

Manuscript Title: Expanding Phenotype of Poirier-Bienvenu Syndrome: New Evidences From An Italian Multicentric Cohort of Patients

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Hereby we provide the form used to collect data from each centre.

Centre		
Patient ID		
Birth date		
Sex		
GENETIC MUTATION		
Variant and Exon involved	Variant	Exon
Type of mutation (e.g.: nonsense, missense, frameshift...)		
Genetic analysis performed		
EPILEPSY		
Epilepsy onset (months)		
Epilepsy type		
Further epilepsy types showed		
DISABILITY		
Intellectual disability (specify scale adopted)		
Motor skills		
Hypotonia		

Dystonia	
Speech	
Communication disability	
Learning disability	
IMAGING AND EEG	
EEG at onset (if available)	
EEG during follow-up	
MRI	
Other imaging (if available)	
OTHER FEATURES	
Facial Dysmorphism	
Micro- /Macrocephaly (yes/no)	
Autistic features	
Immunological features	
Endocrinological features	
Cardiology features	
Gastrointestinal features	

Orthopedic features	
ENT features	
Other abnormalities	