Original article

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Phenotypes of children with 20q13.3 microdeletion affecting *KCNQ2* and *CHRNA4*

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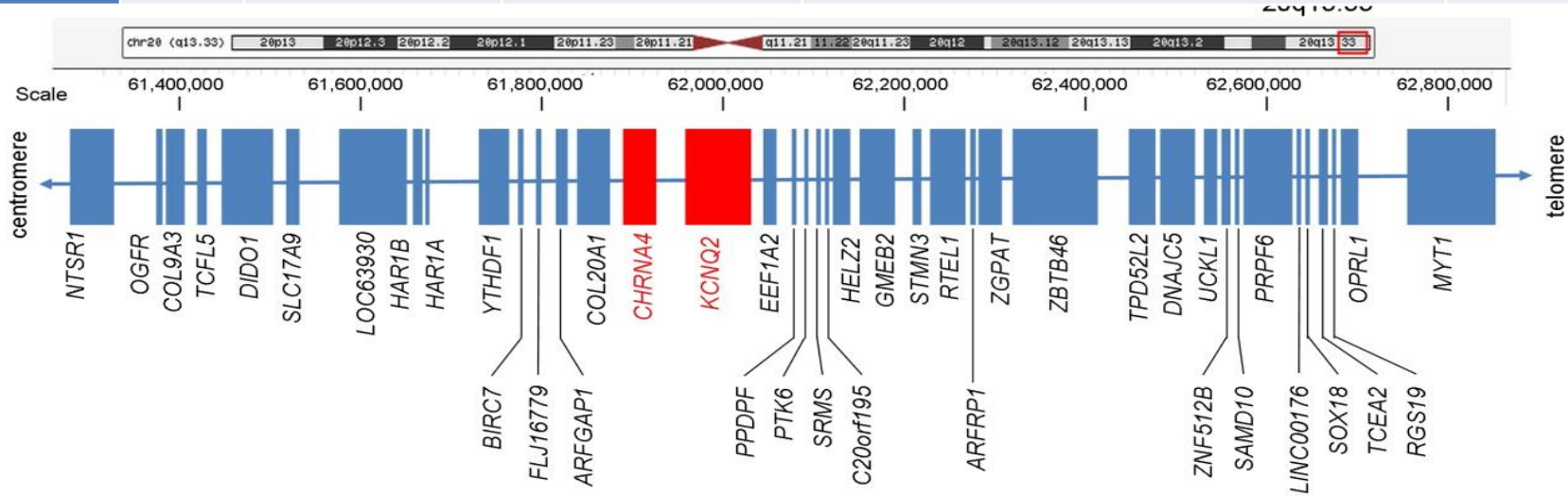
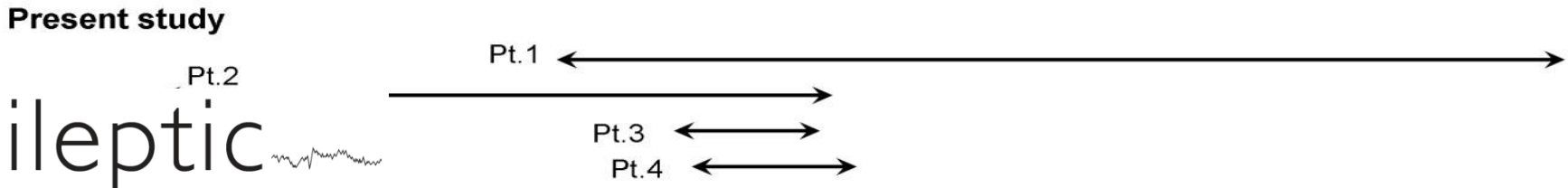
	Sex	Epi onset	Epi offset	Sz semiology	Febrile Sz
Patient 1	F	10 d	4 m	Asymmetric tonic posturing	No
Patient 2	M	3 m	4 m	Right hemiconvulsion	No
Patient 3	F	2 d	3 m	Reduced responsiveness	Yes
Patient 4	F	1 d	3 d	Hemiconvulsion	No





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Clinical features of epilepsy in patients with 20q13.3 microdeletion

	Deletion size	Epilepsy onset	Epilepsy offset	EEG	MRI	Treatment	Psychomotor development	Dysmorphic features
Patient 1	2.47 Mb	10d	4m	Normal	Normal	PB ineffective CBZ effective	Mild delay	None
Patient 2	1.09 Mb	3m	4m	Focal Spikes	Atrophic	PB effective	Mild delay	None
Patient 3	765 kb	2d	3m	Normal	Normal	PB ineffective ZNS effective	Normal	None
Patient 4	136.4 kb	1day	3d	Normal	Normal	None	Normal	None
Traylor et al., 2010 Subject 1	171.8 kb	6m	Single Sz	Normal	N/A	None	Global IQ 40	None
Subject 2	1.1 Mb	2w	Intractable	N/A	Delayed myelination	N/A	Severe delay	Present
Subject 3	1.61 Mb	None		N/A	N/A	N/A	Severe delay	Present
Subject 4	1.08 Mb	Yes	N/A	N/A	N/A	N/A	Delay	Present
Subject 5	560 kb	Yes	N/A	N/A	N/A	N/A	Delay	N/A
Subject 6	1.0 Mb	N/A	N/A	N/A	N/A	N/A	Expired	Present
Béri-Deixheimer et al., 2007 Patient 2	6.8 Mb	2m	Single Sz	Abnormal	Thin CC	None	Severe delay	Present
Mefford et al., 2012	1.6 Mb	2w	8w	hypsarrhythmia	Delayed myelination	PB and Vit B6 effective	Severe delay	None
Pascual et al., 2013 Patient 1	1.5 Mb	7d	1m	Transiently abnormal	Normal	LEV ineffective OXC effective	Mild delay	None
Patient 2	521 kb	2d	6m	Transiently abnormal	Normal	LEV, ZNS, TPM	Global delay	None
Patient 3	520.7 kb	2d	N/A	Multifocal Spikes	Normal	PB effective	Global delay	None

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The outcome of epilepsy in infants with 20q13.3 microdeletion affecting both *CHRNA4* and *KCNQ2* was favourable. Deletion of *KCNQ2* and *CHRNA4* does not appear to affect seizure phenotype.

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