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NOVEDADES CIENTÍFICAS EN INV-DUP 15

Dra. Asun Díaz Gómez
Unidad de Neuropediatría
Hospital de la Santa Creu i Sant Pau

31 de mayo de 2025

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Ciencia y tecnología

Tratan con éxito a un bebé con una enfermedad rara a través de una terapia genética CRISPR "diseñada especialmente para él"

Es el primer caso de una terapia personalizada para un bebé con una enfermedad ultrarrara y severa. Algunos expertos alaban el método pero piden una reflexión y aseguran que es "ética y económica e incluso científicamente cuestionable"

MEDICINA CURATIVA

CURAR CON LOS PROPIOS GENES: LA REVOLUCIÓN SILENCIOSA DE LA TERAPIA GÉNICA

Cada vez han más datos que demuestran los beneficios de la terapia génica, especialmente en enfermedades raras en los que hay pocas opciones

→ ABC → **Salud**

El primer tratamiento con edición genética supera su primer examen: «Es una esperanza para muchas enfermedades genéticas»

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~1 de cada **10** personas tienen una Enfermedad Rara

Representando en América Latina
50 millones de personas afectadas

50-70%
inician síntomas en la infancia

Solo el **5%** tienen un tratamiento aprobado por la **FDA**

Se requieren más de **9 años** para llegar al diagnóstico correcto

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AVANCES EN EL DIAGNÓSTICO DE LAS ENFERMEDADES GENÉTICAS

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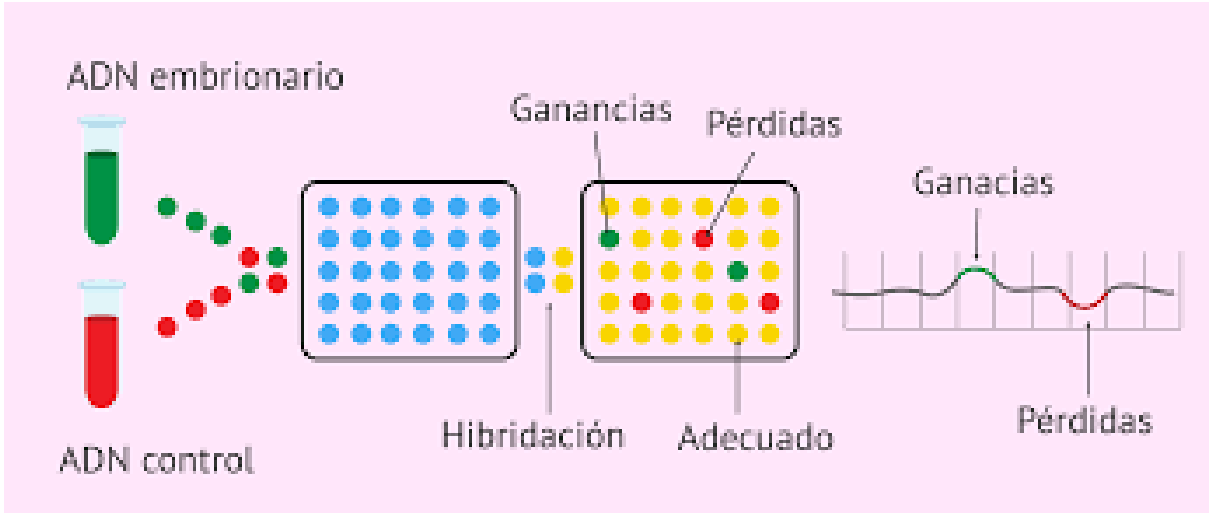

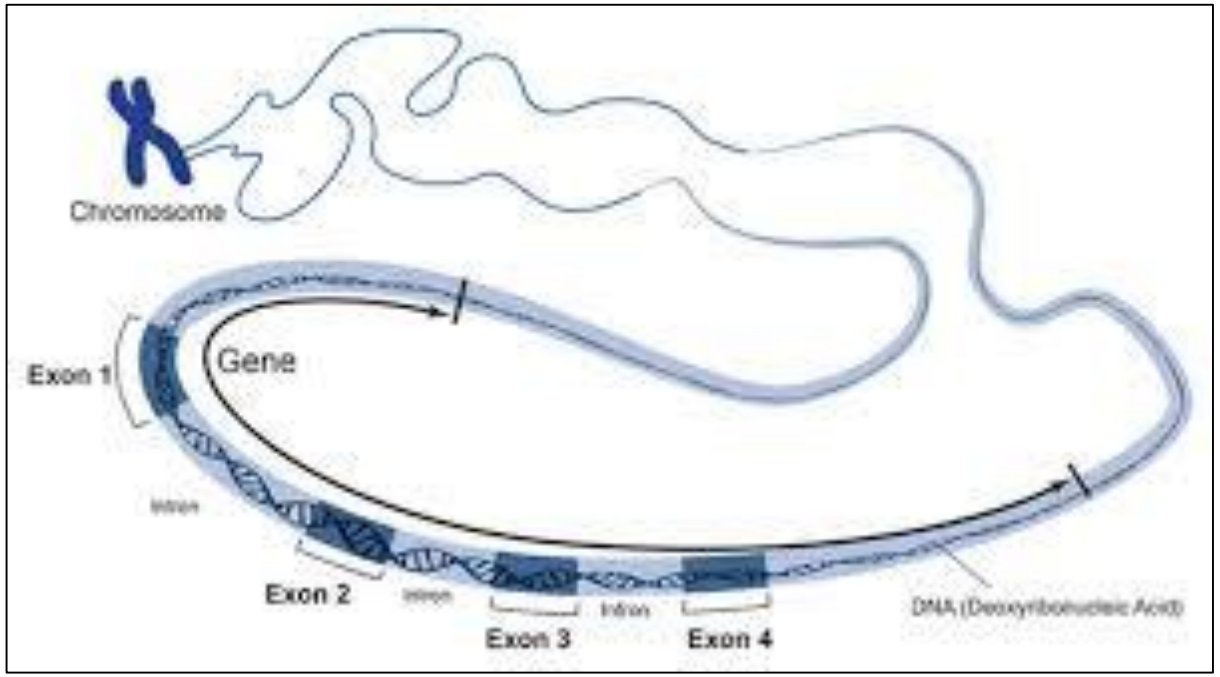

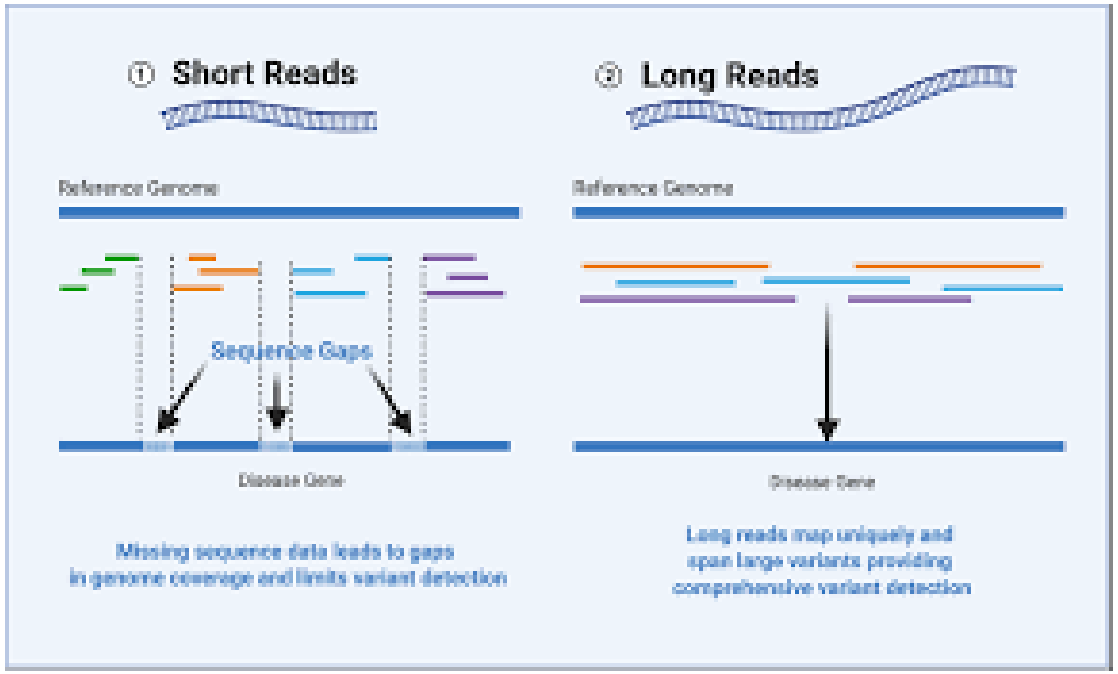

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AVANCES EN EL TRATAMIENTO DE LAS ENFERMEDADES GENÉTICAS

TERAPIA DE REEMPLAZO ENZIMÁTICO

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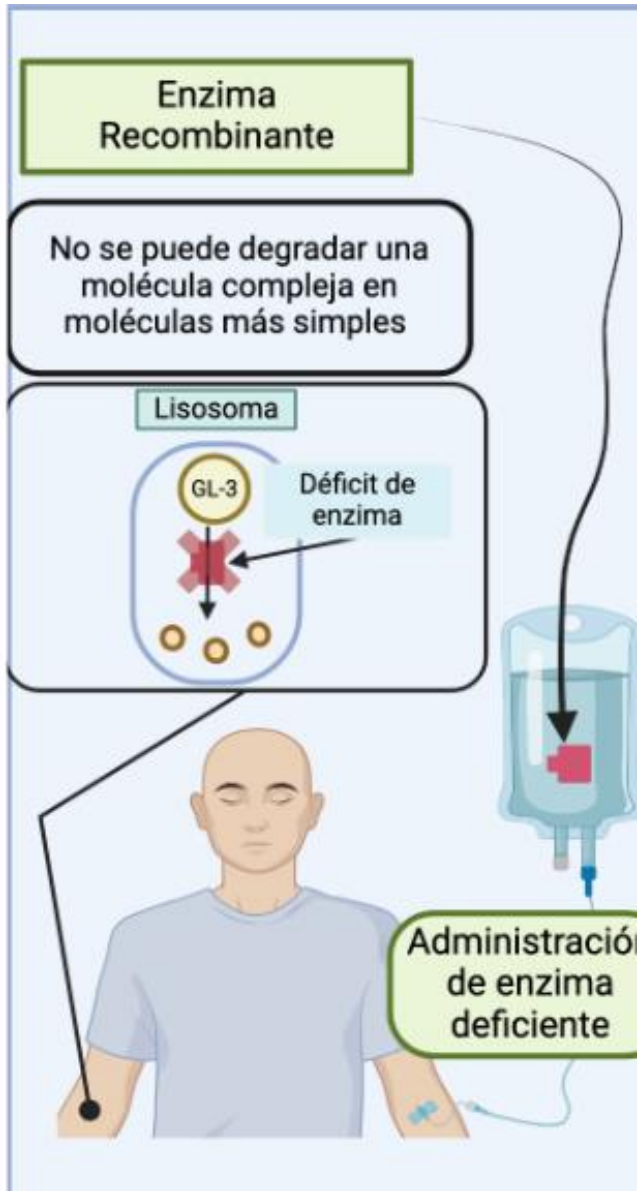

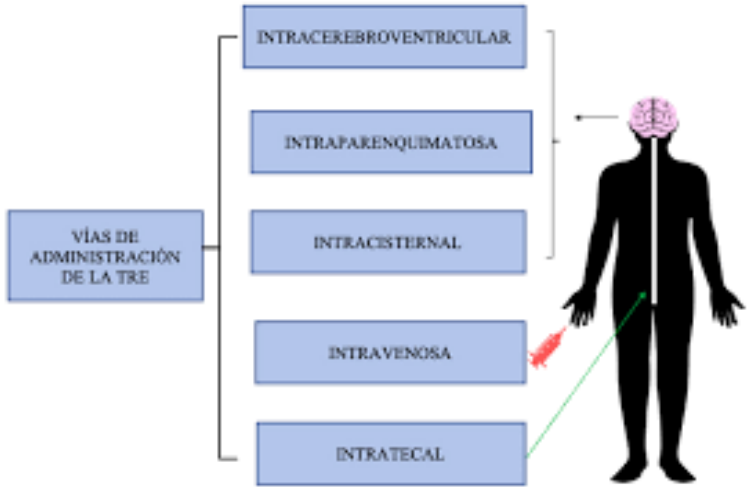

- **ENFERMEDAD DE POMPE:** alglucosidasa
- **ENFERMEDAD DE FABRY:** algilasa alfa y beta
- **ENFERMEDAD DE GAUCHER:** imiglucerasa
- **MUCOPOLISACARIDOSIS I:** laronidasa
- **MUCOPOLISACARIDOSIS II:** idursulfasa
- **MUCOPOLISACARISODIS VI:** galsufasa

TRASPLANTE DE CÉLULAS HEMATOPOYÉTICAS

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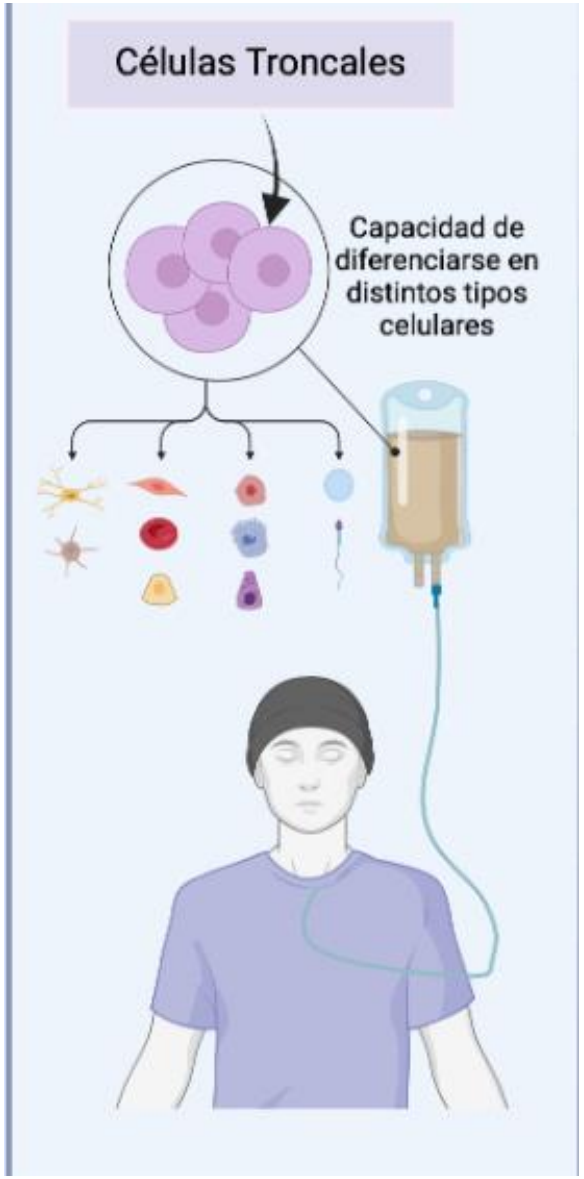

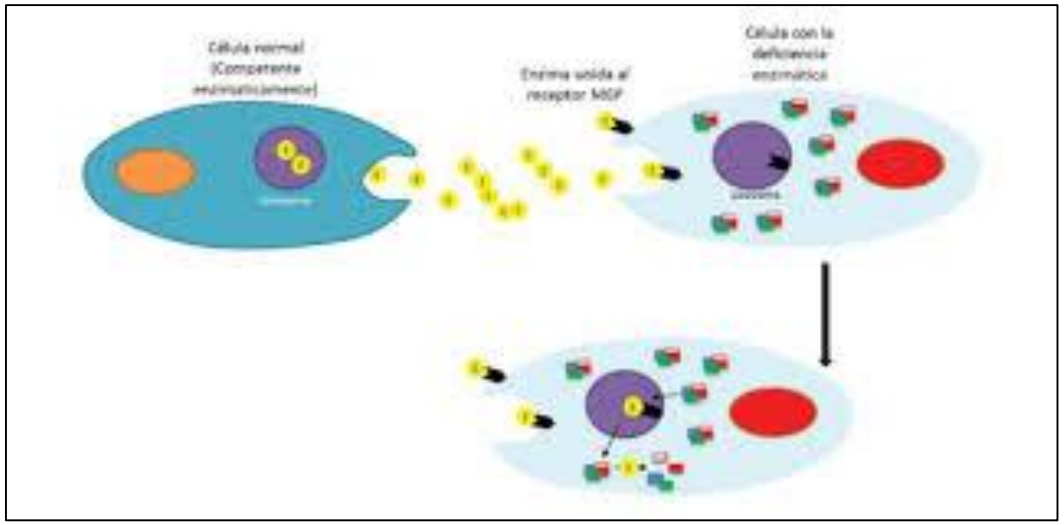

- **ADRENOLEUCODISTROFIA**
- **LEUCODISTROFIA METACROMÁTICA**

TERAPIA CON OLIGONUCLEÓTIDOS ANTISENTIDO

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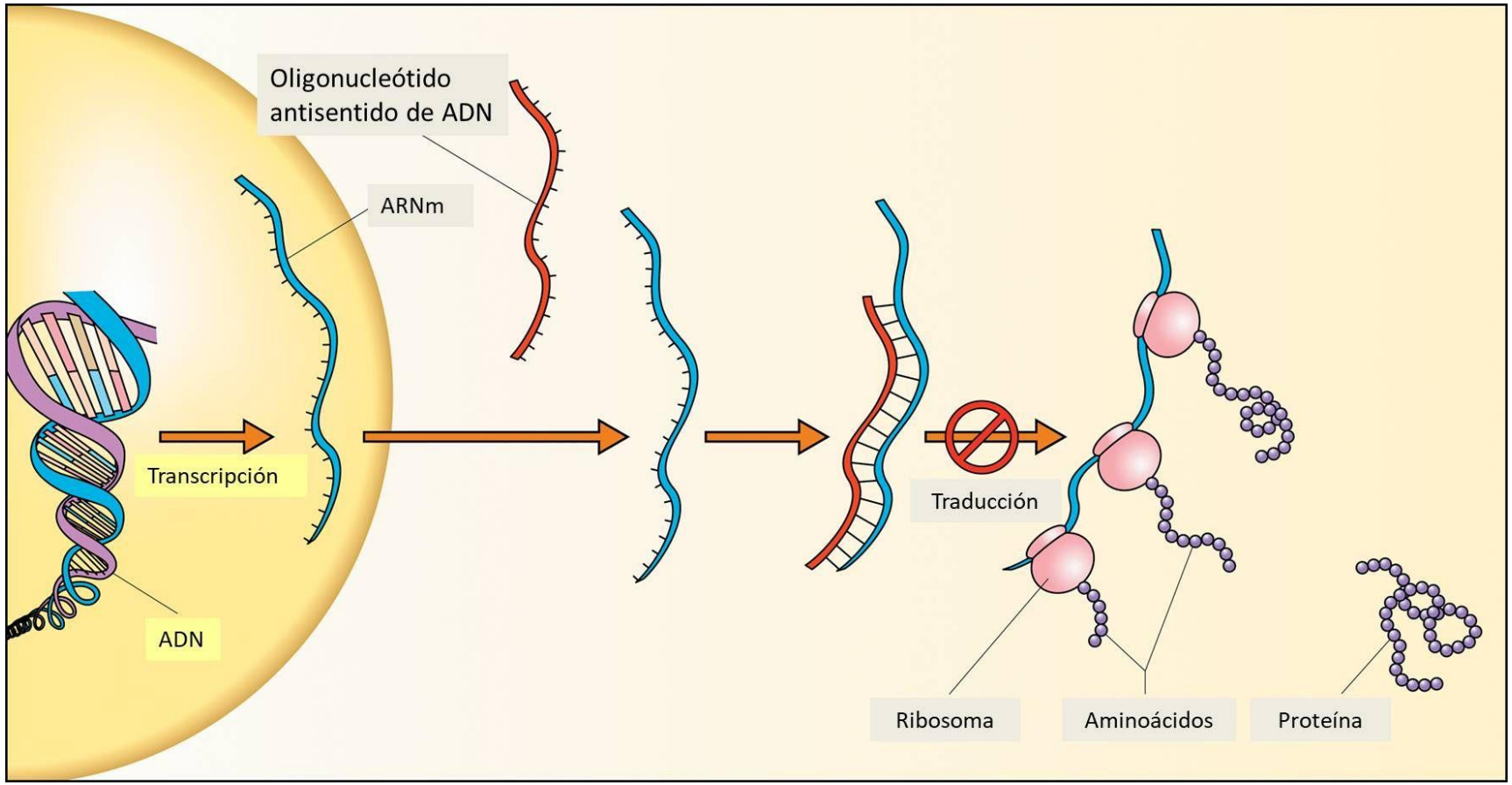

- **AME**
- **DUCHENE**

CRISPR-Cas

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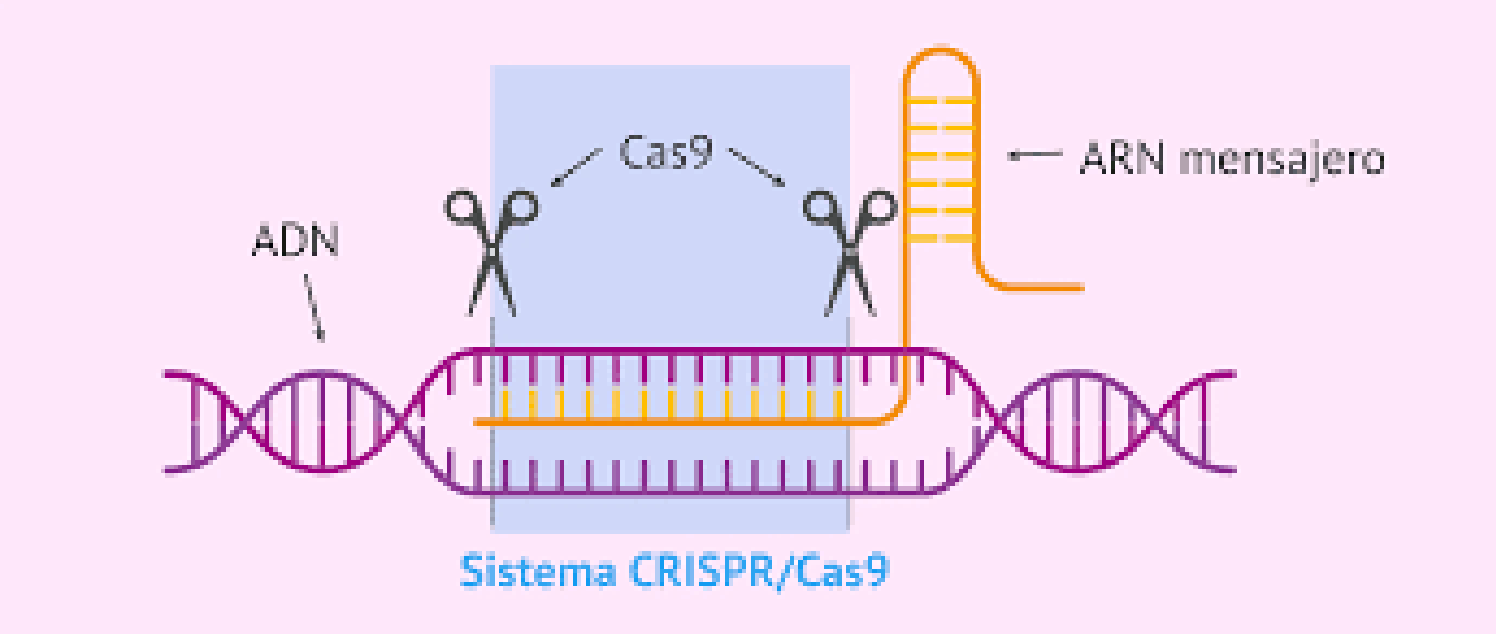

- **FIBROSIS QUÍSTICA**
- **ANEMIA DE CÉLULAS FALCIFORMES**
- **BETA TALASEMIA**
- **DÉFICIT DE CARBAMOIL FOSFATO SINTETASA 1**

REPOSICIONAMIENTO DE FÁRMACOS

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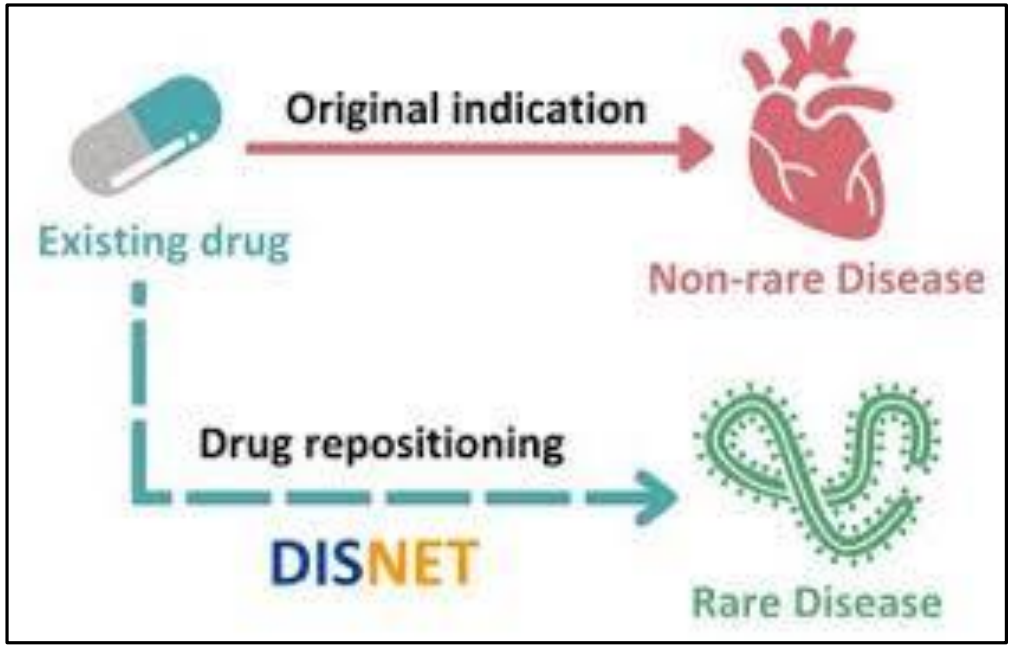
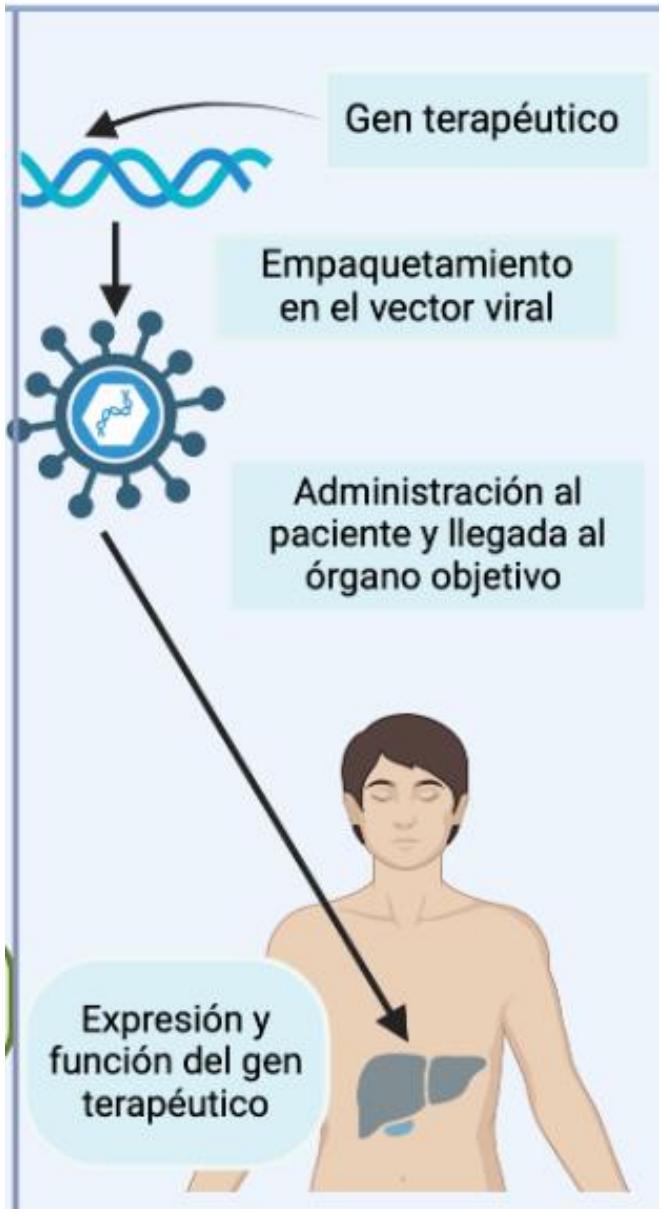
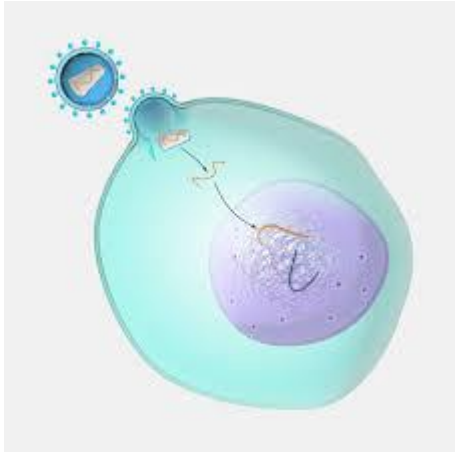


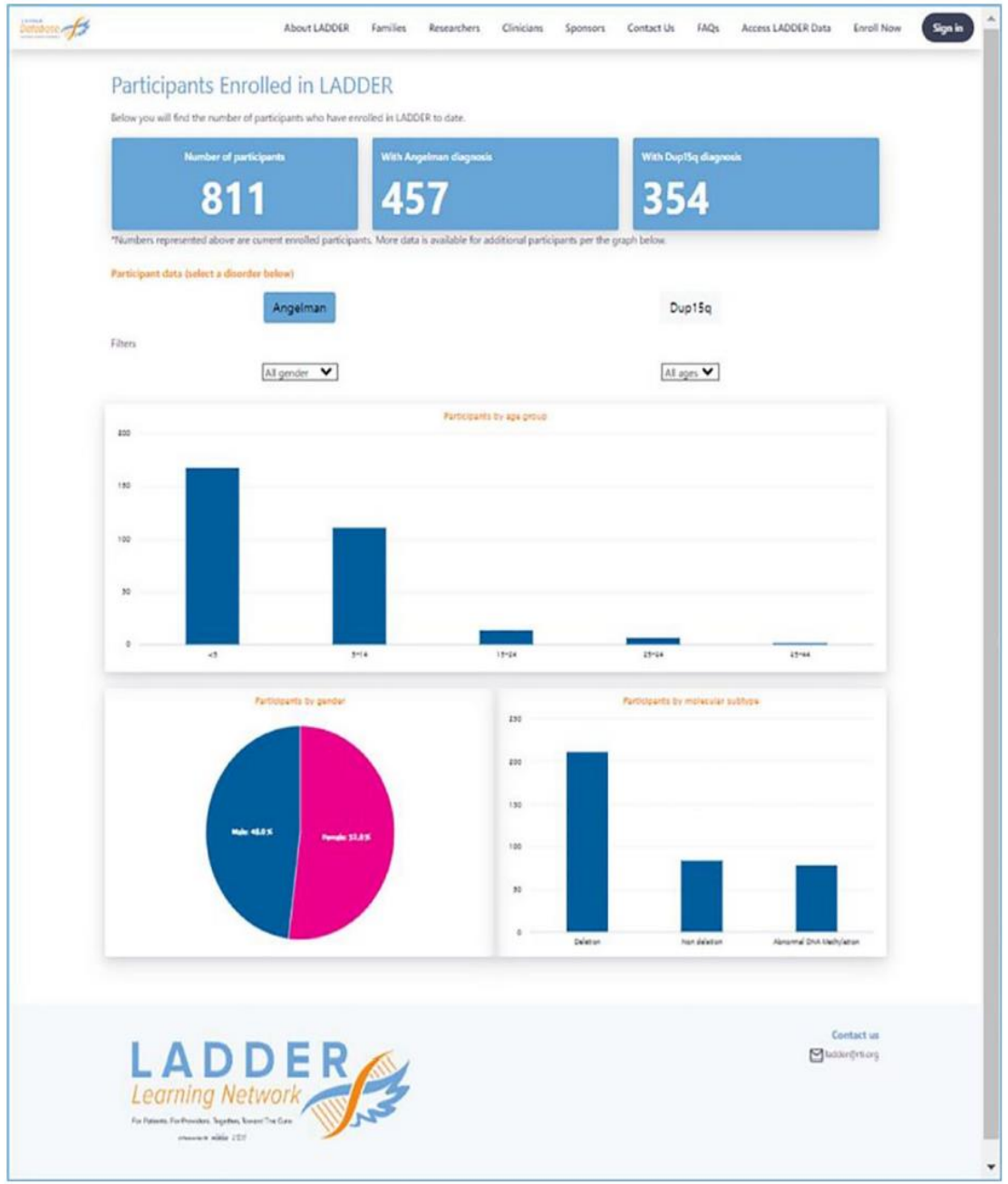
- **FENILBUTIRATO:** SLC6A1
- **GANALOXONA:** CDKL5
- **FENFLURAMINA:** Dravet
- **AMANTADINA:** Parkinson
- **CLONIDINA:**TDAH

TERAPIA GÉNICA





- **ZOLGENSMA:** Atrofia muscular espinal
- **ELEVIDYS:** Distofia muscular de Duchenne
- **LUXTURNA:** Distrofia retinina hereditaria
- **SKYSONA:** Adrenoleudistrofia
- **LIBMELDY:** Leucodistrofia metacromática



THERAPEUTIC ADVANCES in
Rare Disease

Linking Angelman and dup15q data for expanded research (LADDER) database: a model for advancing research, clinical guidance, and therapeutic development for rare conditions

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THERAPEUTIC ADVANCES in
Rare Disease

Patient Perspective

An integrated action plan to fund and support drug development for Dup15q syndrome: a patient organization perspective

Ther Adv Rare Dis

2024, Vol. 5: 1–7

DOI: 10.1177/26330402412349 32

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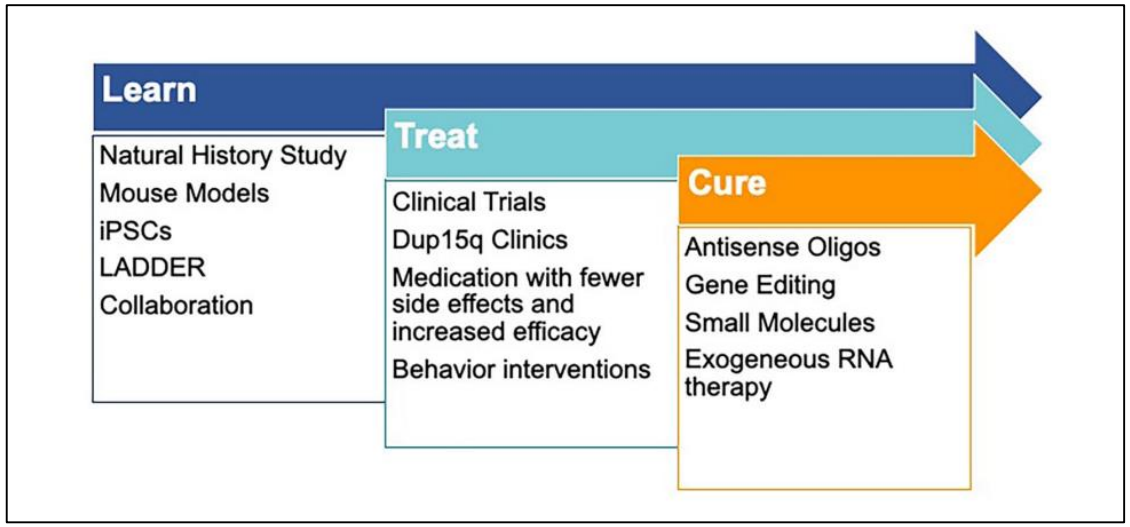
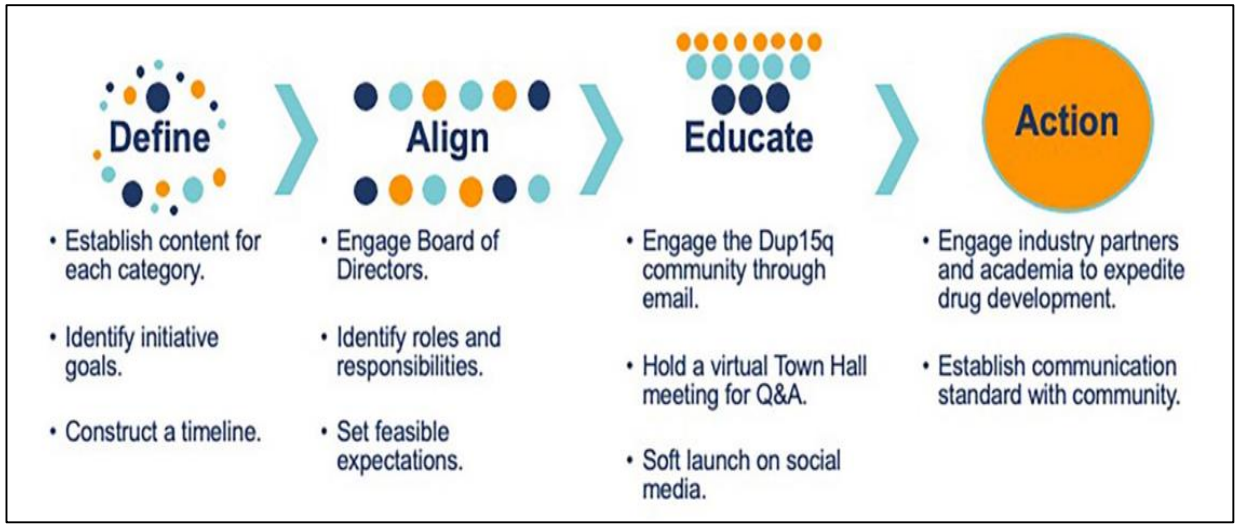
Ryan Rogers-Hammond and Carrie Howell





A CLINICAL TRIAL TO COMPARE THE EFFECTS OF BASMISANIL WITH A PLACEBO IN CHILDREN WITH DUP15q SYNDROME

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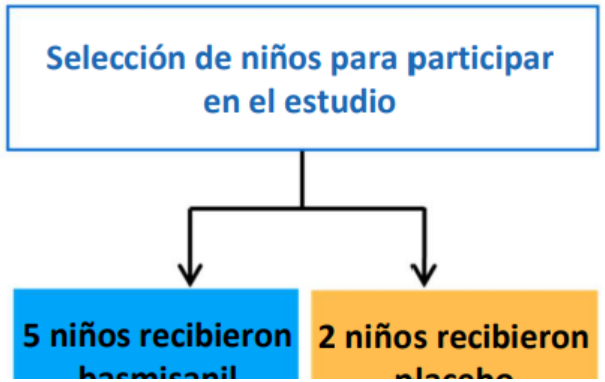
Roche

- ***ATENUACIÓN DEL RC GABA A***
- ***EEUU, UK, España, Portugal, Polonia***
- ***Diciembre 22 a Enero 24***



PRIMERA FASE

- Eficacia, seguridad, farmacodinamia
- 52 semanas
- 2-14 años

SEGUNDA FAS

- Open label exter
- 2 años
- Eficacia y seguri a largo plazo

CANCELLED

Epilepsy & Behavior 142 (2023) 109173

Contents lists available at ScienceDirect

Epilepsy & Behavior

journal homepage: www.elsevier.com/locate/yebeh

Efficacy, safety, and tolerability of soticlestat as adjunctive therapy for the treatment of seizures in patients with Dup15q syndrome or CDKL5 deficiency disorder in an open-label signal-finding phase II study (ARCADE)

Scott Demarest [a], Shafali Jeste [b], Nitin Agarwal [c], Dimitrios Arkilo [d], Mahnaz Asgharnejad [d,*], Samuel Hsiao [d], Ronald Thibert [e]

[a] Department of Pediatrics and Neurology, University of Colorado School of Medicine, Children's Hospital Colorado, Aurora, Colorado, USA
[b] Division of Neurology, Children's Hospital Los Angeles, Los Angeles, California, USA
[c] Division of Pediatric Epilepsy, Minnesota Epilepsy Group, P.A., and Children's Minnesota, St Paul, Minnesota, USA
[d] Takeda Pharmaceutical Company Limited, Cambridge, Massachusetts, USA
[e] Massachusetts General Hospital for Children, Massachusetts General Hospital, Boston, Massachusetts, USA

PHASE II | MULTICENTRIC | OPEN LABEL | 2-55 AÑOS

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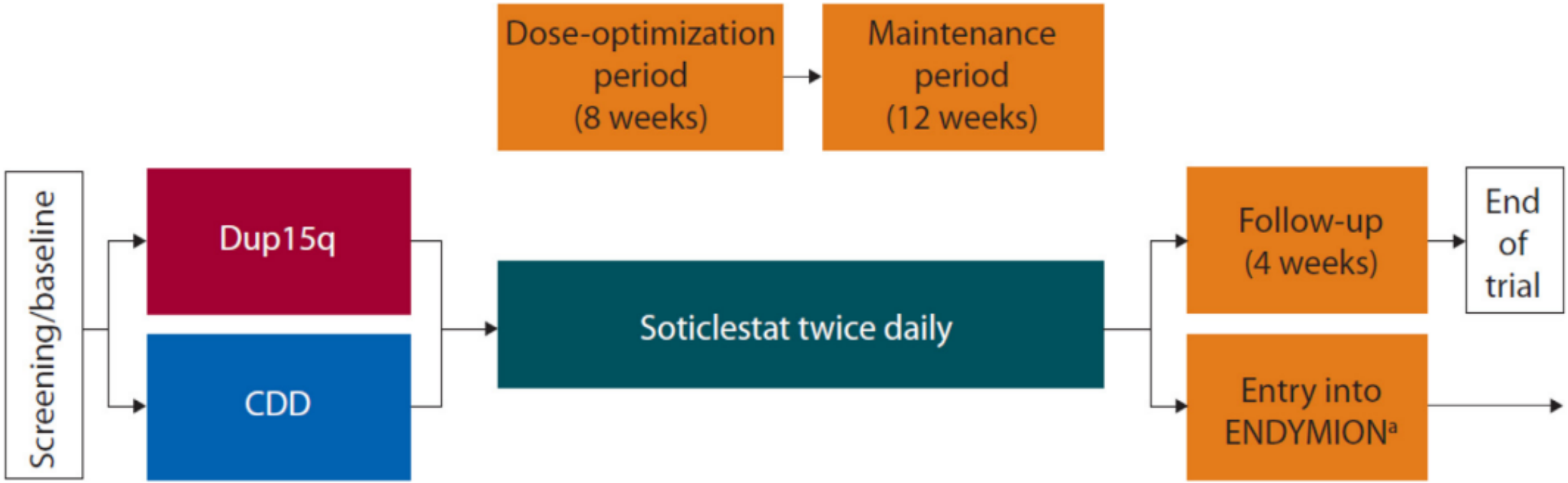
> 3 crisis motoras/mes en los últimos 3 meses



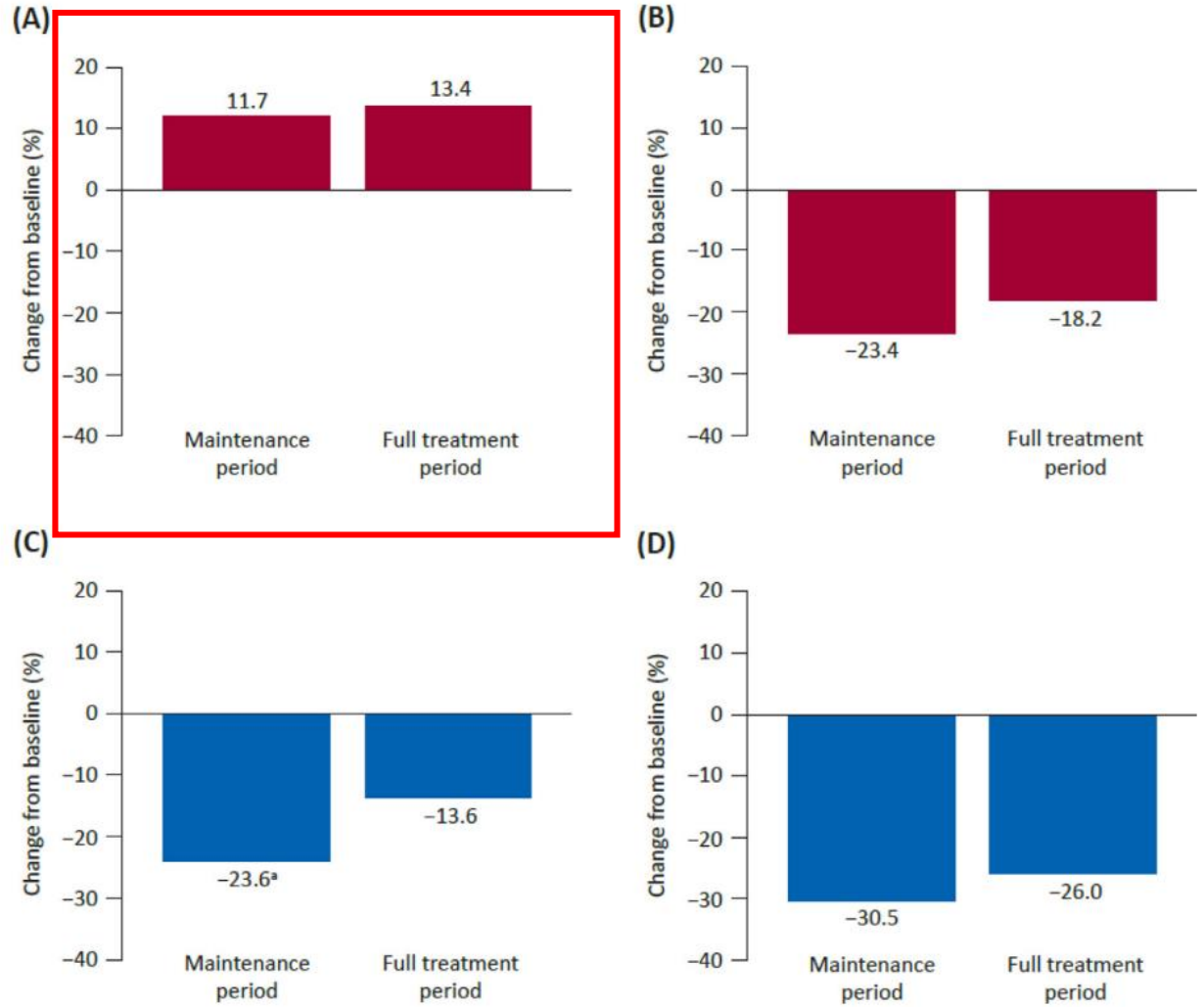


Fig. 2. Change in seizure frequency in patients with Dup15q syndrome or CDKL5 deficiency disorder during the maintenance and full treatment periods. Median percentage change from baseline in (A) motor seizure frequency and (B) all seizure frequency in patients with Dup15q syndrome (n = 8), and median percentage change from baseline in (C) motor seizure frequency and (D) all seizure frequency in patients with CDKL5 deficiency disorder (n = 12). Data are shown for the 12-week maintenance period and the full 20-week treatment period. [a] n = 11.

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OSAKA, Japan and CAMBRIDGE, Massachusetts, January 30, 2025 – Takeda (TSE:4502/NYSE:TAK) today announced the decision to discontinue its soticlestat (TAK-935) development program.

This decision follows the June 2024 announcement that the soticlestat Phase 3 SKYLINE study in Dravet syndrome (DS) and Phase 3 SKYWAY study in Lennox-Gastaut syndrome (LGS) missed their primary endpoints.[1] Subsequently, Takeda discontinued the soticlestat LGS development program [2,3] and engaged with the U.S. Food and Drug Administration (FDA) around the totality of evidence for soticlestat treatment for DS. The FDA informed Takeda that the current clinical data package would not be capable of demonstrating substantial evidence of effectiveness to support a New Drug Application (NDA) for soticlestat in DS. Data from SKYLINE and SKYWAY studies are publicly available on ClinicalTrials.gov.[4,5]

Dup15q Syndrome Market Insight, Epidemiology And Market Forecast - 2034

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Study Period	**2020–2034**
Forecast Period	**2025–2034**
Geographies Covered	**US, EU4 (Germany, France, Italy, and Spain) and the UK, and Japan**
Dup15q Syndrome	**Segmented by** ● Total Prevalent Cases of Dup15q Syndrome in the 7MM [2020–2034] ● Total Diagnosed Prevalent Cases of Dup15q Syndrome in the 7MM [2020–2034] ● Type-specific Diagnosed Prevalent Cases of Dup15q Syndrome in the 7MM [2020–2034]
Dup15q Syndrome Companies	● Lundbeck ● Quiver Bioscience
Dup15q Syndrome Therapies	● Bexicaserin ● UBE3A (ASO)

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March 3, 2025 • General

Acadia Pharmaceuticals and Saniona Announce Initial Positive Results from ACP-711 (formerly SAN711) Phase 1 Study

The Acadia/Saniona deal secures financing enabling Saniona to start Phase II for SAN2465 in MDD. Please note that, as per the company, this drug has an additional opportunity to treat the neuropsychiatric symptoms in Dup15q syndrome (although at the moment no trial is ongoing).

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European Journal of Cell Biology 103 (2024) 151446

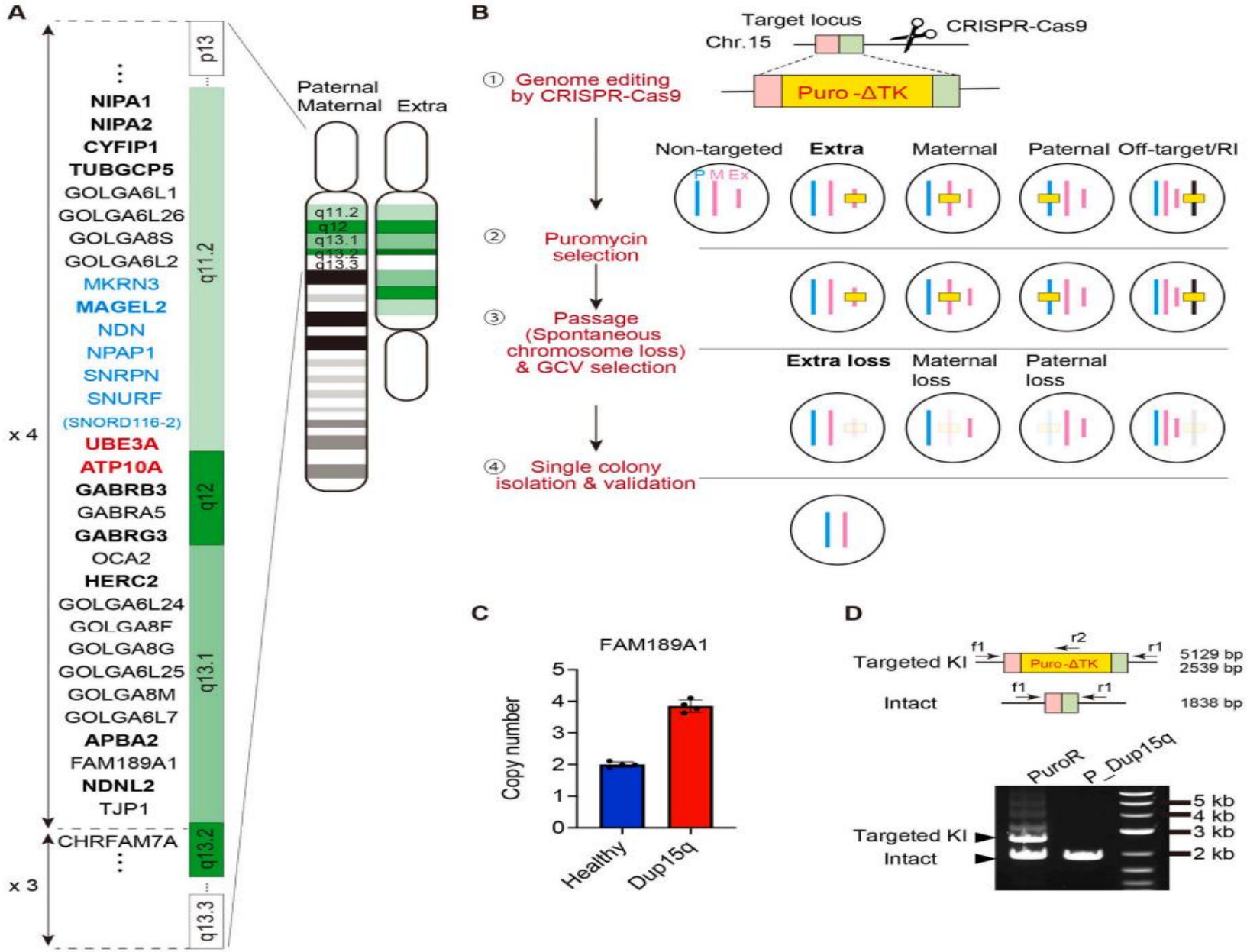
Contents lists available at ScienceDirect

European Journal of Cell Biology

journal homepage: www.elsevier.com/locate/ejcb

Elimination of the extra chromosome of Dup15q syndrome iPSCs for cellular and molecular investigation

Haruka Munezane [a,b], Keiko Imamura [a,b,c,d], Naoko Fujimoto [a,b], Akitsu Hotta [a,b], Hiroshi Yukitake [b,e], Haruhisa Inoue [a,b,c,d,*]



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