de un gen
- **Añadir** un fragmento nuevo a un gen
- **Marcar** un gen
- ...



CRISPR

Ratones avatar





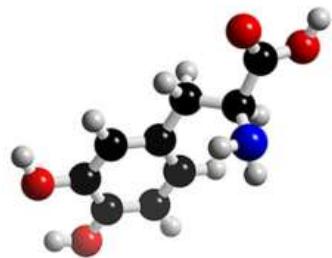
Pepe

OCA1



Patty

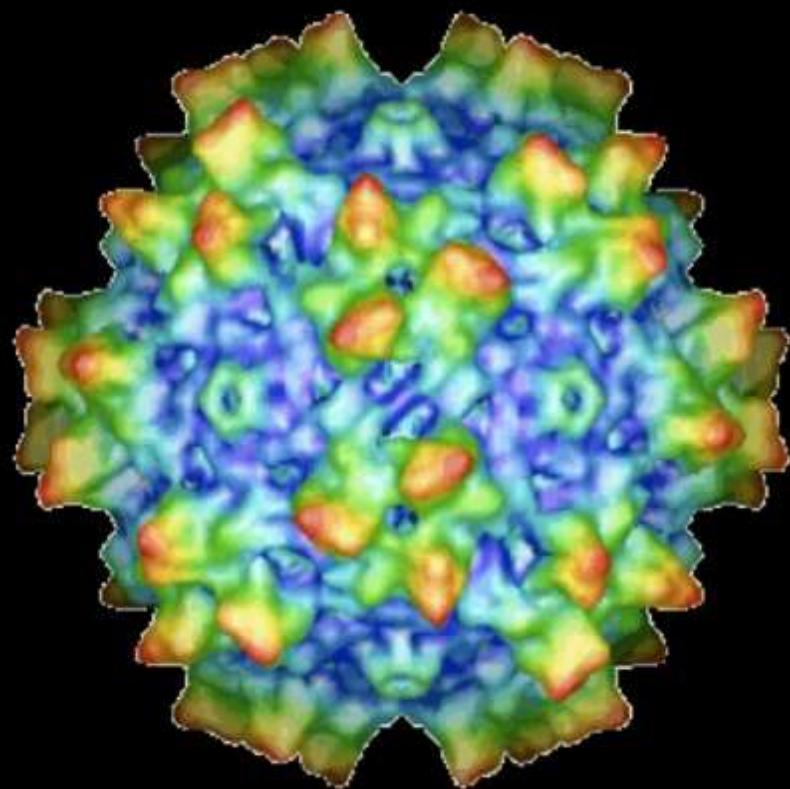
OCA4



Tratamientos



CRISPR



Enfermedades raras tratadas con CRISPR en animales

- Distrofia Muscular de Duchenne (DMD)
- Deficiencia en Ornitina transcarbamila (OTC)
- Tirosinemia Hereditaria de tipo I (FAH)
- Cataratas congénitas (CRYGC)
- Enfermedad Crónica Granulomatosa (CGD)
- Retinosis pigmentaria (RP)
- Amaurosis Congénita de Leber (LCA)
- Enfermedad de Huntington (HD)
- ...



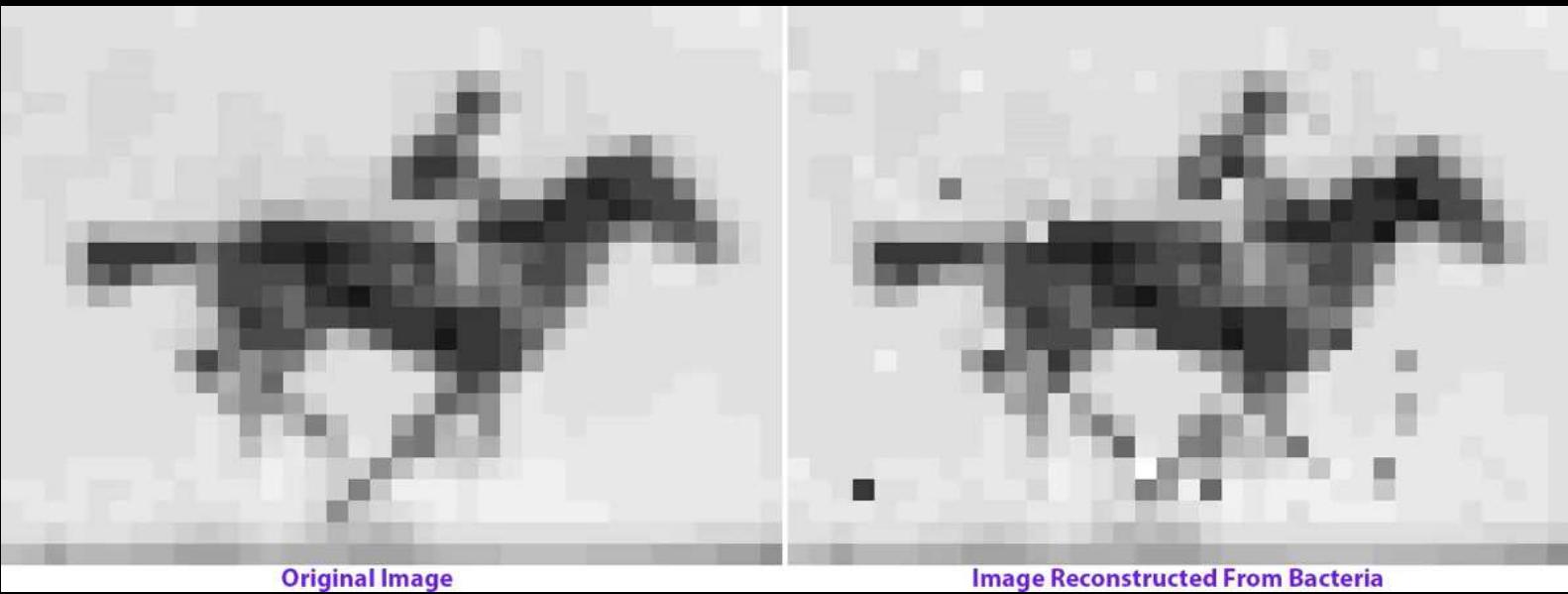
PROBLEMA

Múltiples correcciones



EVERWHERE

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GOBIERNO
DE ESPAÑA

MINISTERIO
DE ECONOMÍA, INDUSTRIA
Y COMPETITIVIDAD

ciberer isciii



EMMA
mouse repository



INFRAFRONTIER
mouse disease models