

Epilepsia partialis continua associated with the p.Arg403Cys variant of the *DNM1L* gene: an unusual clinical progression with two episodes of super-refractory status epilepticus with a 13-year remission interval

Sara Minghetti¹, Roberto Giorda¹, Massimo Mastrangelo², Laura Tassi³, Nicoletta Zanotta¹, Sara Galbiati¹, Maria Teresa Bassi^{1,*}, Claudio Zucca^{1,*}

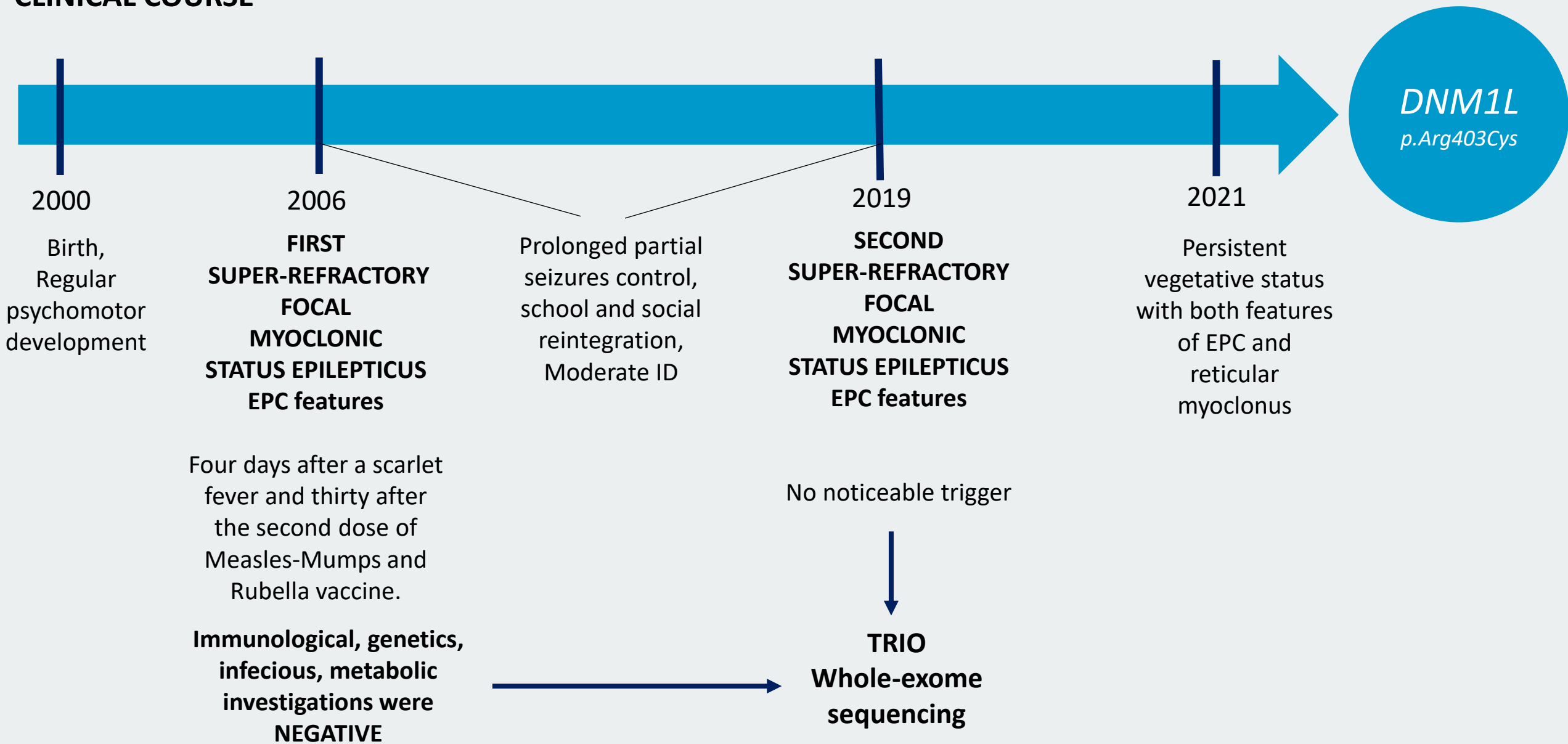
¹ Scientific Institute, IRCCS E. Medea, Bosisio Parini, Lecco, Italy

² Child Neurology Unit, V. Buzzi Children's Hospital, Milan, Italy

³ "Claudio Munari" Epilepsy Surgery Center, Niguarda Hospital, Milan, Italy

*Authors contributed equally

CLINICAL COURSE



NEURORADIOLOGICAL COURSE



2000
Birth
Regular
psychomotor
development

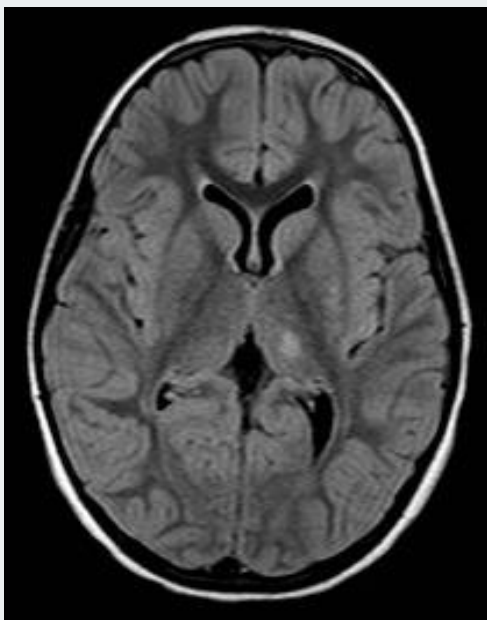
**FIRST
SUPER-REFRACTORY
STATUS EPILEPTICUS**

Partial seizures controlled,
school and social
reintegration,
Moderate ID

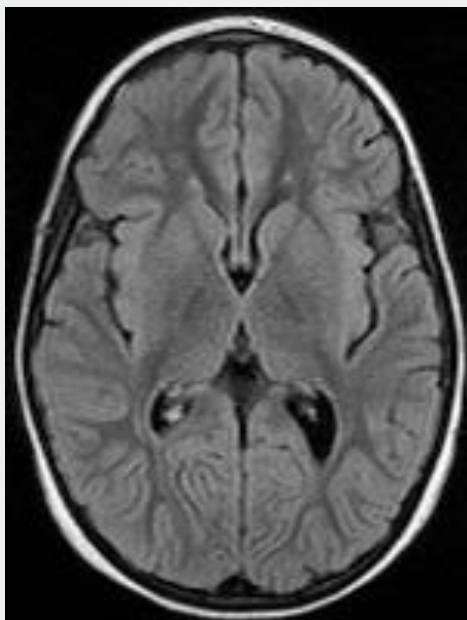
**SECOND
SUPER-REFRACTORY
STATUS EPILEPTICUS**

Persistent
vegetative status

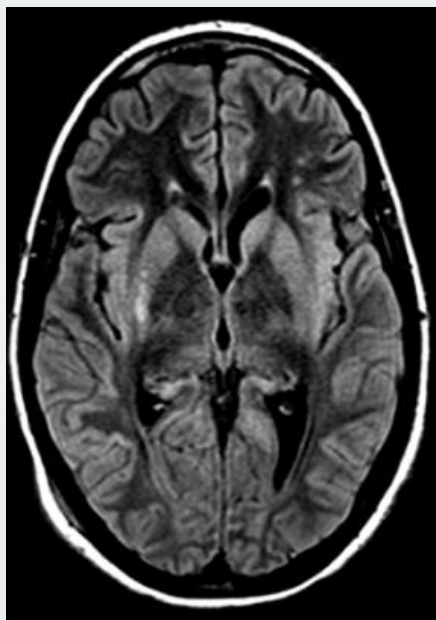
DNM1L
p.Arg403Cys



T2 FLAIR hyperintensity on the left posterior thalamus and putamen



Complete resolution of the FLAIR hyperintensity

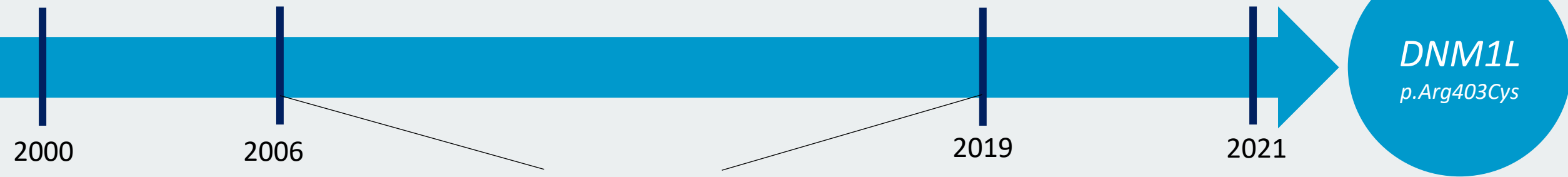


T2-FLAIR hyperintensity on the right capsular, thalamic, posterior lentiform nucleus, frontal areas and centro-parietal cortex



Bilateral dorso-medial thalamic hyperintensity

ELECTROENCEPHALOGRAPHIC COURSE



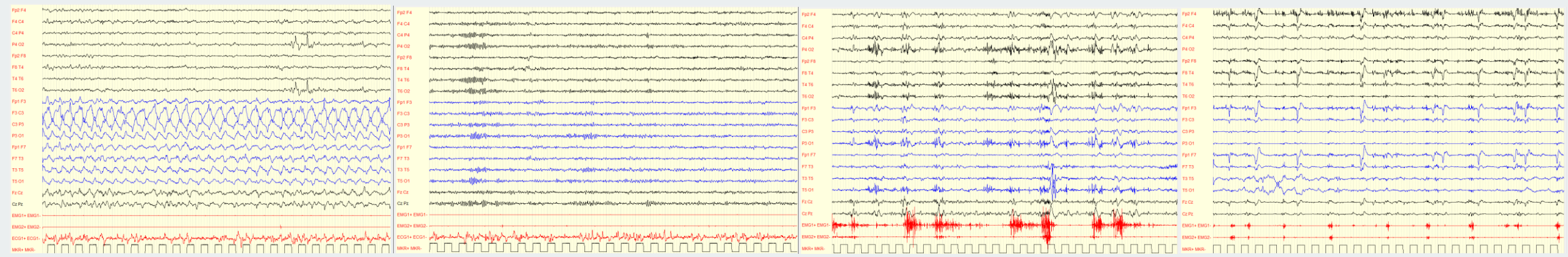
Birth
Regular psychomotor development

**FIRST
SUPER-REFRACTORY
STATUS EPILEPTICUS**

Prolonged partial seizures controlled, school and social reintegration, Moderate ID

**SECOND
SUPER-REFRACTORY
STATUS EPILEPTICUS**

Persistent vegetative status with epilepsy partialis continua



Asymmetry of background activity with continuous slow paroxysmal activity on the left frontal regions (March 2006)

11-Hz background activity without asymmetry and without epileptiform discharges recorded (January 2019)

Irregular background activity (8-9 Hz) with polyspike-wave discharges on the right frontal region; myoclonic phenomena associated with diffuse polyspike-wave discharges, prevalent on the right fronto-central region (April 2020)

Irregular, low-amplitude background activity (6-7 Hz) with small groups of diffuse polyspike-wave discharges inconstantly associated with myoclonic phenomena, mostly observed on the right arm (November 2020)

CLINICAL

Childhood-onset super-refractory focal myoclonic status epilepticus with features of EPC

Risk of relapse after prolonged partial seizure control

Muscle biopsy not pathognomonic

ELECTROENCEPHALOGRAPHIC

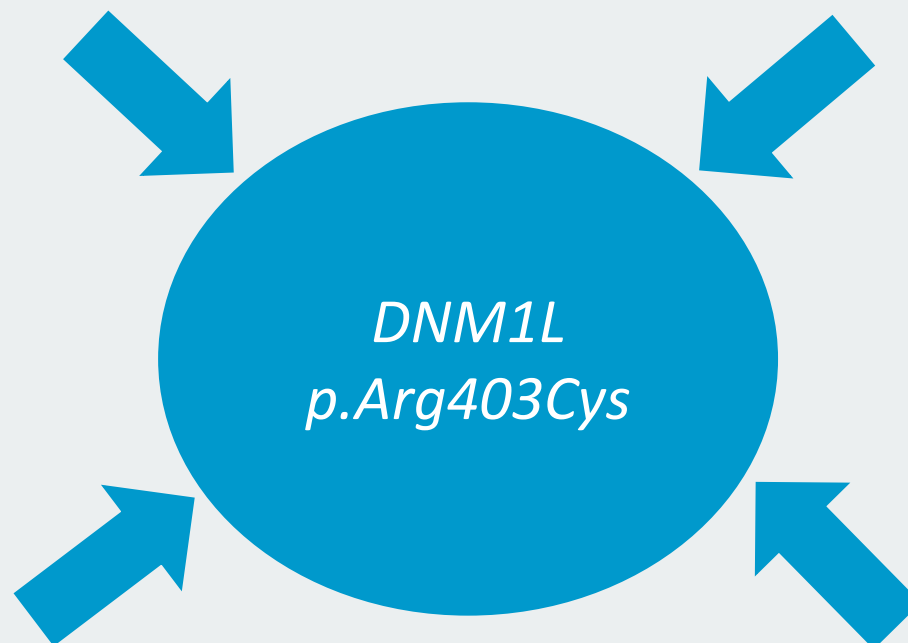
This mutation could be considered as a new EPILEPSIA PARTIALIS CONTINUA etiology

Shifting focal hemispheric refractory status epilepticus

EEG progressive normalization during the remission interval

Coexistence of both epileptic and non epileptic myoclonic phenomena

TAKE HOME MESSAGE



NEURORADIOLOGICAL

Transient and shifting T2 FLAIR thalamic hyperintensity during status

Progressive brain atrophy

GENETICS

Whole-exome sequencing is pivotal in childhood-onset super-refractory status epilepticus

Functional studies are required to investigate this unusual biphasic disease course