

Current Overview of Spinocerebellar Ataxias in Mexican Population: High Prevalence of SCA7



Jonathan J. Magaña PhD.

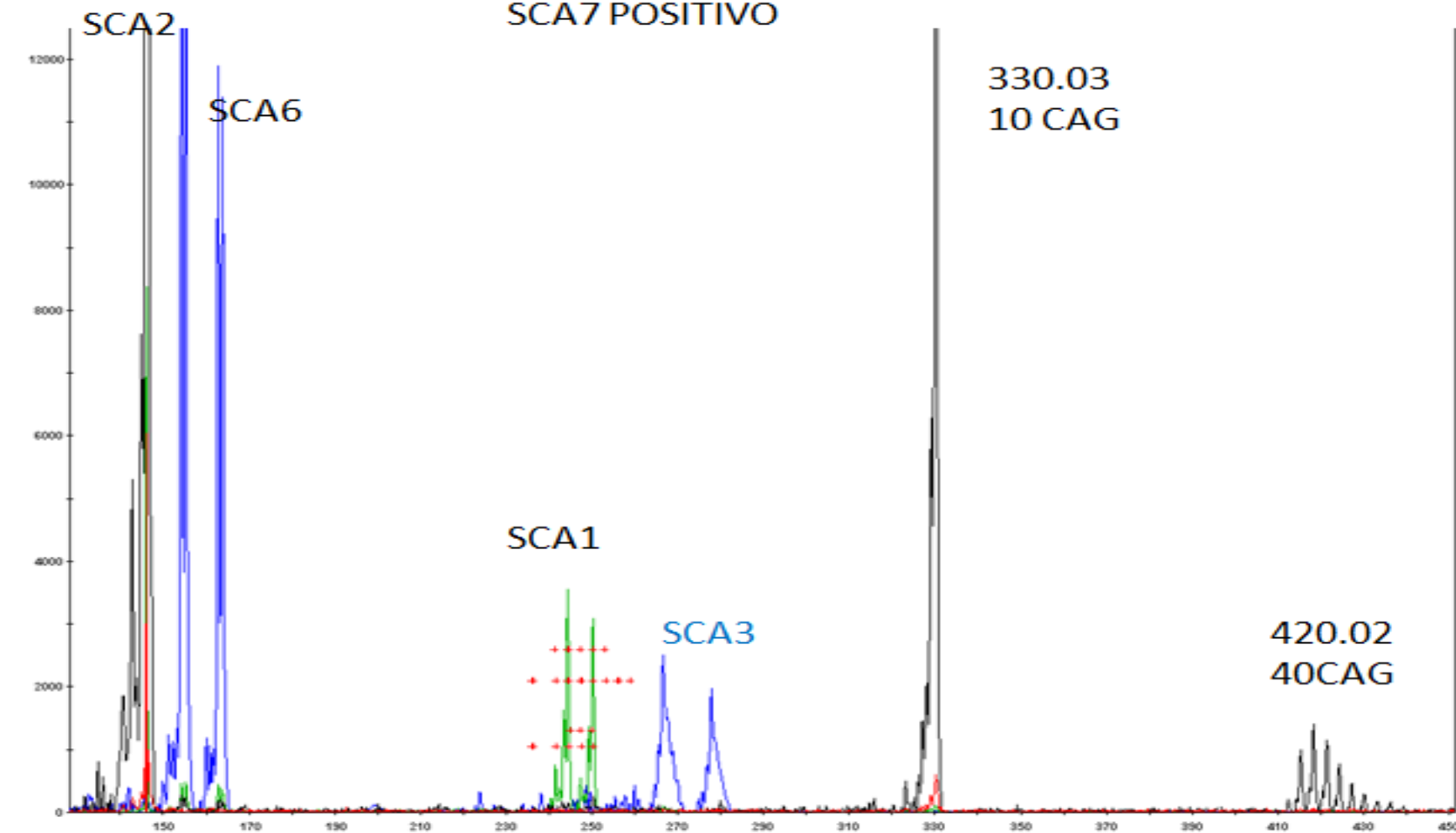
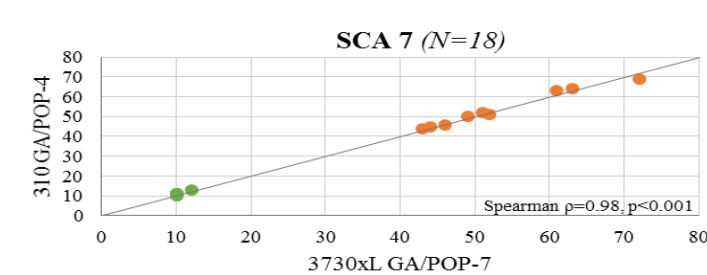
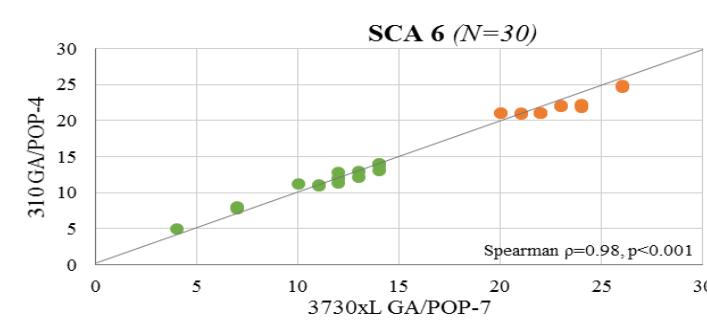
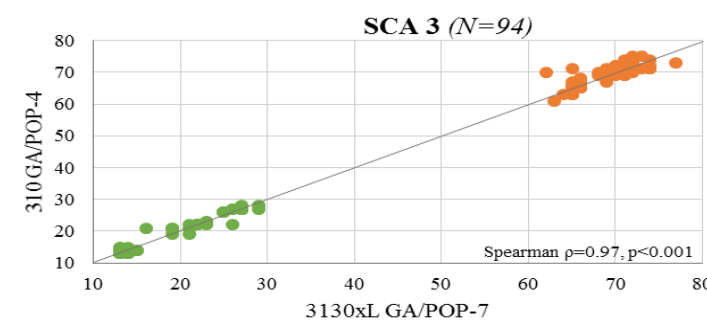
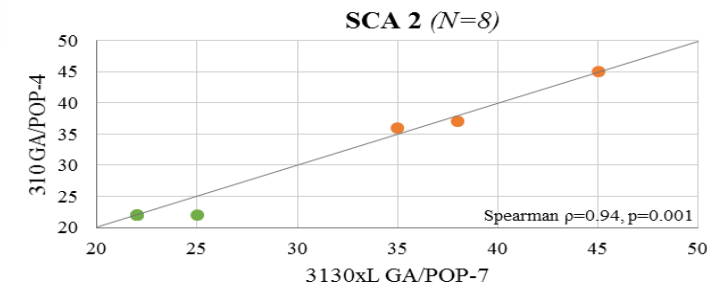
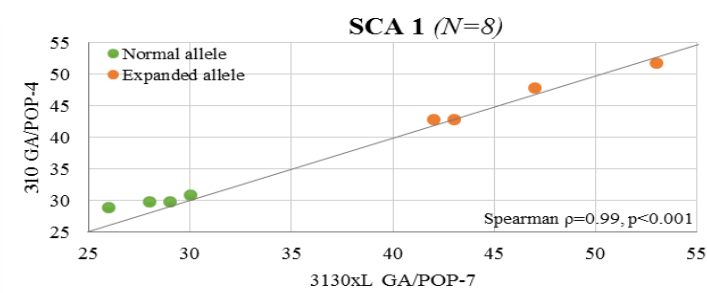
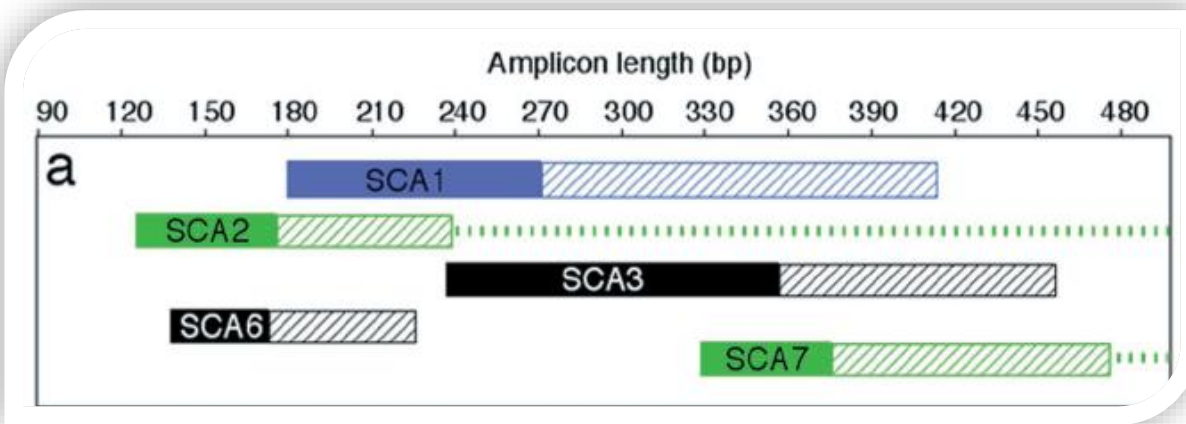
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Molecular Diagnosis of Spinocerebellar Ataxias



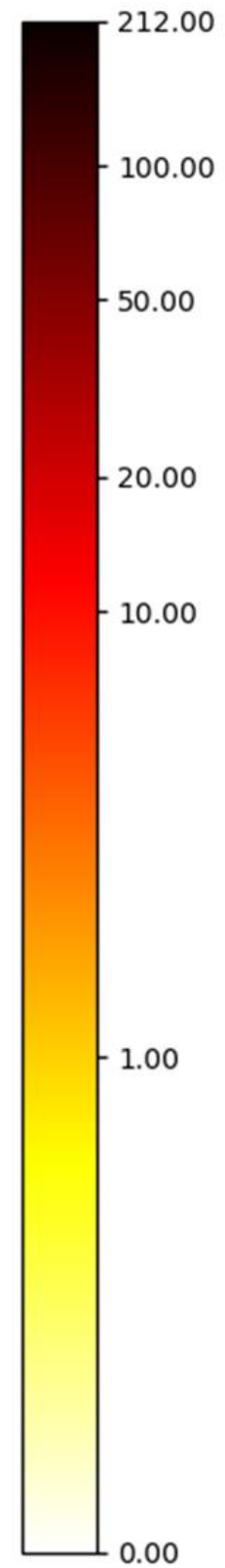
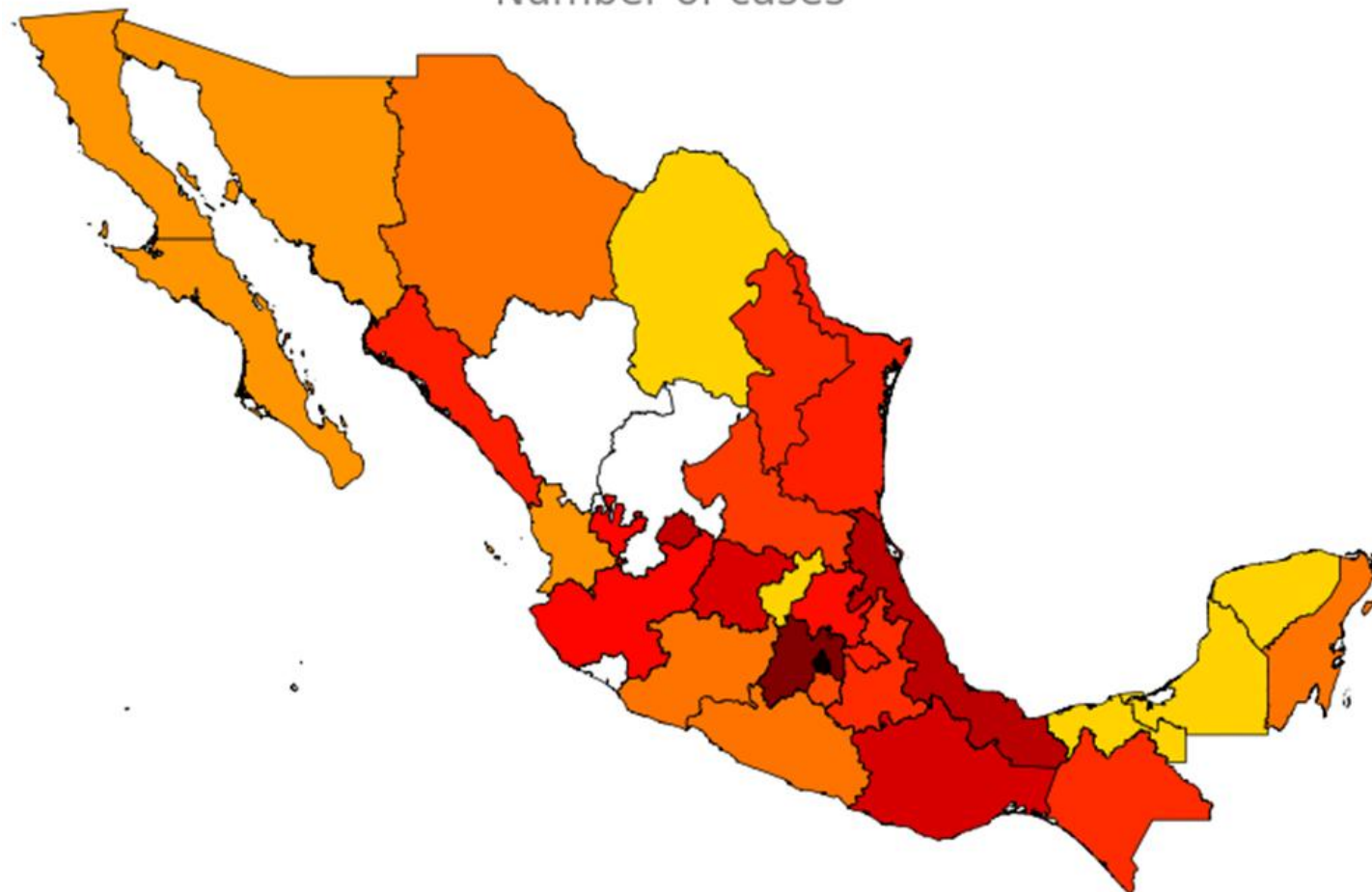
Verification of Inter-laboratorial Genotyping Consistency in the Molecular Diagnosis of Polyglutamine Spinocerebellar Ataxias

Amanda Ramos^{1,2,3} • Mafalda Raposo^{1,2,3} • Montserrat Milà⁴ • Conceição Bettencourt⁵ • Henry Houlden⁵ • Bulmaro Cisneros⁶ • Jonathan J. Magaña⁷ • Bruno Filipe Bettencourt^{2,3,8} • Jácome Bruges-Armas^{2,3,8} • Cristina Santos⁹ • Manuela Lima^{1,2,3}



Geographical Distribution

Number of cases



SCA2 45%

SCA3 24%

SCA7 7%

SCA1 3%

SCA6 2%

UNDIAGNOSED 19%

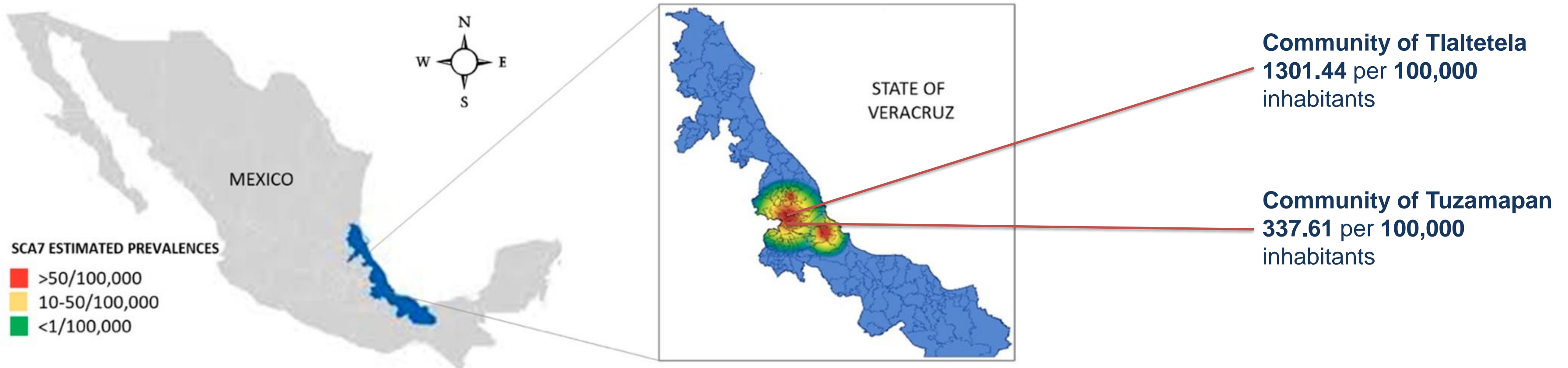


Table 1. Prevalence for patients with SCA7 in the central region of the state of Veracruz.

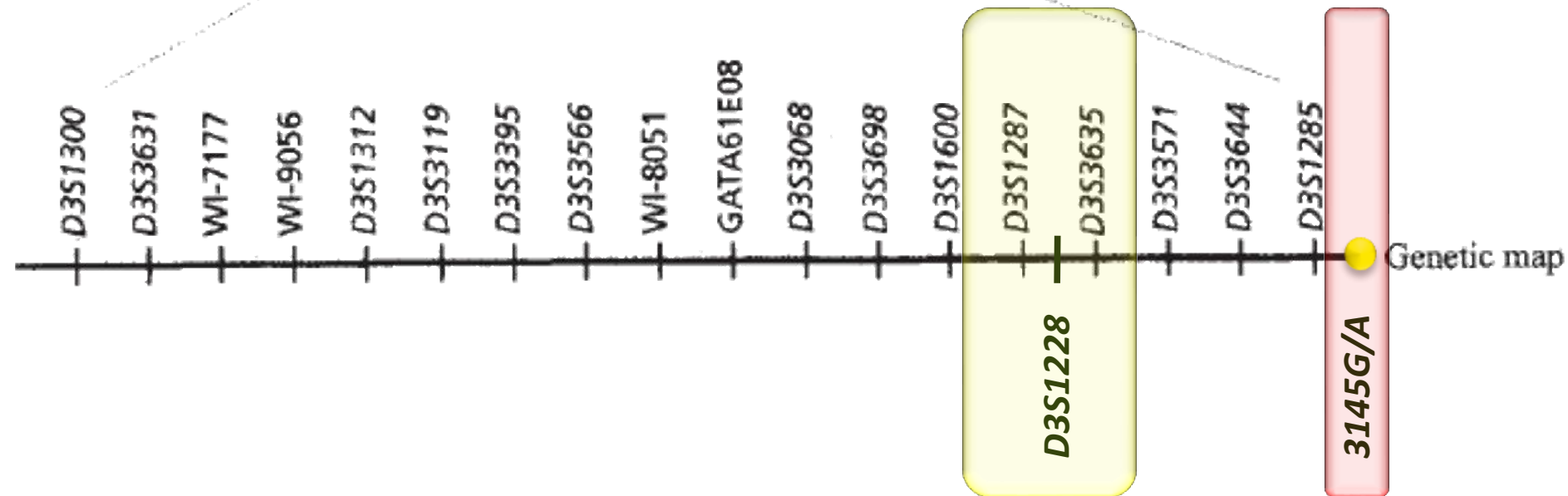
Municipality	Inhabitants	SCA7 Patients	Prevalence Rate
Tlaltetela	16,486	74	448.87
Cosautlan	16,167	20	123.71
Coatepec	93,911	35	37.27
Teocelo	16,957	6	35.38
Naolinco	22,835	4	17.52
Acajete	9701	1	10.31
Tezonapa	54,537	2	3.67
Cuitlahuac	28,875	1	3.46
Xalapa	488,531	14	2.87
Cordoba	204,721	1	0.49
Emiliano Zapata	85,489	1	1.17

Given that several communities have fewer than 6000 inhabitants. Given that several communities have fewer than 6000 inhabitants, we decided to report the prevalence by municipality.

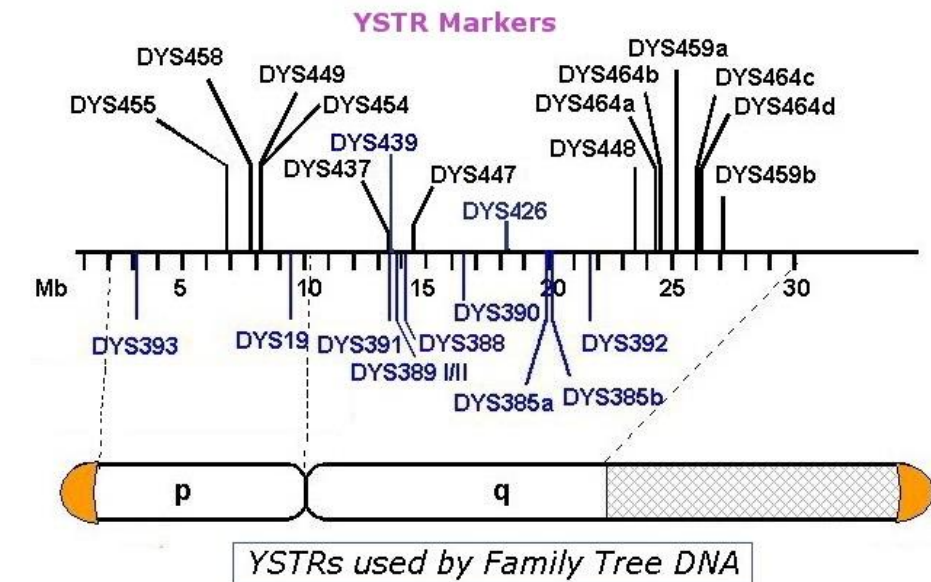
(populations exceeding 10,000 people).



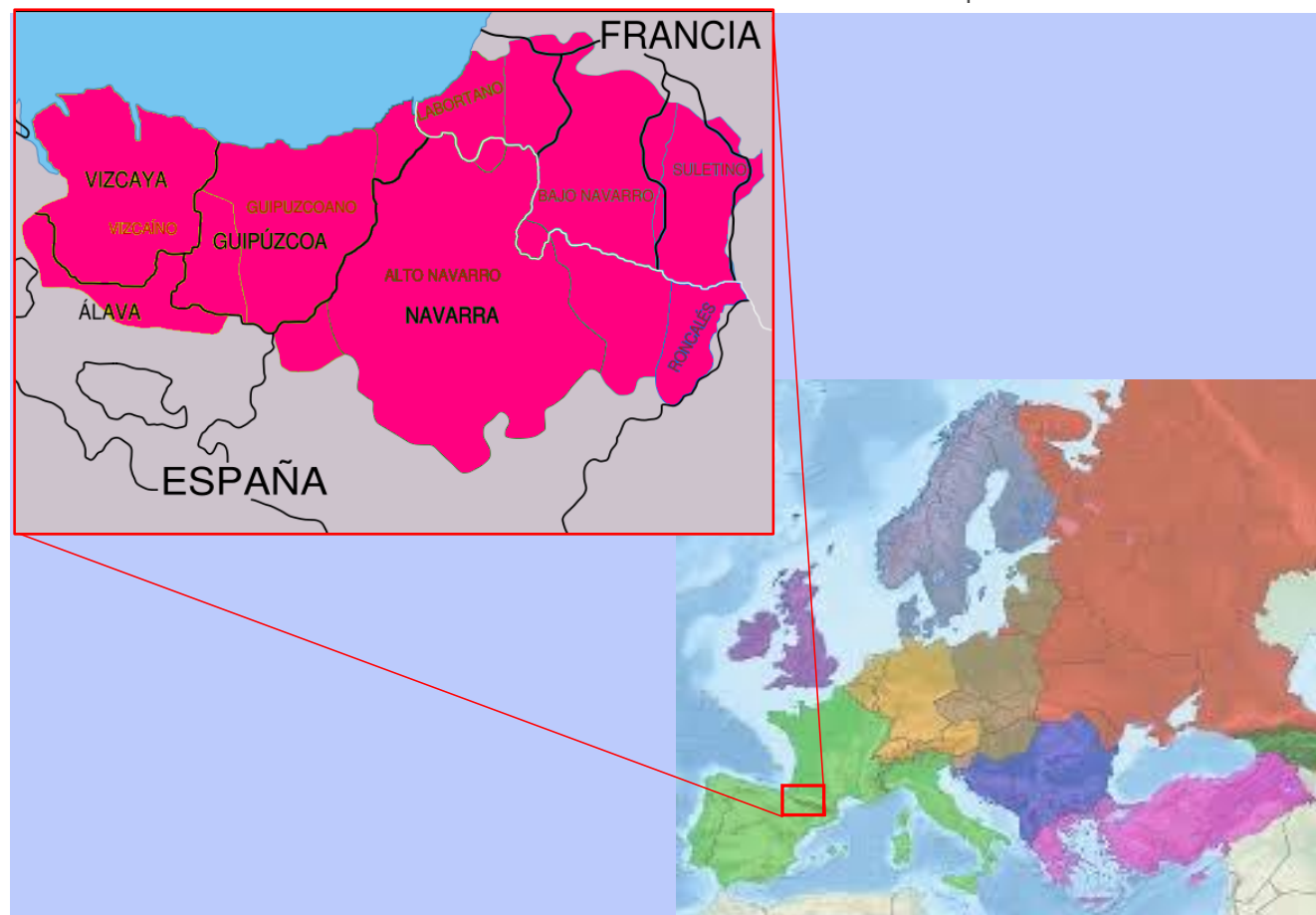
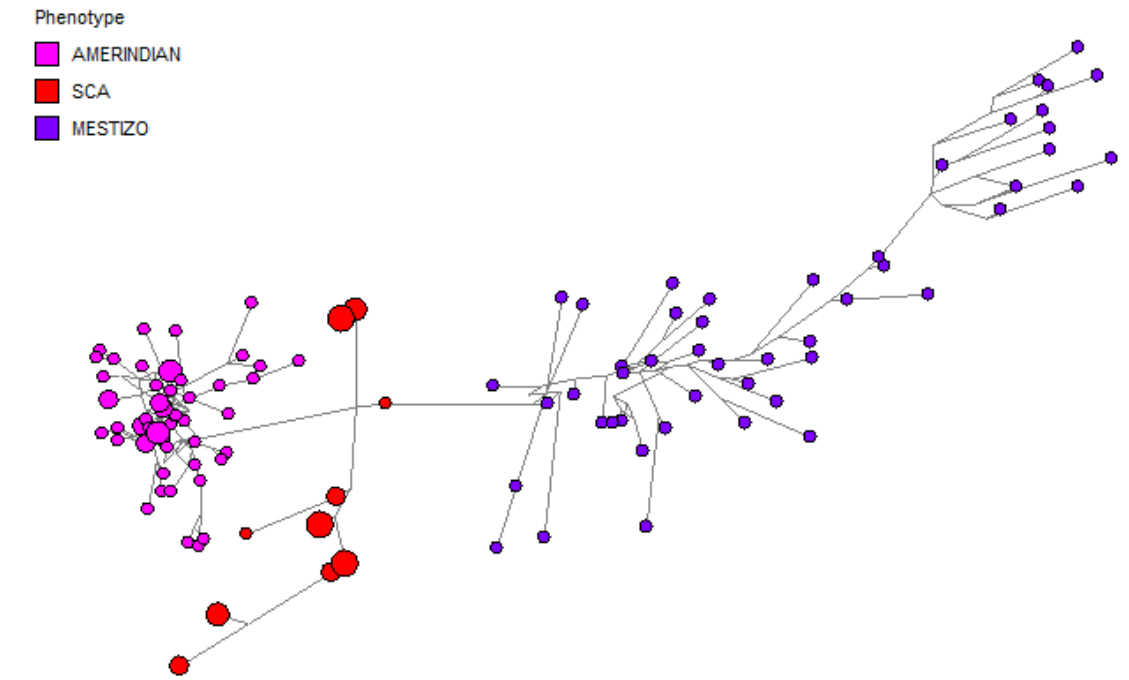
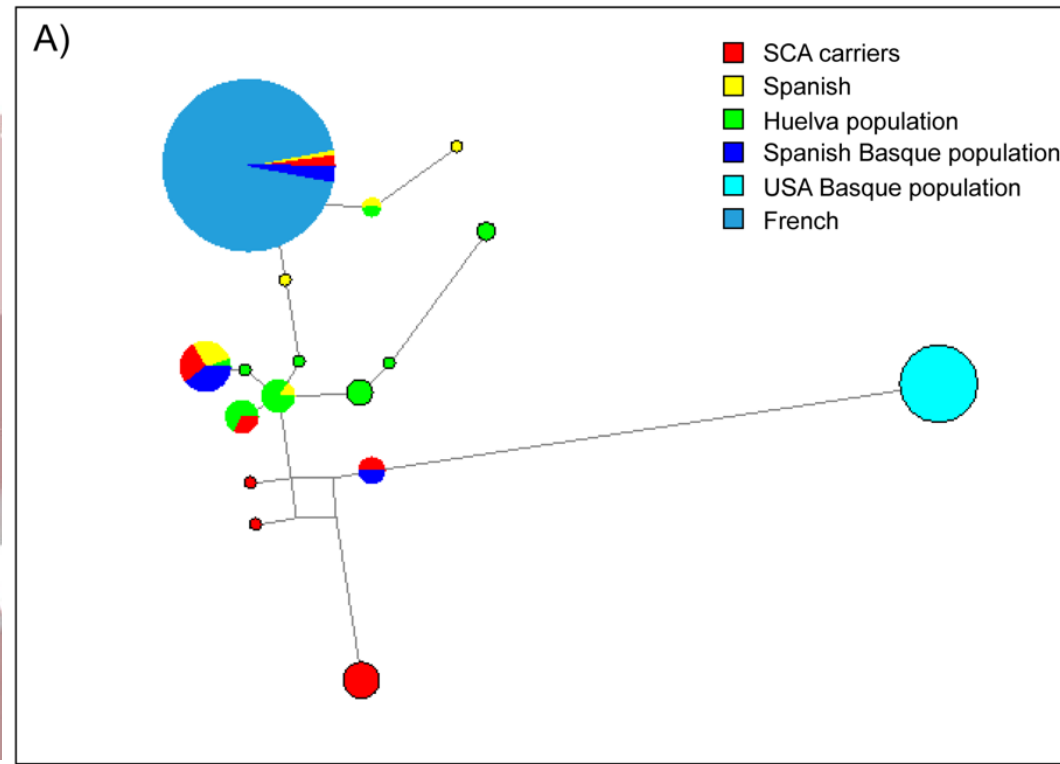
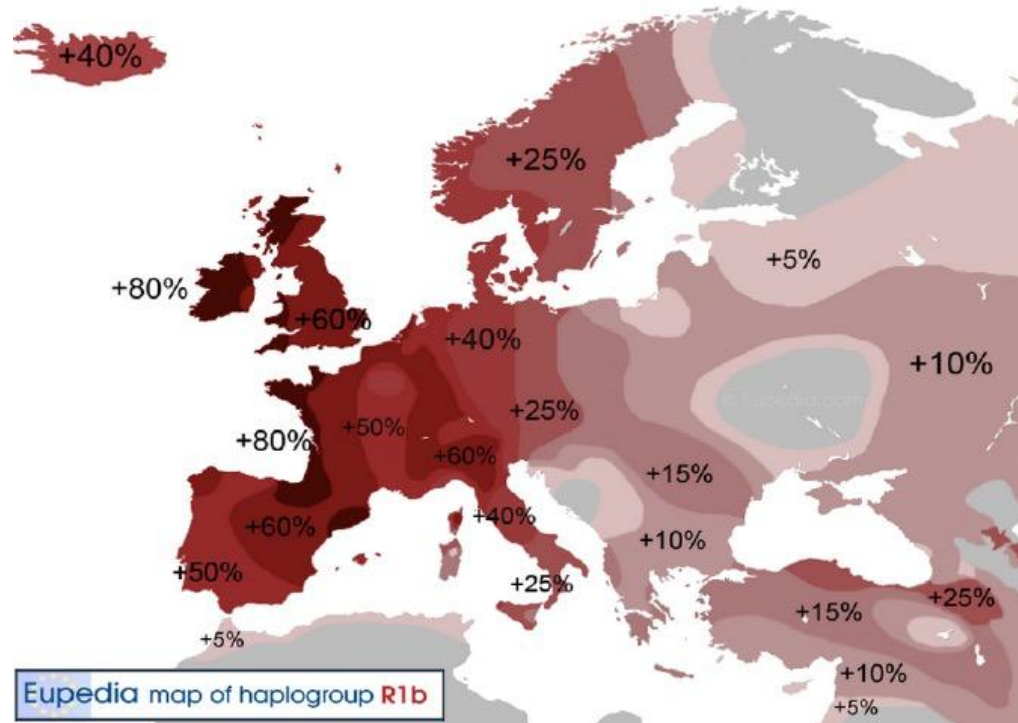
HAPLOTYPE : 254, 82, 98, A



YSTR Positions along Y Chromosome



Haplotype	Non-Admixed		Unrelated population		General population	
	SPAR Satellites (%) (n=R2)	SPAR Unrelated (%) (n=R5)	p-value	Control (%) (n=R0)	p-value	General population (%) (n=300)
A-25L-82-98	100.00	6.66	0.001	5.71	0.001	6.00



Cerebellum (2013) 12:902–905
DOI 10.1007/s12311-013-0505-8

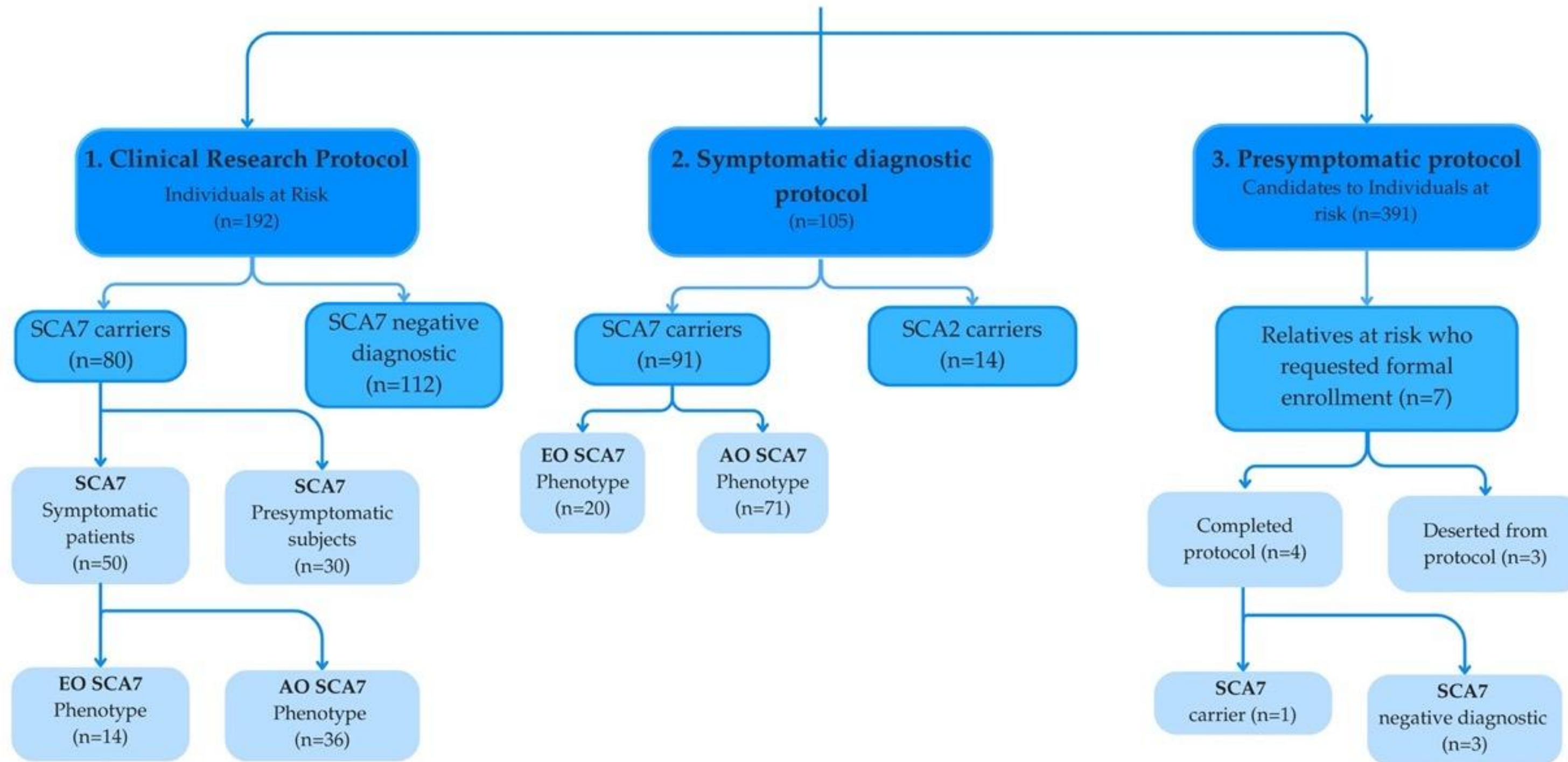
ORIGINAL PAPER

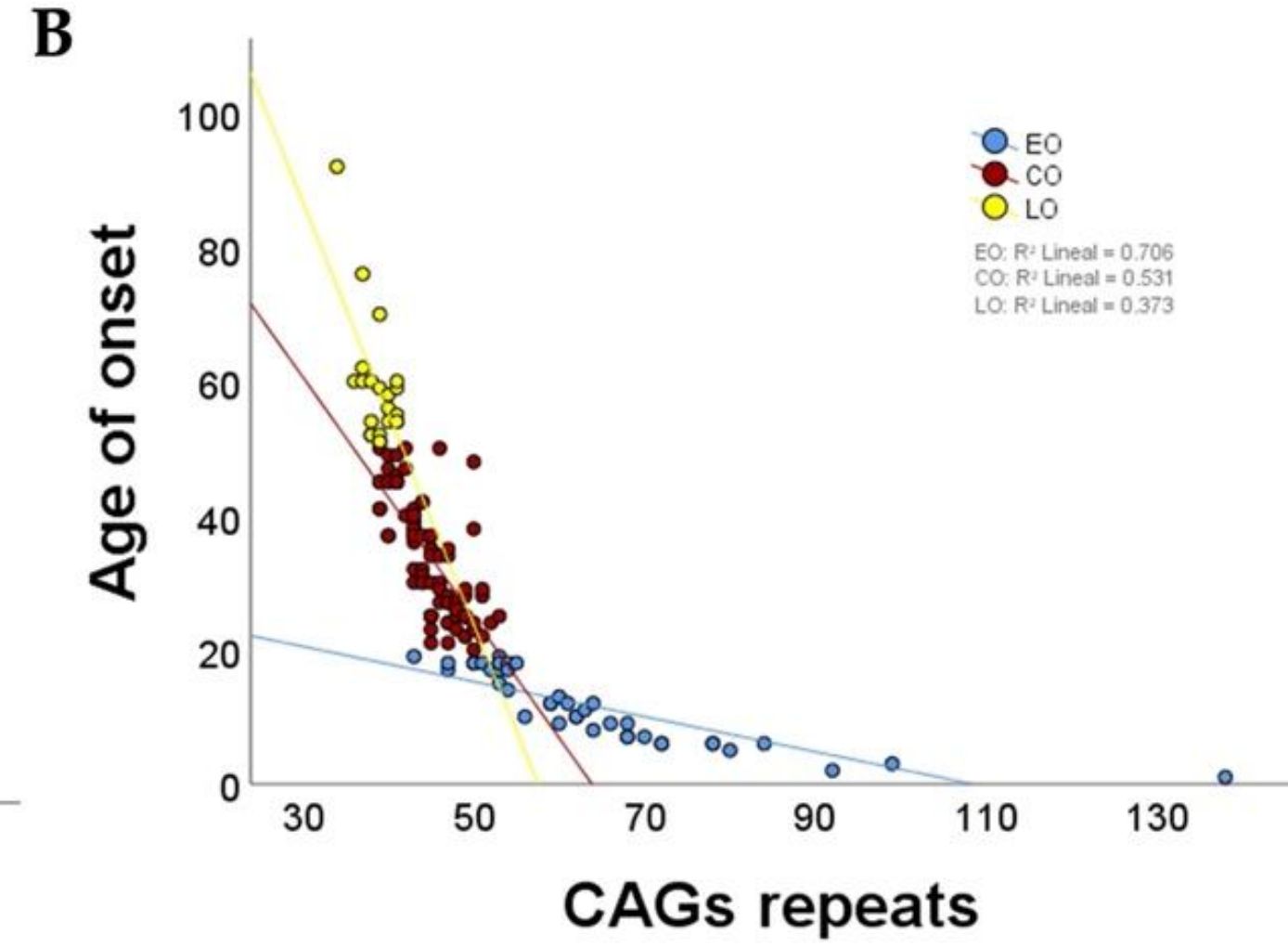
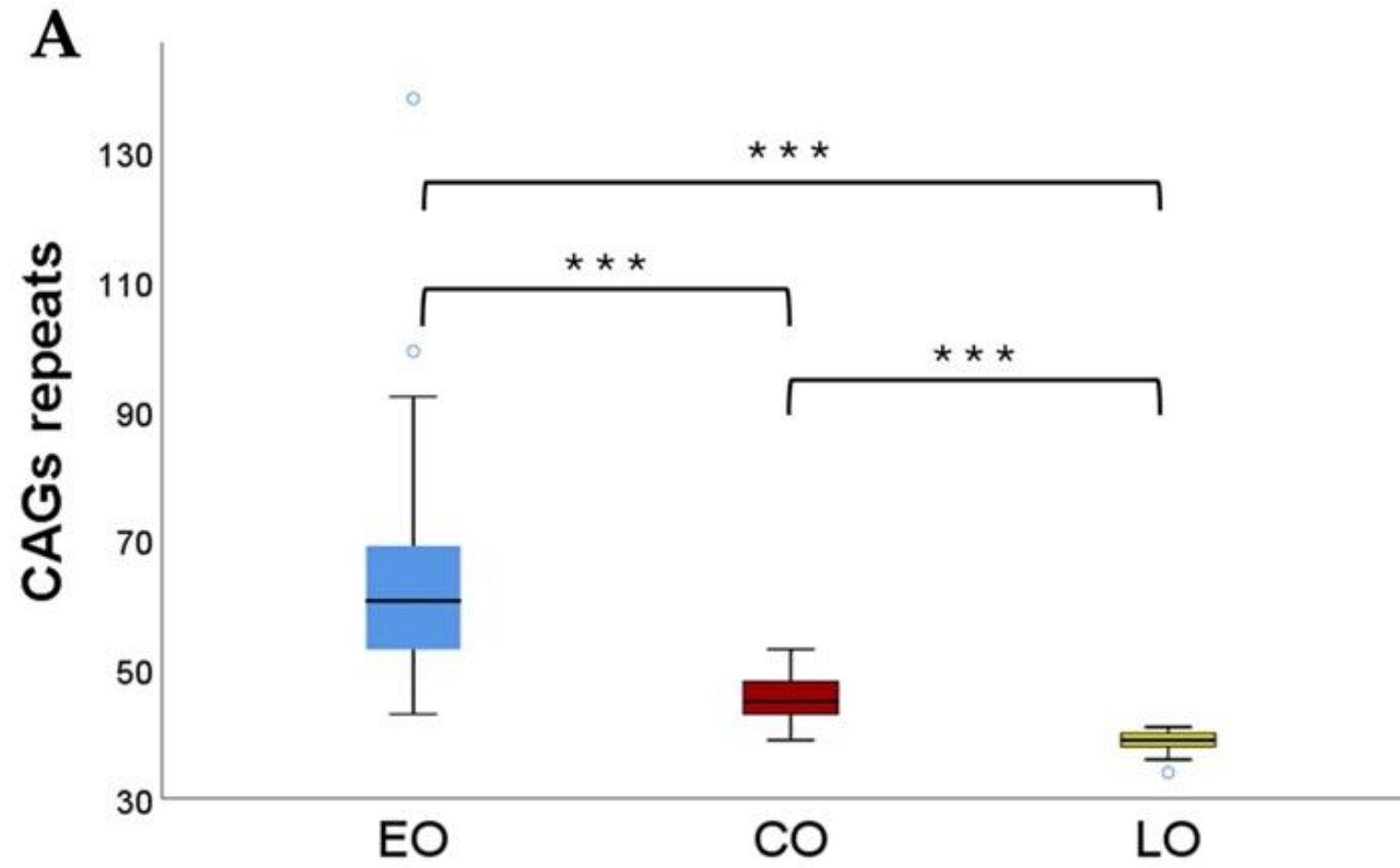
Origin of the Spinocerebellar Ataxia Type 7 Gene Mutation in Mexican Population

J. J. Magaña • R. Gómez • M. Maldonado-Rodríguez •
L. Velázquez-Pérez • Y. S. Tapia-Guerrero • H. Cortés •
N. Leyva-García • O. Hernández-Hernández • B. Cisneros



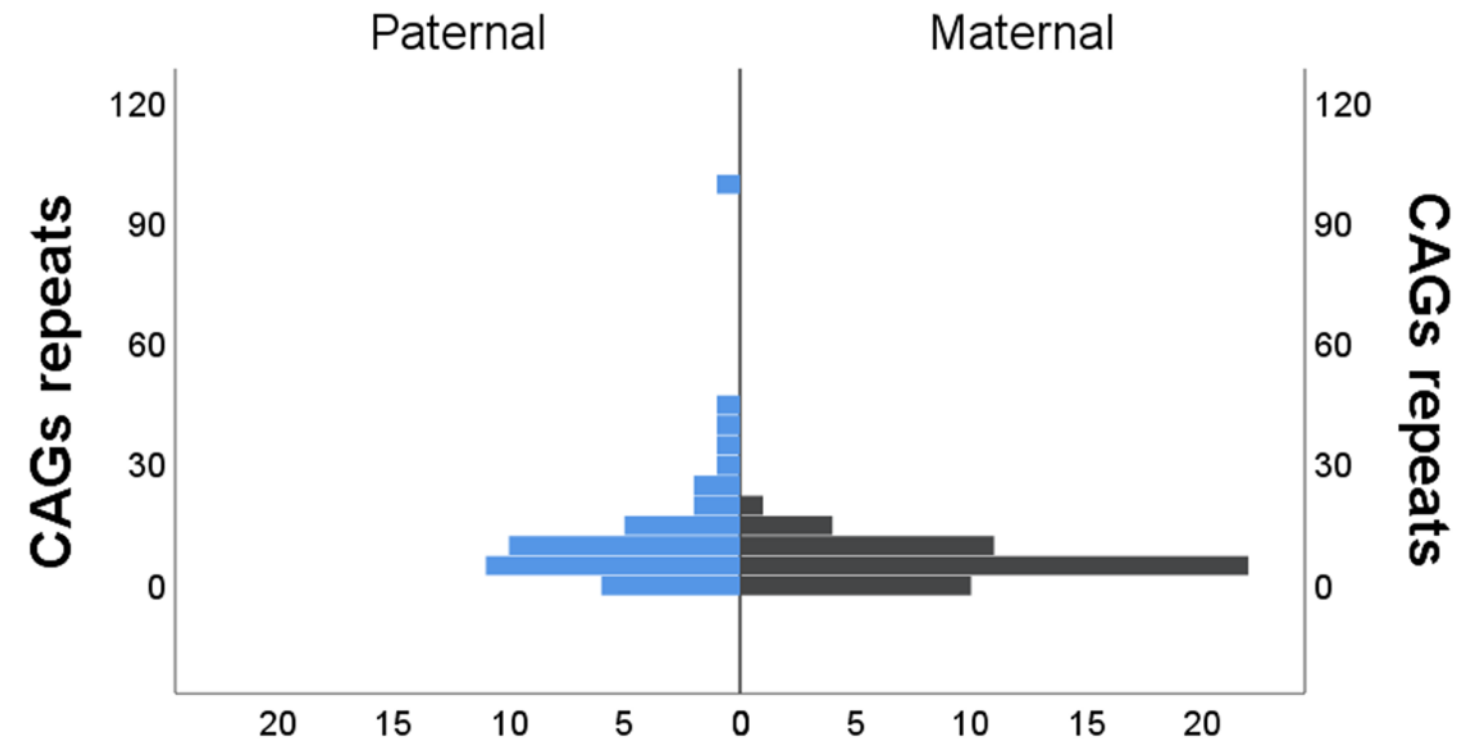
SCA7 testing protocol (2013-2024)





Dr. César Cerecedo Zapata

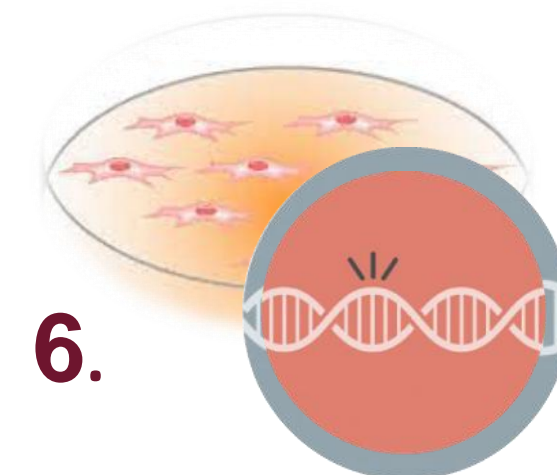
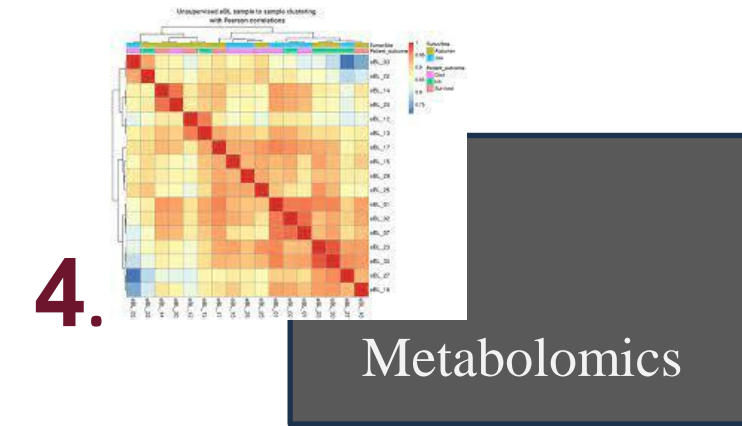
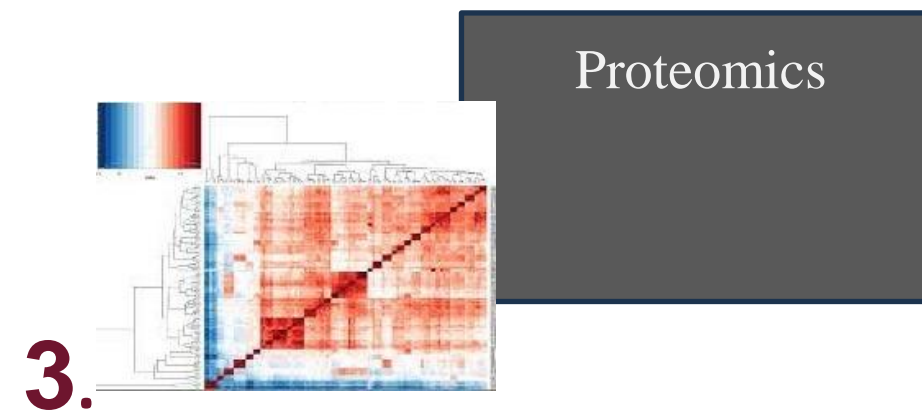
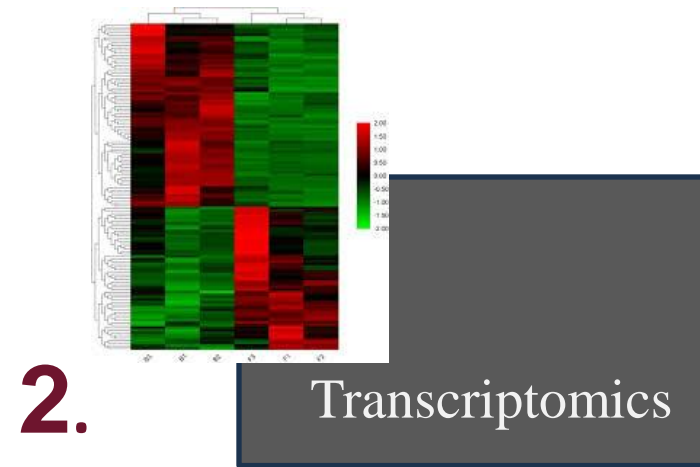
A





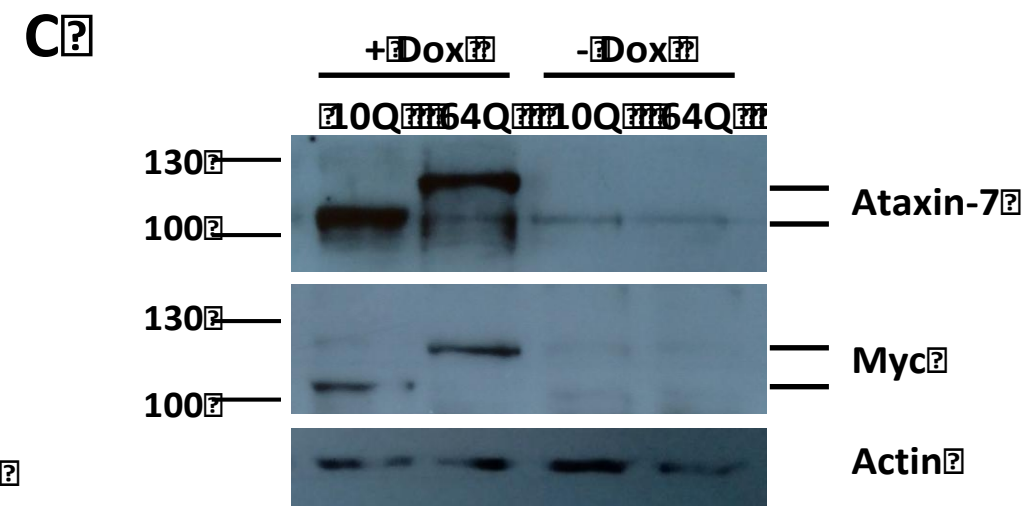
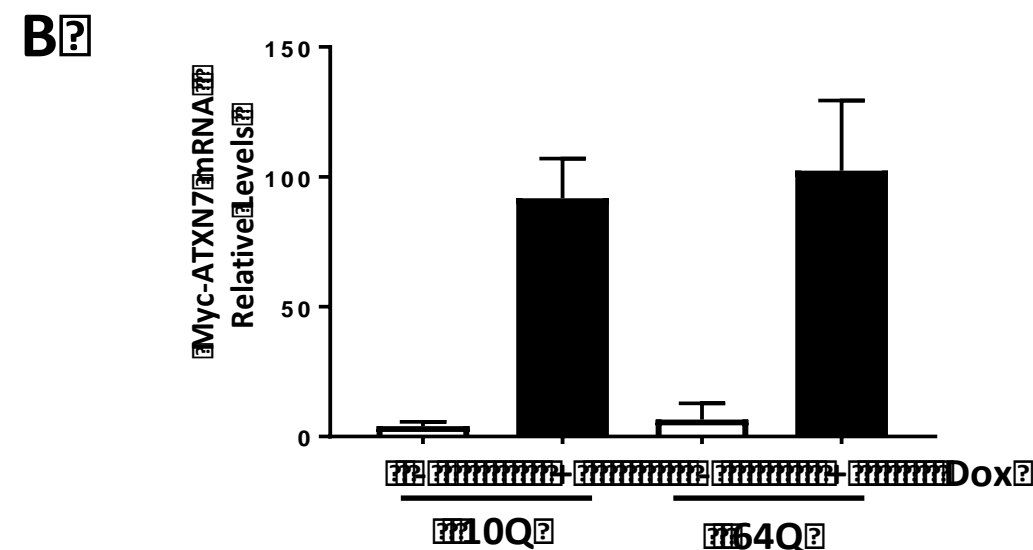
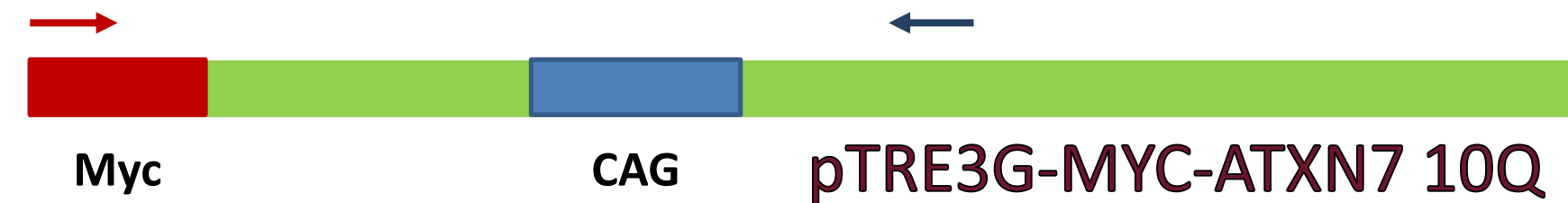
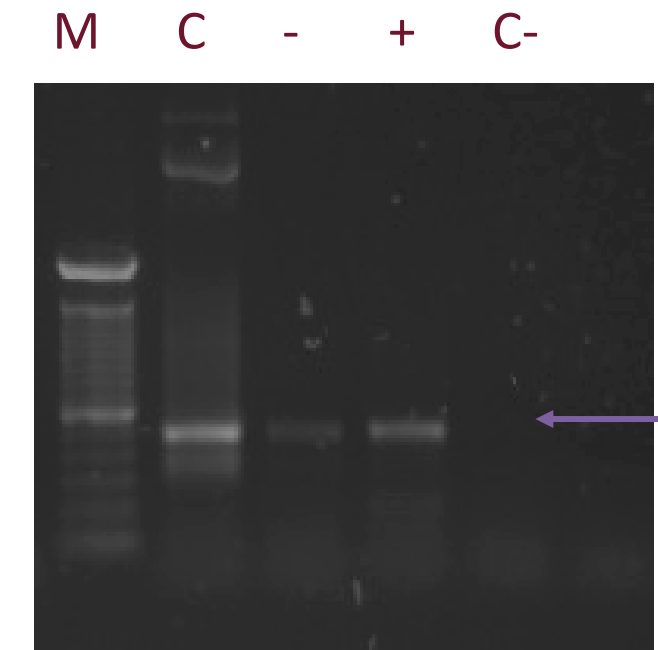
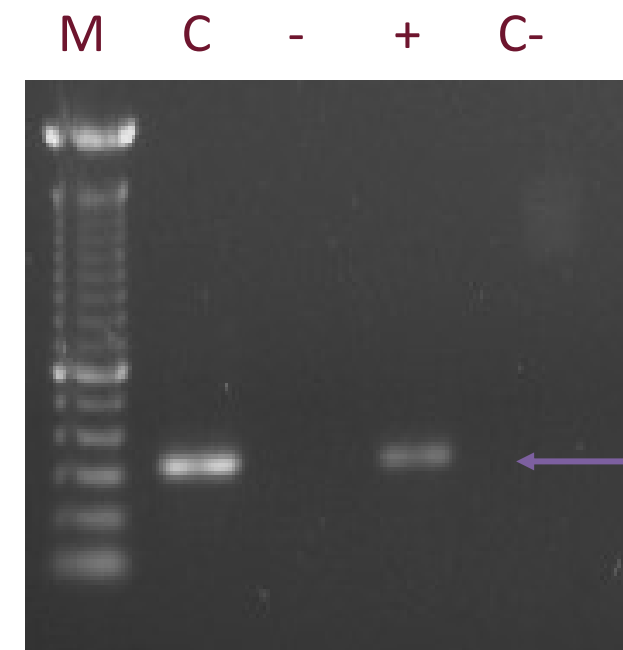
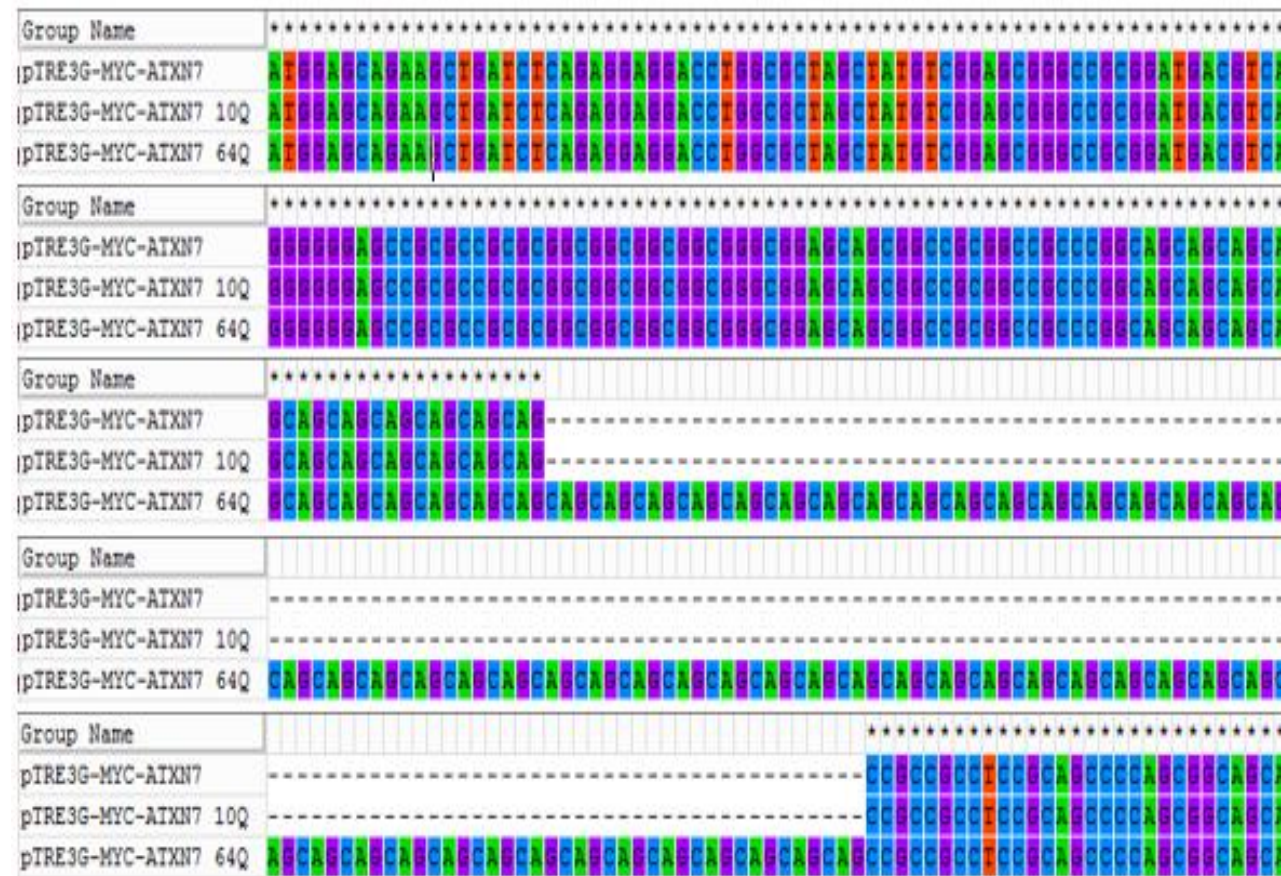
BIOMARKERS

1. Methods of
Biochemical
Analysis





EXPERIMENTAL CELL MODELS FOR SCA7



Dr. Ian A. García Aguirre

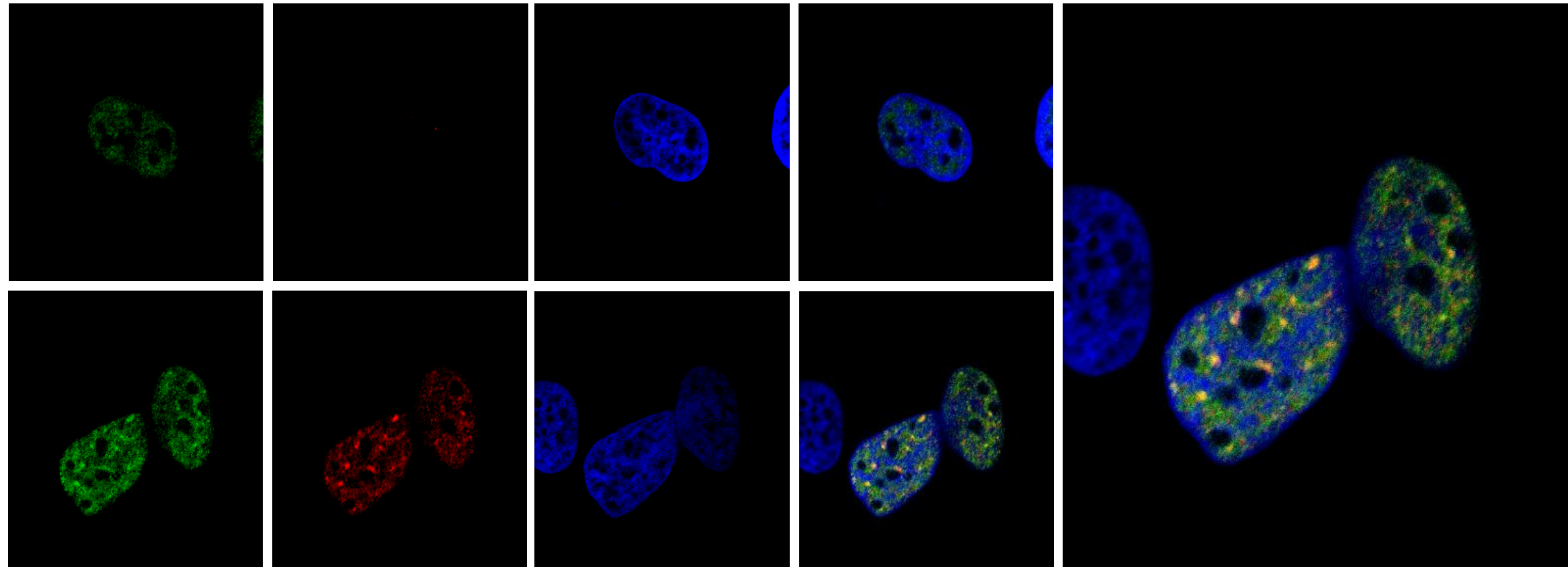


Dr. Oscar Hernández H.



ATXN7 64Q Model

unstimulated



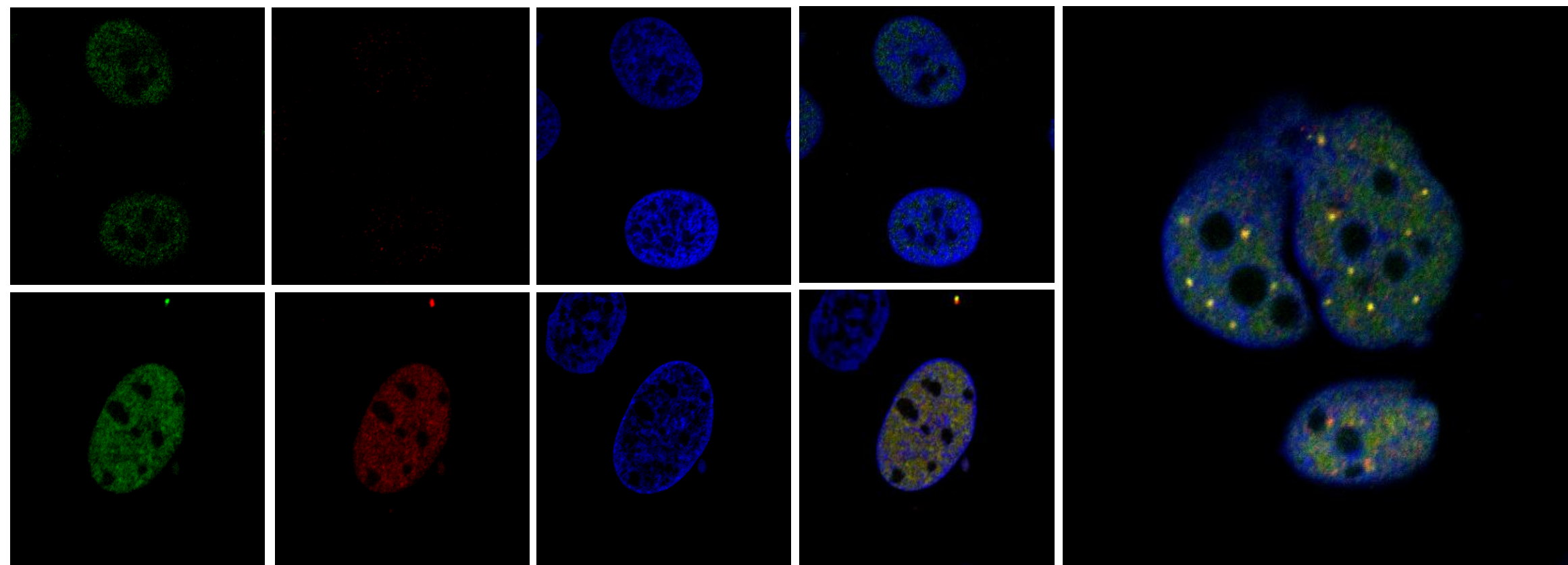
Dox



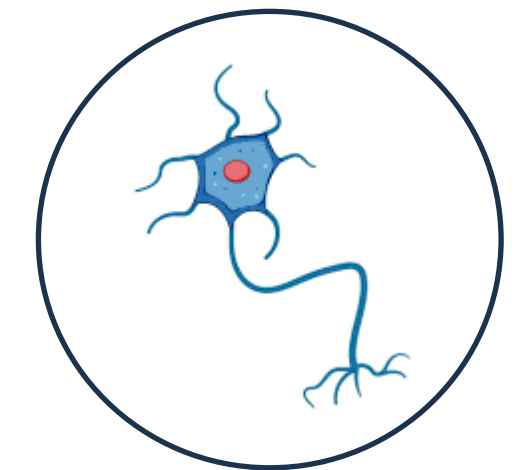
MIO-M1 Inducible Model

ATXN7 10Q Model

unstimulated



Dox



SH-SY5Y Inducible Model
SH-SY5Y Constitutive Model

ATXN 7

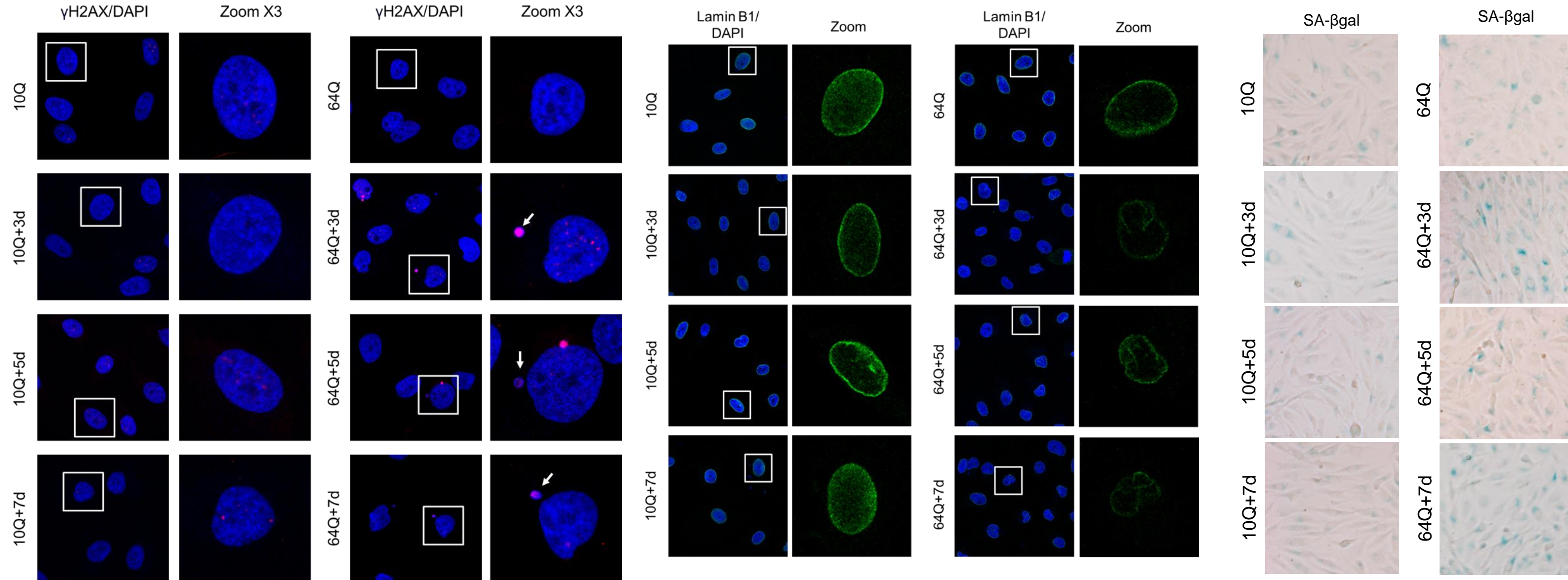
Myc

DAPI

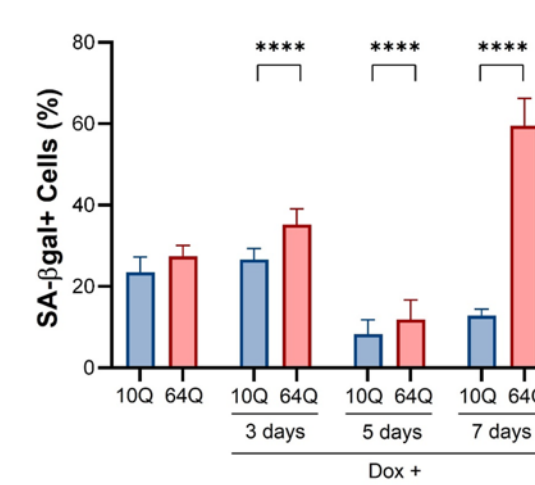
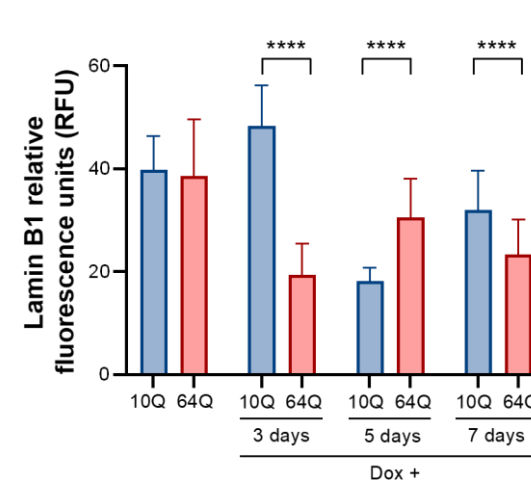
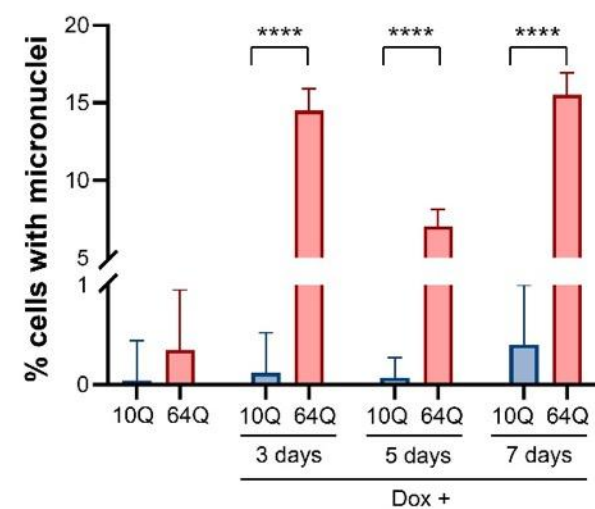
Merge



Molecular Mechanisms



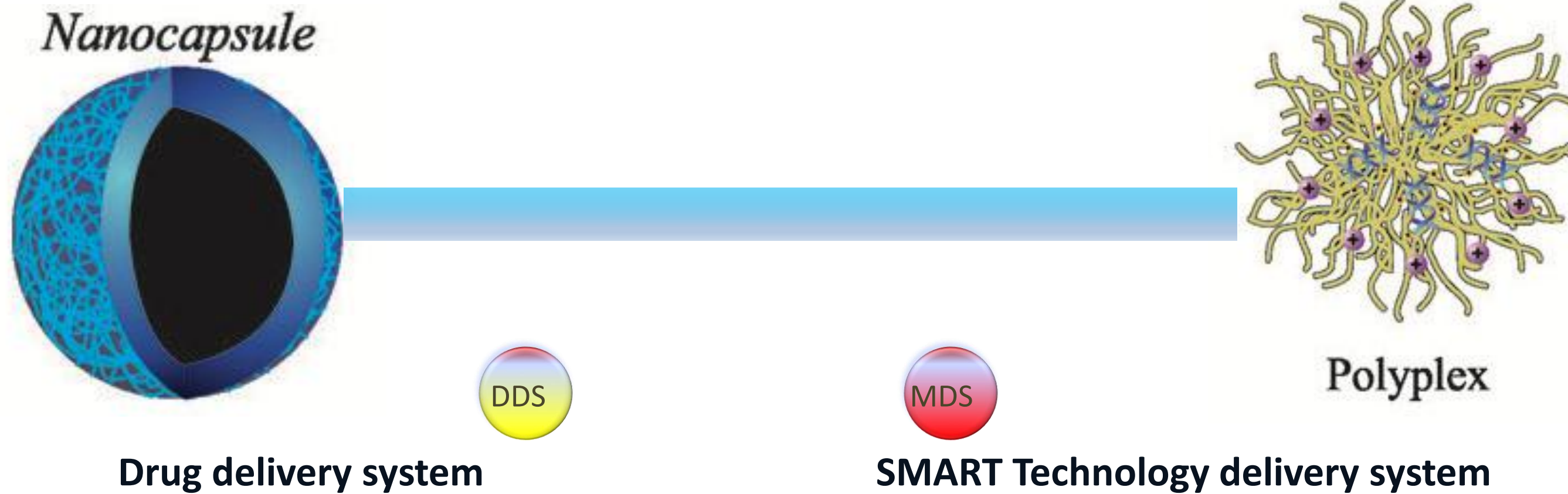
M. en C. Vanessa Ruiz Esparza



IBT. Aranza Meza Dorantes



THERAPEUTIC ALTERNATIVES FOR SCA7



Dra.en C. Yessica S. Tapia Guerrero



Dra.en C. Fabiola Borbolla Jiménez



Dr. Gerardo Leyva

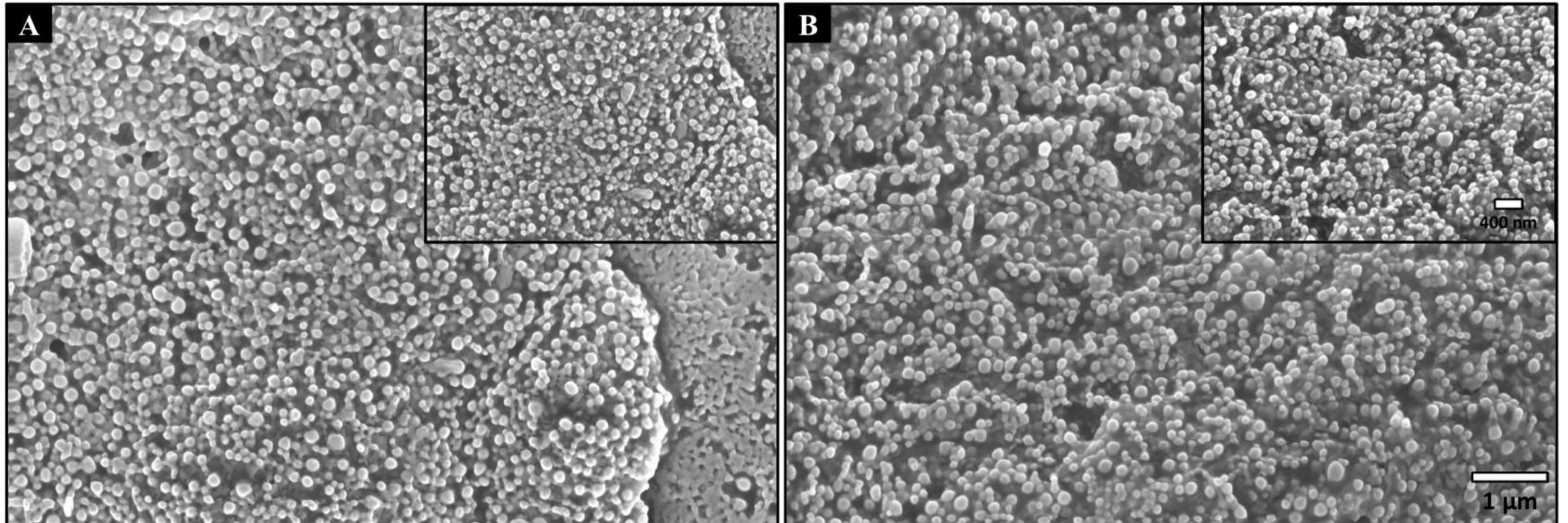
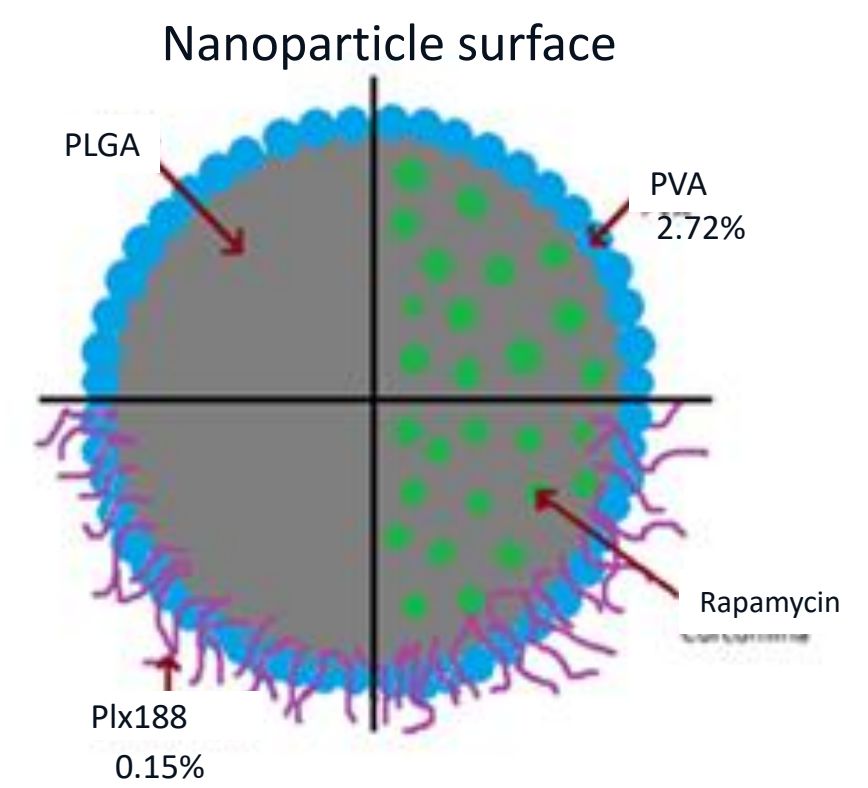


Dra.en C. Mireya Murillo-Melo



Dra.en C. Magali Hernández

Nanoparticle Carrier Drug Delivery System





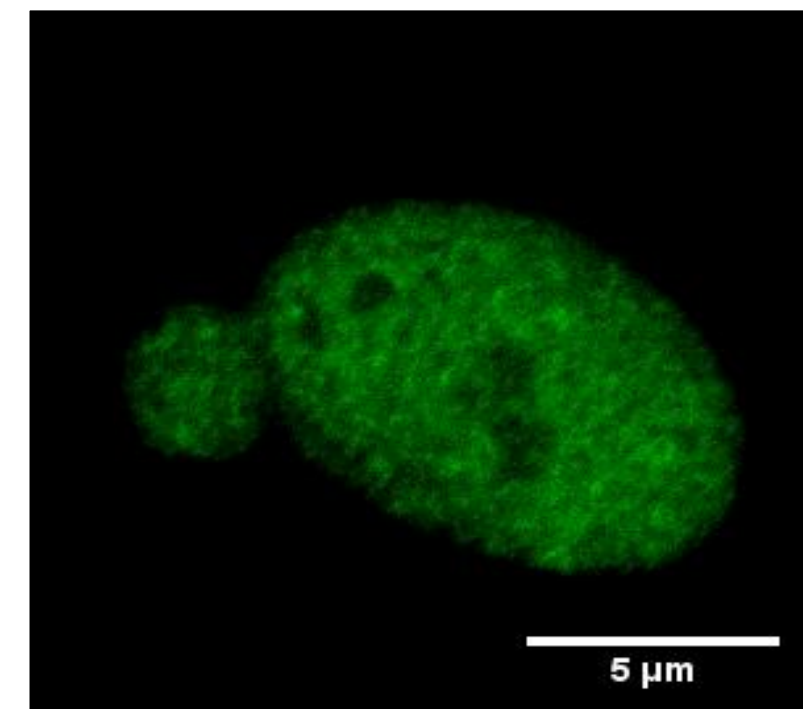
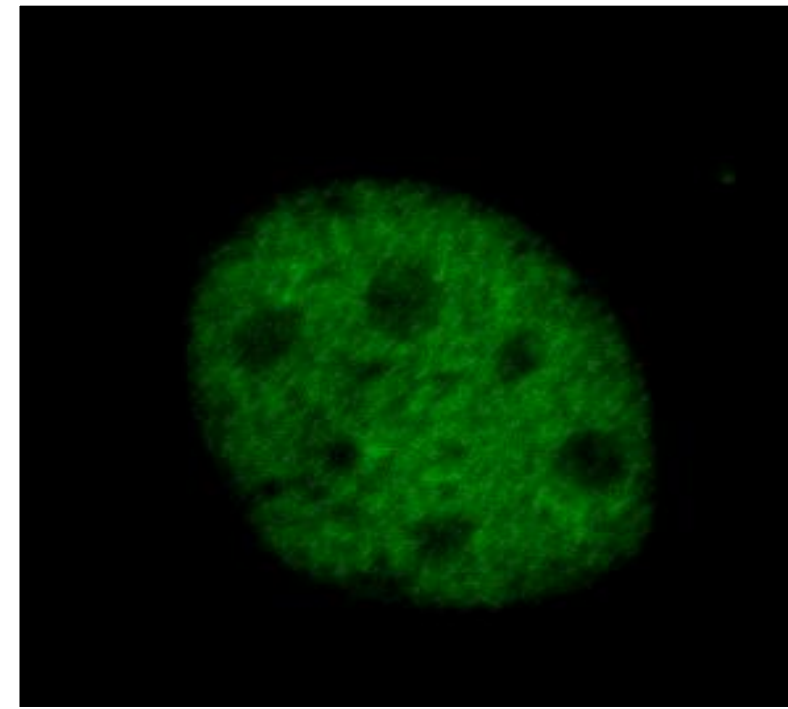
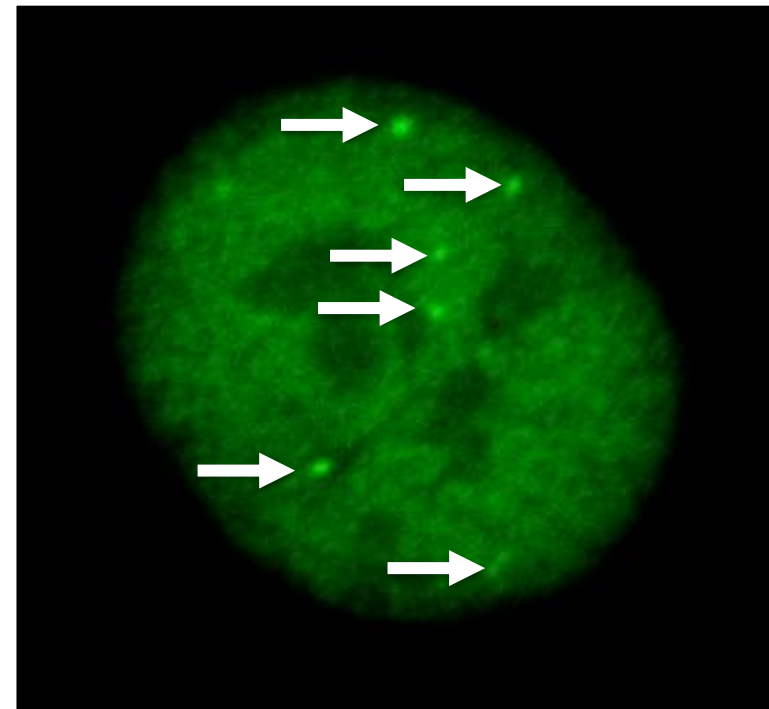
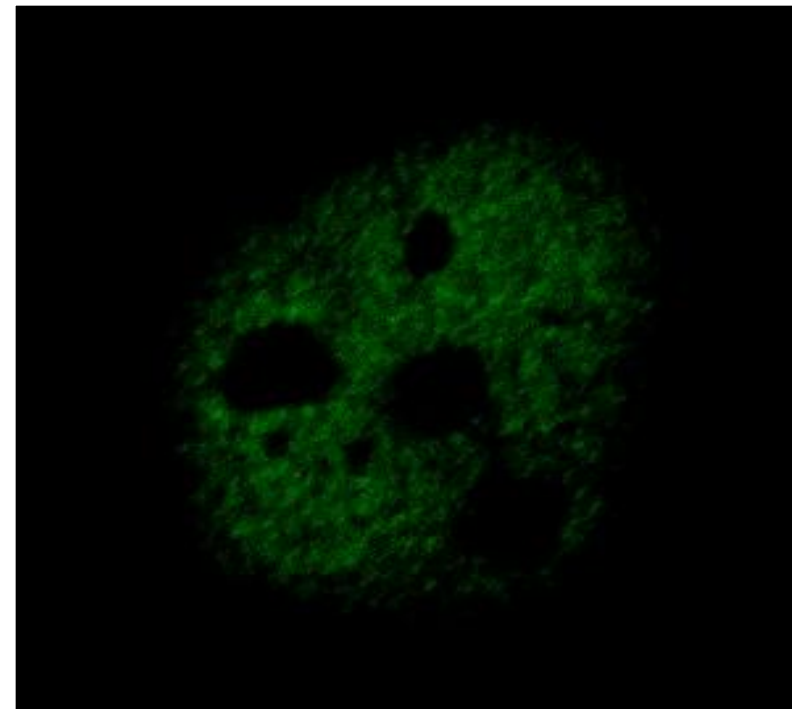
Therapeutic Efficiency Drug Delivery System

DOX

Plx188 NP

Plx188/Rap NP

Rapamycin

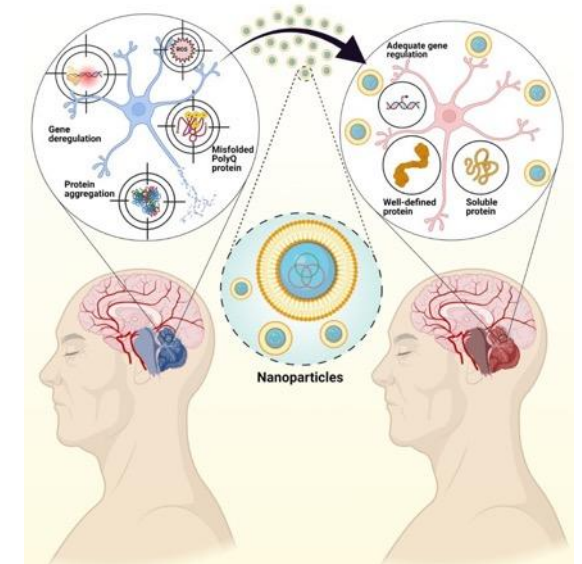
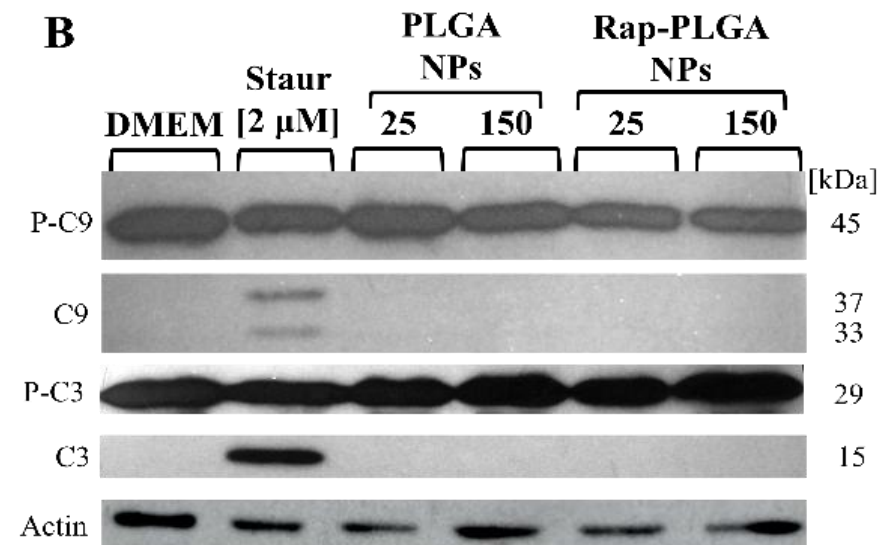
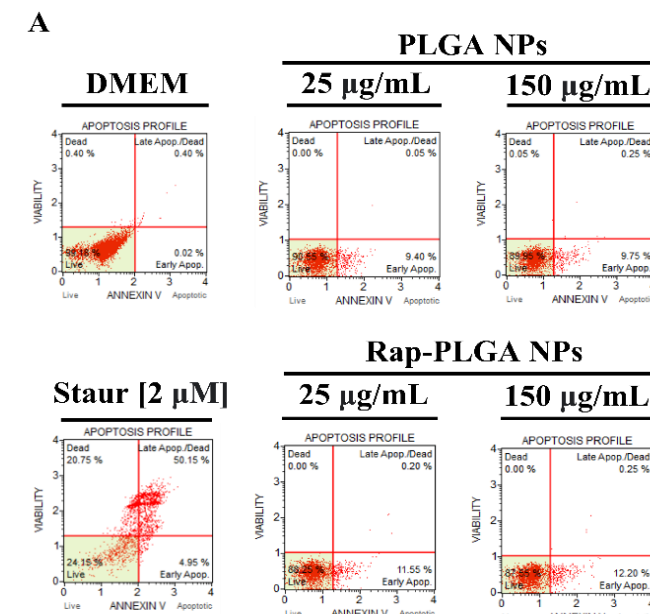


5 μm

Q64



Dr. Ian A. García Aguirre





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- Dra. Claudia Rangel Escareño

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- Dr. Zbigniew K. Wszolek

THANK YOU!!!!

