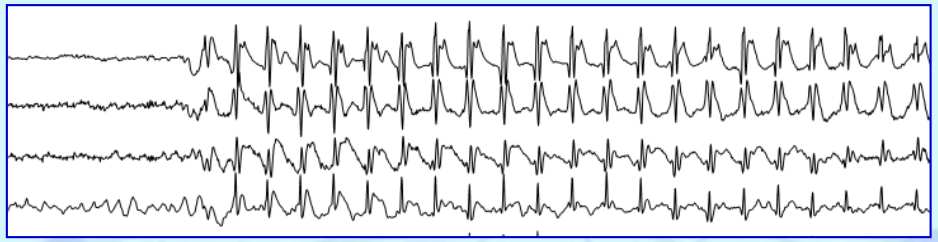
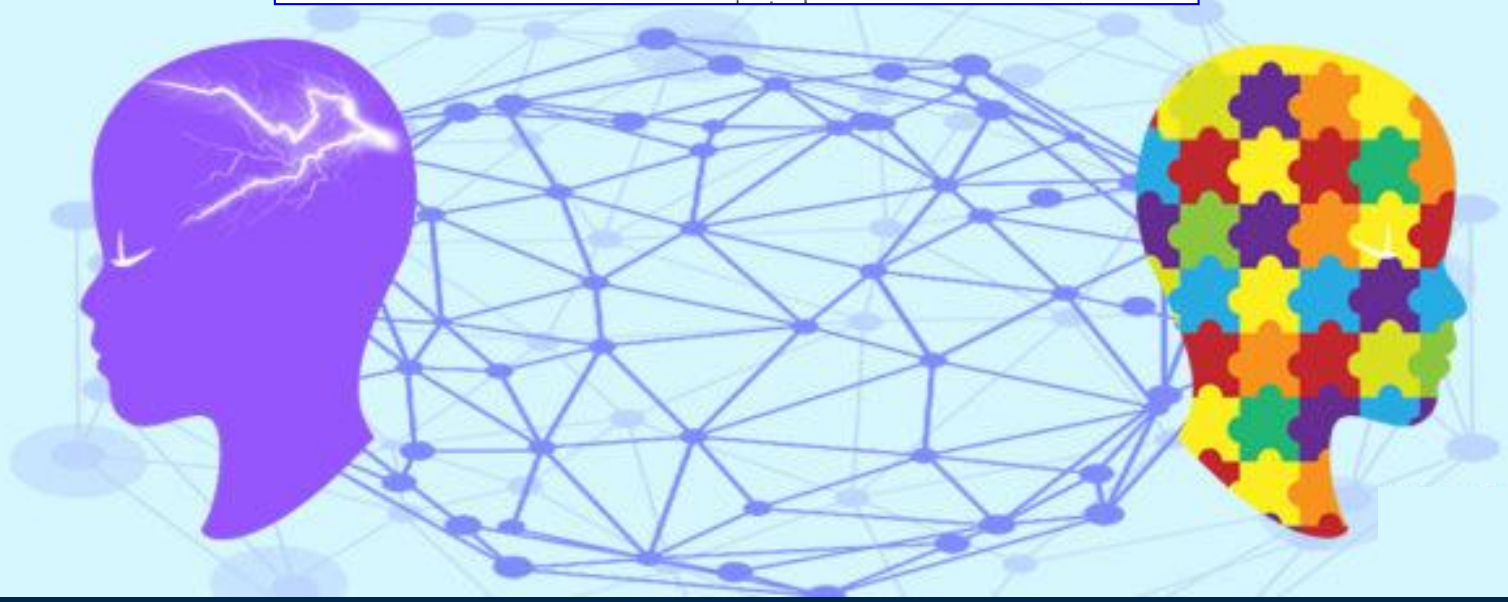
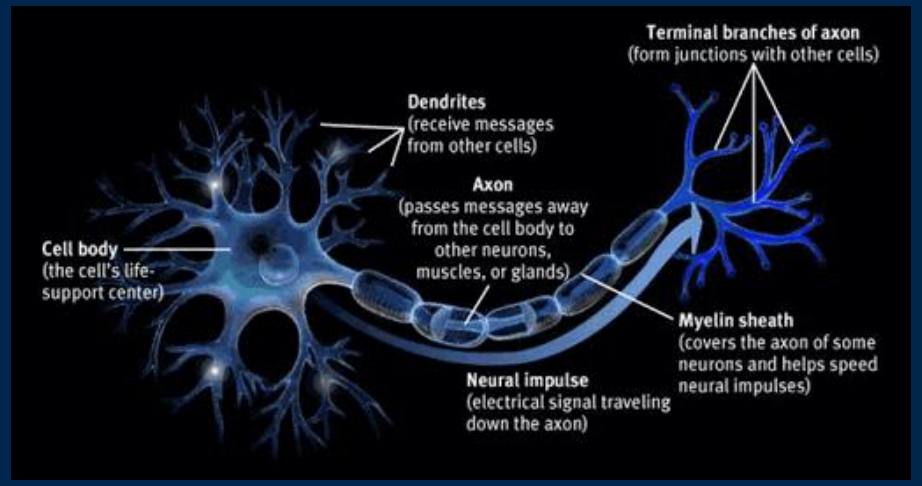
AUTISMO & INCLUSIONE
9 APRILE 2022

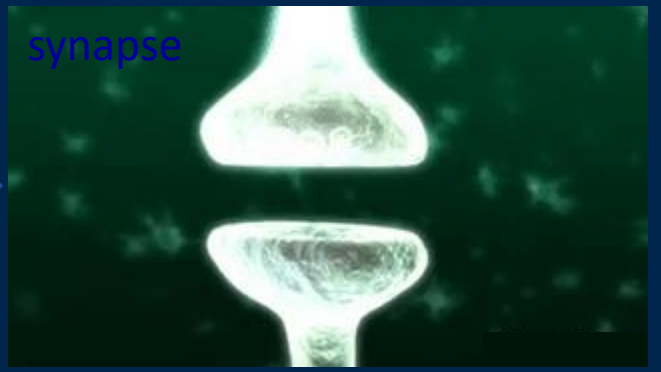
EEG, EPILESSIA E DISTURBI DELLO SPETTRO AUTISTICO



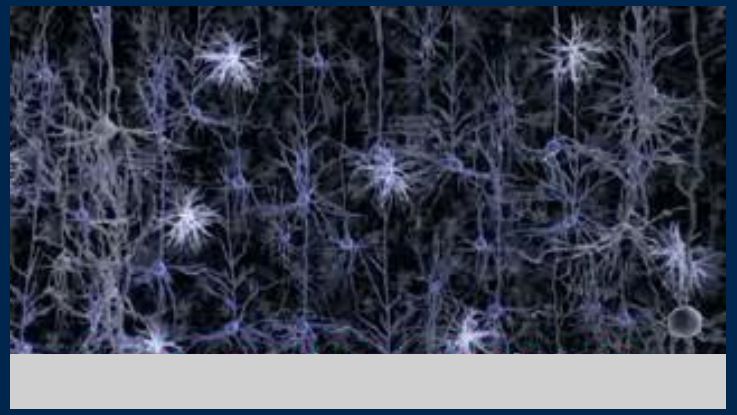


Federico Sicca
Neuropsichiatra Infantile
SERVIZIO EPILESSIA INFANZIA E ADOLESCENZA
USL TOSCANA CENTRO

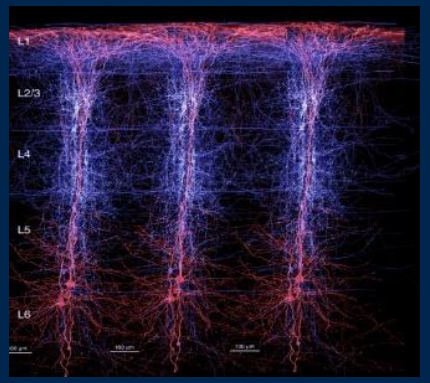




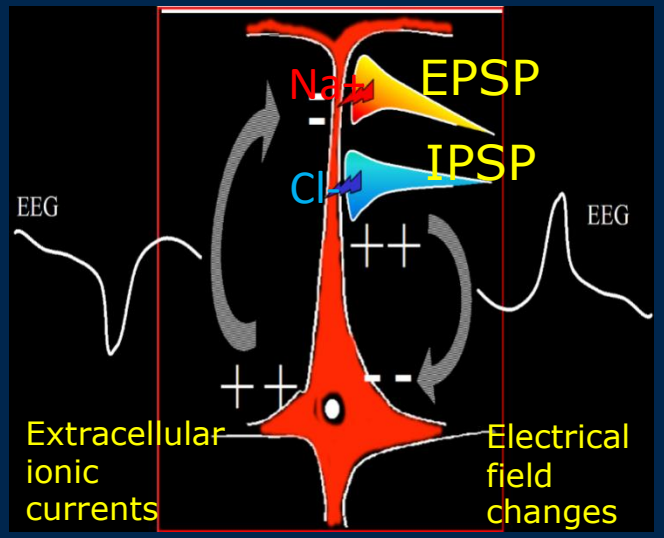
To neural population



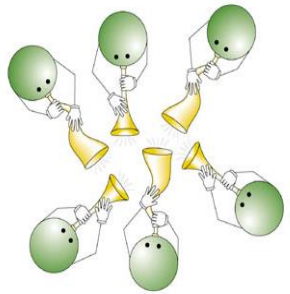
A single pyramidal cell:
> 10^4 synapses



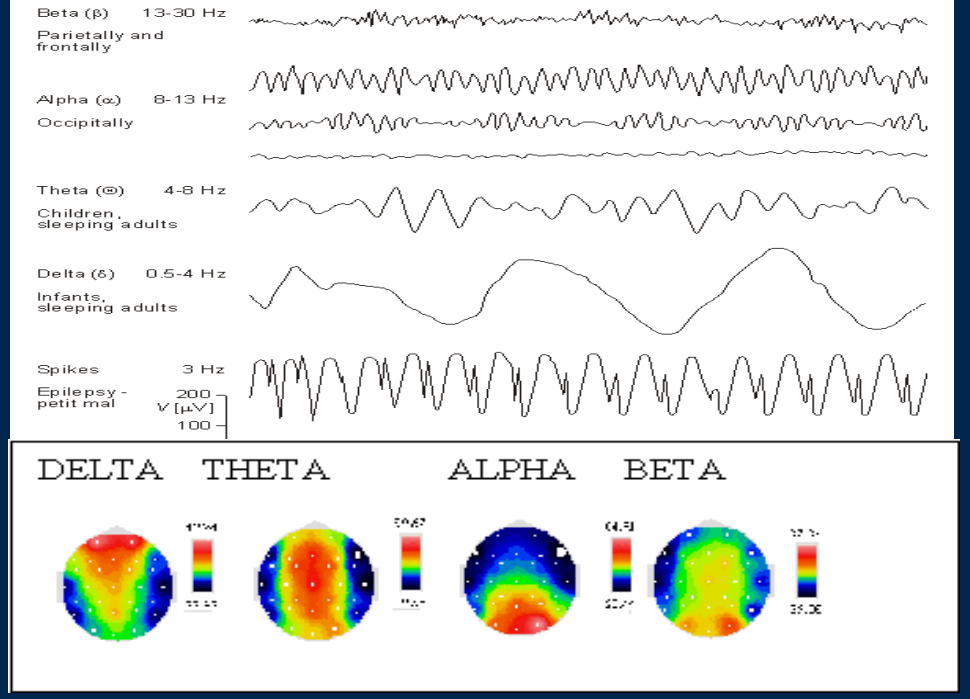
From single neuron

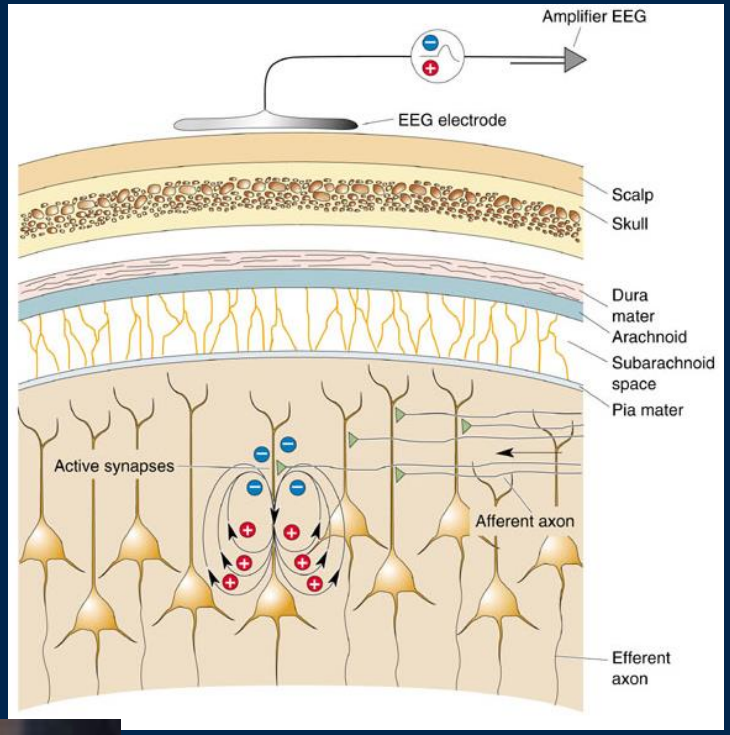


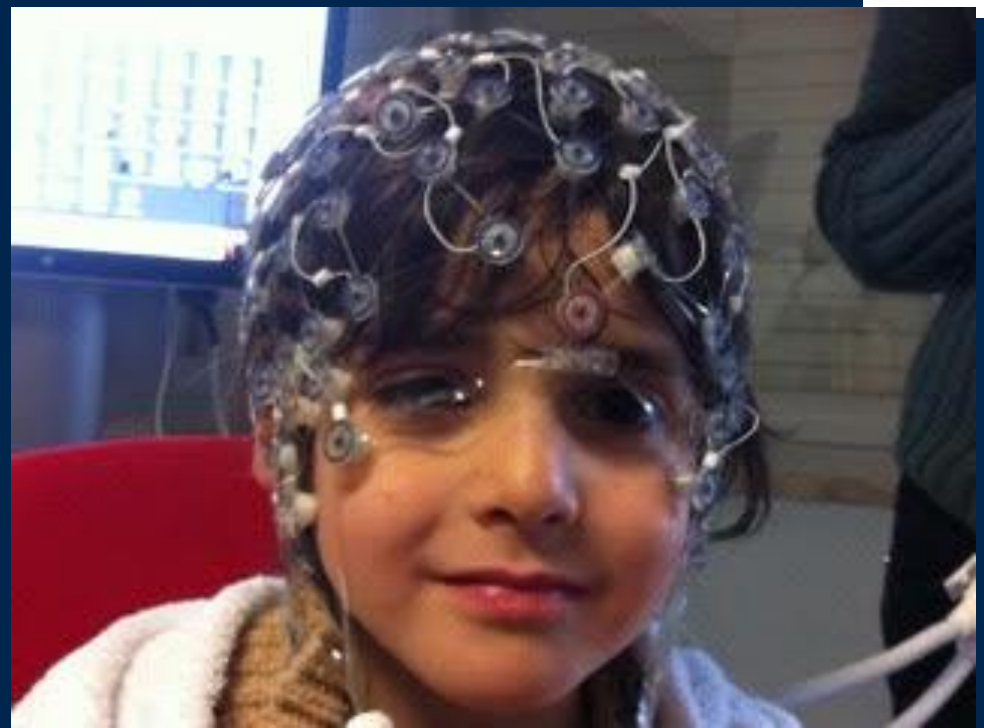
Sincronizzazione reciproca



Pacemaker







APPLICAZIONE EEG NELL' ASD

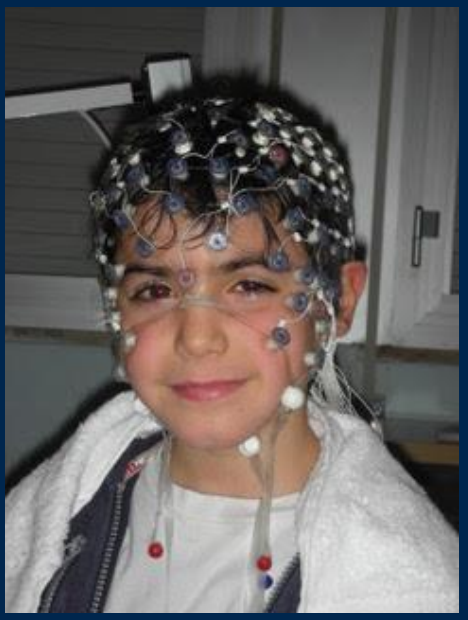
Autism is a disorder of neural development characterized by impaired social interaction and communication...

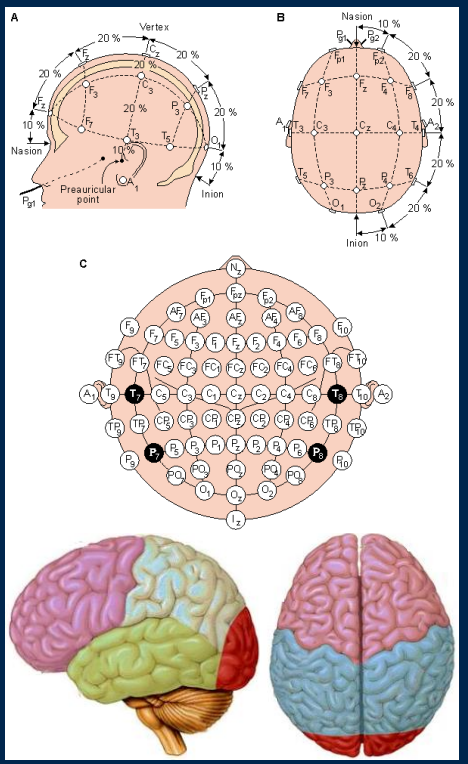
?

ASPETTI PSICOFISIOLOGICI

ASPETTI NEUROFISIOLOGICI

ASPETTI PSICOFISIOLOGICI





EEG QUANTITATIVO (Q-EEG) - ANALISI SPETTRALE

- FREQUENZA
- AMPIEZZA
- POTENZA (POWER)
  - ASSOLUTA: ESTREMA VARIABILITÀ
  - RELATIVA: MINORE VARIABILITÀ

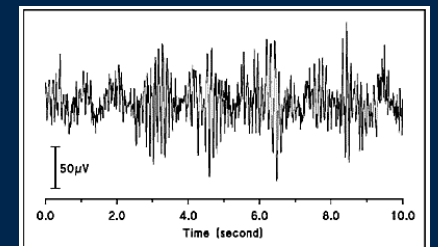
ERP – POTENZIALI EVENTO CORRELATI

VARIAZIONI DEL POTENZIALE ELETTRICO DERIVANTI DA UNO STIMOLO RIPETUTO (VISIVO, SOMESTESICO O UDITIVO)

CONNETTIVITÀ FUNZIONALE

- COERENZA: CONCORDANZA FASE TRA COPPIE DI ELETTRODI PER CIASCUNA BANDA FREQUENZA - COMPORTAMENTO DINAMICO NELLO SPAZIO E NEL TEMPO
- SINCROSTATI: STATI BEN DEFINITI E STABILI (MSEC) DI SINCRONIZZAZIONE DI FASE OSSERVATI DURANTE L'ESECUZIONE DI UN TASK DI PERCEZIONE DI VOLTI

ANALISI MATEMATICA: FAST FOURIER TRANSFORMATION (FFT)

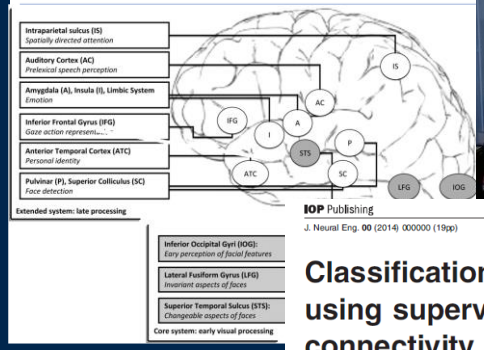


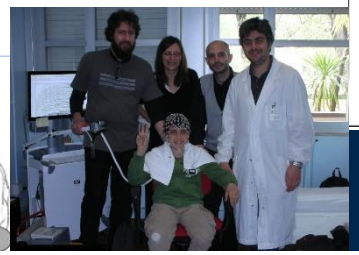
Research report

Fusiform Gyrus responses to neutral and emotional faces in children with Autism Spectrum Disorders: a High Density ERP study

Fabio Apicella[a,*], Federico Sicca[a], Rosario R. Federico[b], Giulia Campatelli[a], Filippo Muratori[a,b]

[a] Stella Maris Scientific Institute, Viale del Tirreno, 331, I-56018, Calambrone, Pisa, Italy
[b] Division of Child and Adolescent Psychiatry and Neurology, University of Pisa, Italy



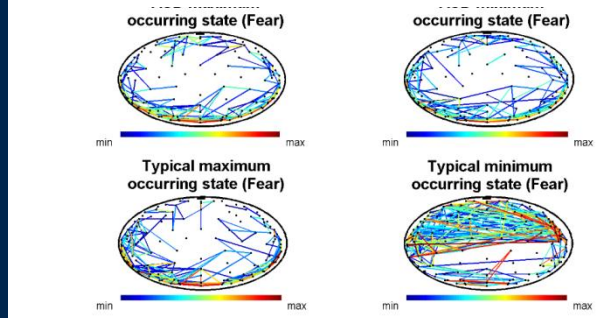


IOP Publishing
Journal of Neural Engineering
J. Neural Eng. 00 (2014) 000000 (19pp)

Classification of autism spectrum disorder using supervised learning of brain connectivity measures extracted from synchrostates

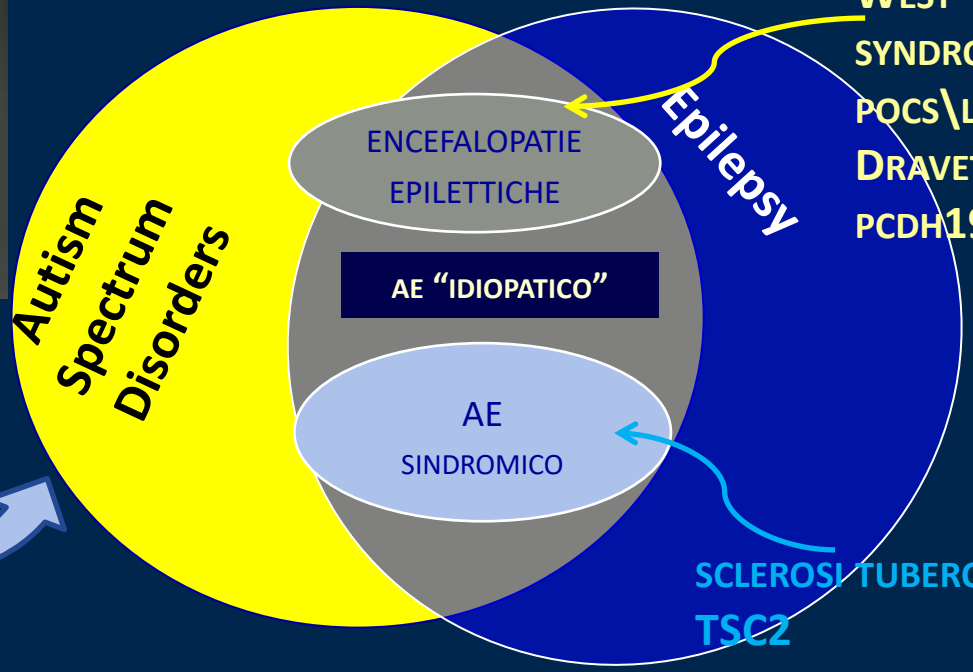
Wasifa Jamal[1], Saptarshi Das[1], Ioana-Anastasia Oprescu[1], Koushik Maharatna[1], Fabio Apicella[2] and Federico Sicca[2]

[1] School of Electronics and Computer Science, University of Southampton, Southampton SO17 1BJ, UK
[2] IRCCS Stella Maris Foundation, Pisa (Calambrone), Italy



ASPETTI NEUROFISIOLOGICI DELL' ASD

ANALISI PREVALENTEMENTE VISIVA



WEST SYNDROME
POCS\LKS
DRAVET
PCDH19

SCLEROSI TUBEROSA
TSC2
X-FRAGILE
RETT SYNDROME\CDKL5

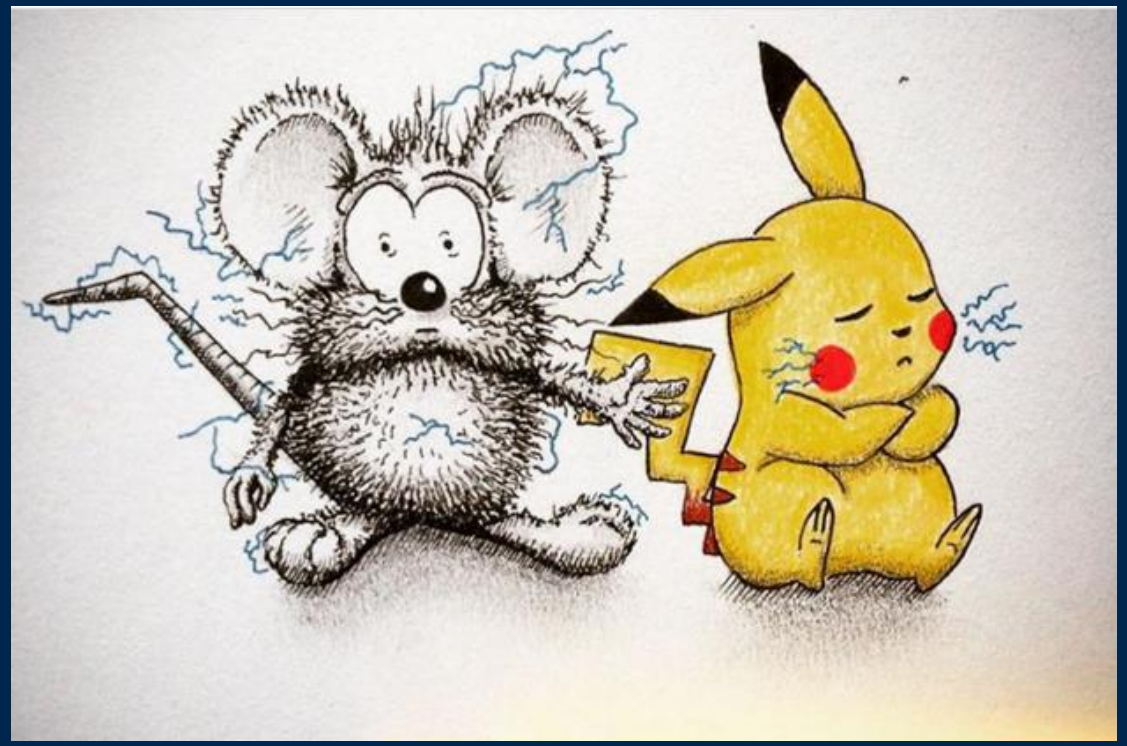
TERAPIA

Sclerosi tuberosa → spasmi infantili

mTORc inhibitors

vigabratin

- CRISI EPILETTICHE IN ASD: 5-46 %
- ANOMALIE EEG IN ASD: 6-75%
- ASD IN PAZIENTI CON EPILESSIA: 32%



DISABILITÀ INTELLETTIVA (ID):

PREVALENZA EPILESSIA 6-8% (NO ID) VS 21-40 % (ID)

GENERE FEMMINILE: F:M = 2:1 (ID F>M)

ETÀ: DISTRIBUZIONE BIMODALE

PICCO PRECOCE: < 5 ANNI

PICCO TARDIVO: > 10 ANNI

ENDOFENOTIPI CLINICI

OPEN ACCESS Freely available online

PLOS ONE

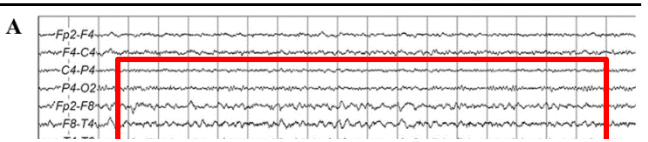
Somatic Overgrowth Predisposes to Seizures in Autism Spectrum Disorders

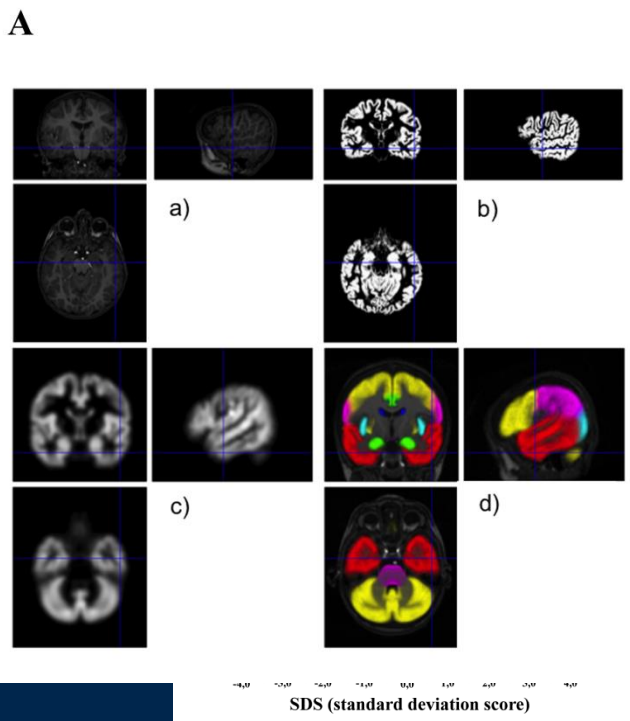
Giulia Valvo
Renzo Guerr
Raffaella Tar

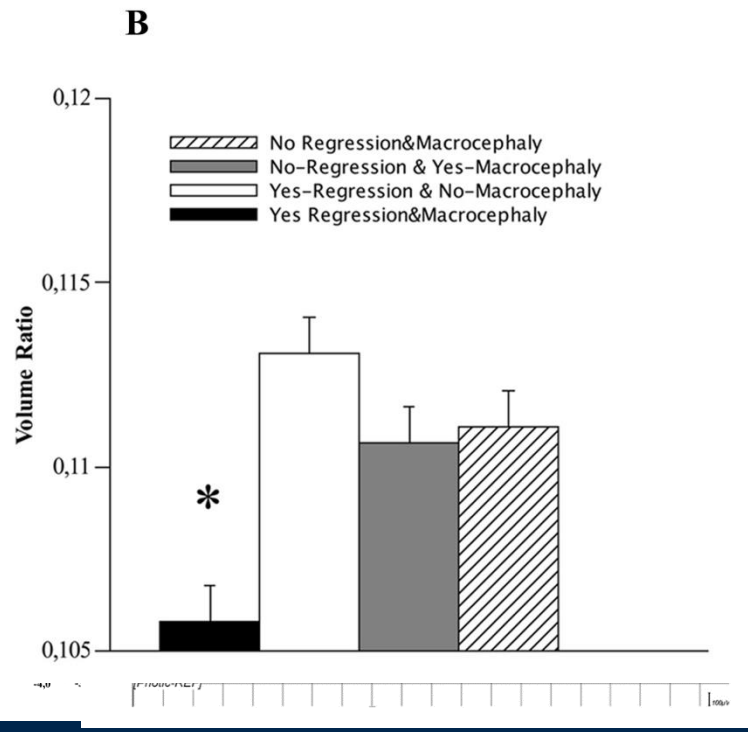
Eur Child Adolesc Psychiatry
DOI 10.1007/s00787-015-0746-9

CrossMark

ORIGINAL CONTRIBUTION







IDENTIFICAZIONE DI POSSIBILI MECCANISMI GENETICI

MECCANISMI GENETICI CONDIVISI

Cytoband	Maximum coordinates	Phenotype	Deletion/duplication
1q21.1	144.9–146.2	ASD, E, ID, SCZ	Deletion/duplication
3q29	197.1–198.9	ASD, E, ID, SCZ	Deletion/duplication
7q11.23	71.9–74.2	ASD, E, ID, SCZ	Deletion/duplication
15q11.2–13.1	21.1–26.2	ASD, E, ID, SCZ	Duplication
15q13.3	28.2–30.7	ASD, E, ID, SCZ	Deletion/duplication
16p11.2	29.4–30.1	ASD, E, ID, SCZ	Duplication
16p11.2	29.4–30.1	ASD, E, ID	Deletion
16p13.11	14.6–18.7	ASD, E, ID, SCZ	Deletion/duplication
17q12	31.5–33.1	ASD, E, ID, SCZ	Deletion/duplication
22q11.2	16.9–20.6	ASD, E, ID, SCZ	Deletion

sm spectrum disorders: Are there
evelopmental mechanisms?

Amy Brooks-Kayal *

Brain & Development 32 (2010) 731–738

University of Colorado Denver School of Medicine, The Children's Hospital Denver, Avenue, B155, Aurora, CO 80045, United States

Table 1 Genes implicated in autism, epilepsy and/or intellectual disability

Gene	Locus	Mutation	Transmission	Phenotype	Protein
SCN1A	2q24	Point mutation	*De novo*	ASD, E, ID	$Na_v1.1$ (Na^+ channel)
		Deletion	Dominant inheritance		
SCN2A	2q23–q24.3	Deletion	*De novo*	ASD, E, ID	$Na_v1.2$ (Na^+ channel)
		Point mutation	Inherited		
SCN3A	2q24	Deletion	*De novo*	E, ID	$Na_v1.3$ (Na^+ channel)
SCN1B	19q13.1	Point mutation	Dominant inheritance	E	β_1 subunit (Na^+ channel)
KCNA1	12p13	Point mutation	Dominant inheritance	E, ID	$K_v1.1$ (K^+ channel)
KCNQ2	20q13.3	Point mutation	Dominant inheritance	E, ID	$K_v7.2$ (K^+ channel)
		Deletion	*De novo*		
KCNQ3	8q24	Point mutation	Not known	E	$K_v7.3$ (K^+ channel)
KCNMA1	10q22	Point mutation	Dominant inheritance	ASD, E, ID	$K_v1.1$ (K^+ channel)
			De novo		
CACNA1A	19p13	Point mutation	*De novo*	E, ID	$Ca_v2.1$ (Ca^{2+} channel)
			Dominant inheritance		
GABRA1	5q34–q35	Point mutation	Dominant inheritance	E	α_1 subunit ($GABA_A$ receptor)
GABRG2	5q34	Point mutation	Dominant inheritance	E, ID	γ_2 subunit ($GABA_A$ receptor)
CHRNA2	8p21	Point mutation	Dominant inheritance	E	α_2 subunit (nACh receptor)
CHRNA4	20q13.2–q13.3	Point mutation	Dominant inheritance	E, ID	α_4 subunit (nACh receptor)
			De novo		
CHRNB2	1q21	Point mutation	Dominant inheritance	E	β_2 subunit (nACh receptor)
			De novo		
NLGN3	Xq13.1	Point mutation	Inherited	ASD	Inhibitory synapse formation
NLGN4	Xp22.31	Point mutation	Inherited	ASD, ID	Synapse formation
		Deletion			
CDH8	16q21	Deletion	Inherited	ASD	Synapse formation
PCDH10	4q28.3	Deletion	Inherited	ASD	Synapse formation
PCDH19	Xq22	Deletion	*De novo*	E, ID	Synapse formation
		Point mutation	Inherited		
NRXN1	2p16.3	Deletion	Recessive inheritance	ASD, E, ID, SCZ	Synapse formation
		Point mutation	*De novo*		
CNTNAP2	7q35	Deletion	Recessive inheritance	ASD, E, ID, SCZ	Synapse formation

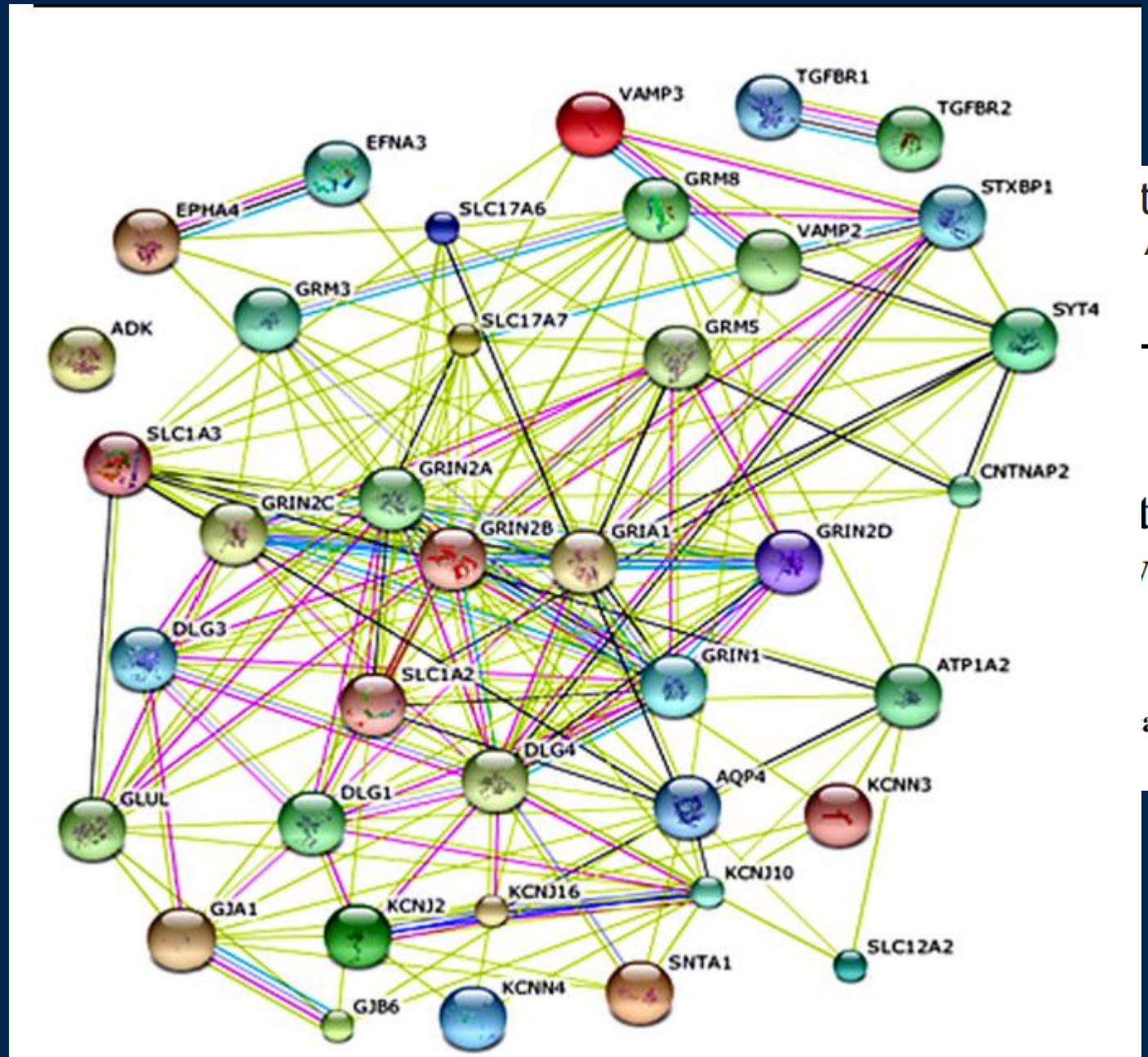
FUNZIONE SINAPTICA

CRESCITA CELLULARE

PLASTICITÀ CEREBRALE

ECCITABILITÀ CELLULARE

IDENTIFICAZIONE DI POSSIBILI MECCANISMI GENETICI



th macrocephaly
AM genetic

CrossMark

to Explore
Phenotype

a^2 ·

IDENTIFICAZIONE DI POSSIBILI MECCANISMI GENETICI

Neurobiology of Disease 43 (2011) 239–247

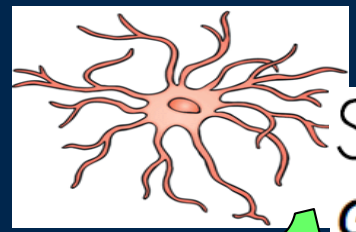
Contents lists available at ScienceDirect

Neurobiology of Disease

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

Autism with Seizures and Intellectual Disability: Possible Causative Role of *Gain-of-function* of the Inwardly-Rectifying K^+ Channel Kir4.1

.Kir4.1



.Kir2.1

Genetically induced dysfunctions of Kir2.1 channels: implications for short QT3 syndrome and autism–epilepsy phenotype

Human Molecular Genetics, 2014
doi:10.1093/hmg/ddu201

Elena Ambrosini[1,†,*], Federico Sicca[2,†], Maria S. Brignone[1], Maria C. D'Adamo[4], Carlo Napolitano[7], Ilenio Servettini[4], Francesca Moro[2], Yanfei Ruan[7], Luca Guglielmi[4], S Servillo[5], Angela Lanciotti[1], Giulia Valvo[2], Luigi Catacuzzeno[6], Fabio F

SCIENTIFIC REPORTS

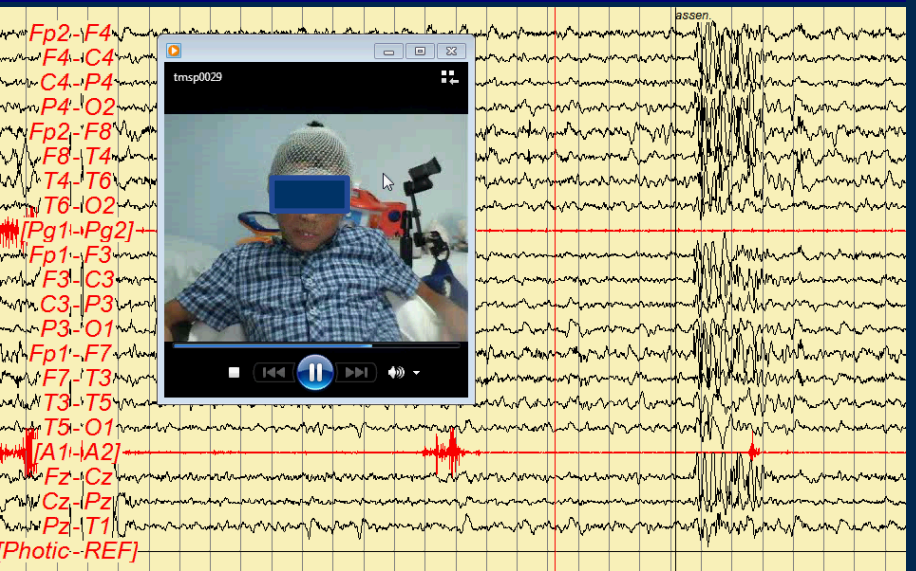
Gain-of-function defects of astrocytic Kir4.1 channels in children with autism spectrum disorders and epilepsy

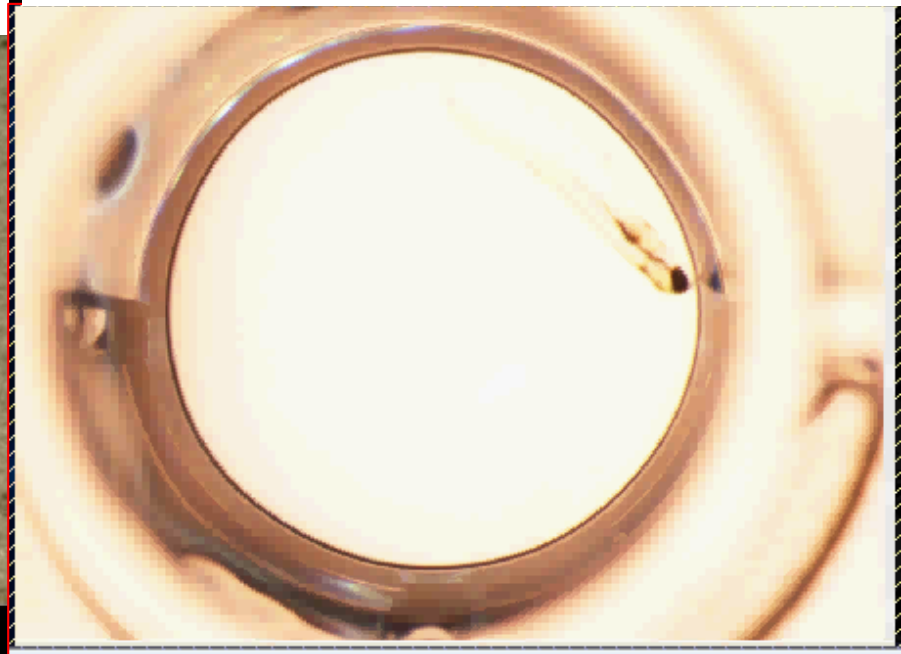
Federico Sicca[1,2,*], Elena Ambrosini[3,*], Maria Marchese[2], Luigi Sforna[4], Ilenio Servettini[4], Giulia Valvo[1], Maria Stefania Brignone[3], Angela Lanciotti[3], Francesca Moro[2], Alessandro Grottesi[5], Luigi Catacuzzeno[6], Sara Baldini[1], Sonia Hasan[4], Maria Cristina D'Adamo[4,7], Fabio Franciolini[6], Paola Molinari[8], Filippo M. Santorelli[2] & Mauro Pessia[4,7]

J Vet Intern Med 2014;28:871–877

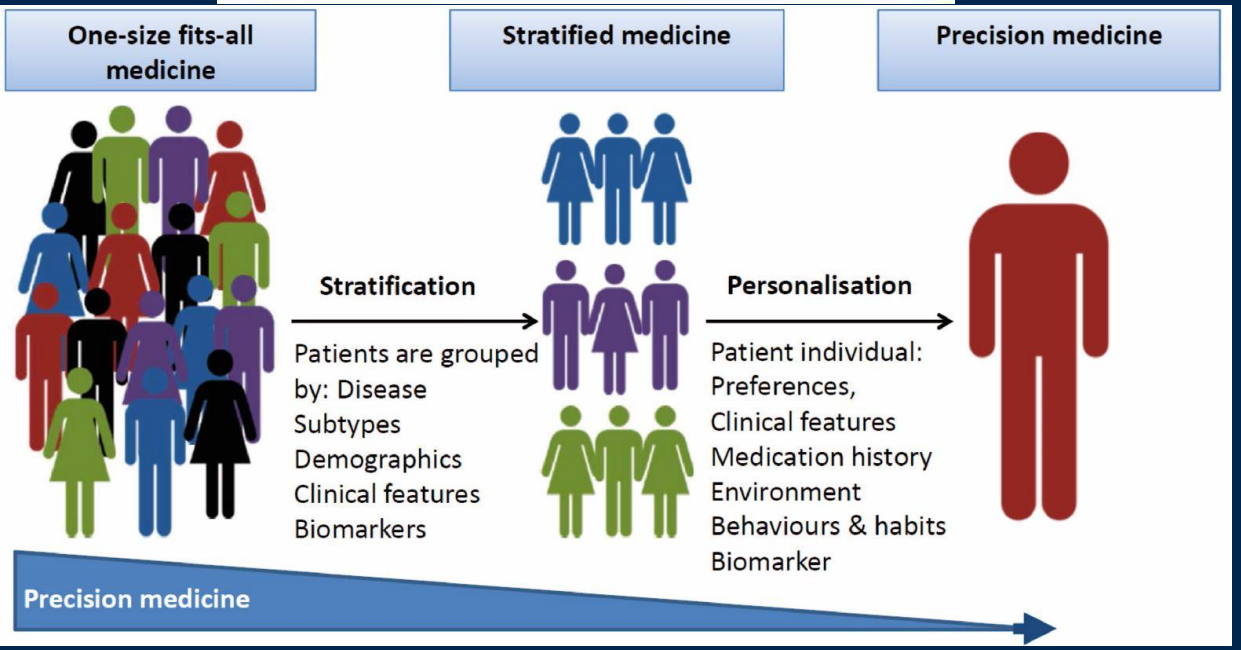
A Homozygous *KCNJ10* Mutation in Jack Russell Terriers and Related Breeds with Spinocerebellar Ataxia with Myokymia, Seizures, or Both

R18Q AEP with subtle seizures





IN CONCLUSIONE



GRAZIE PER L'ATTENZIONE