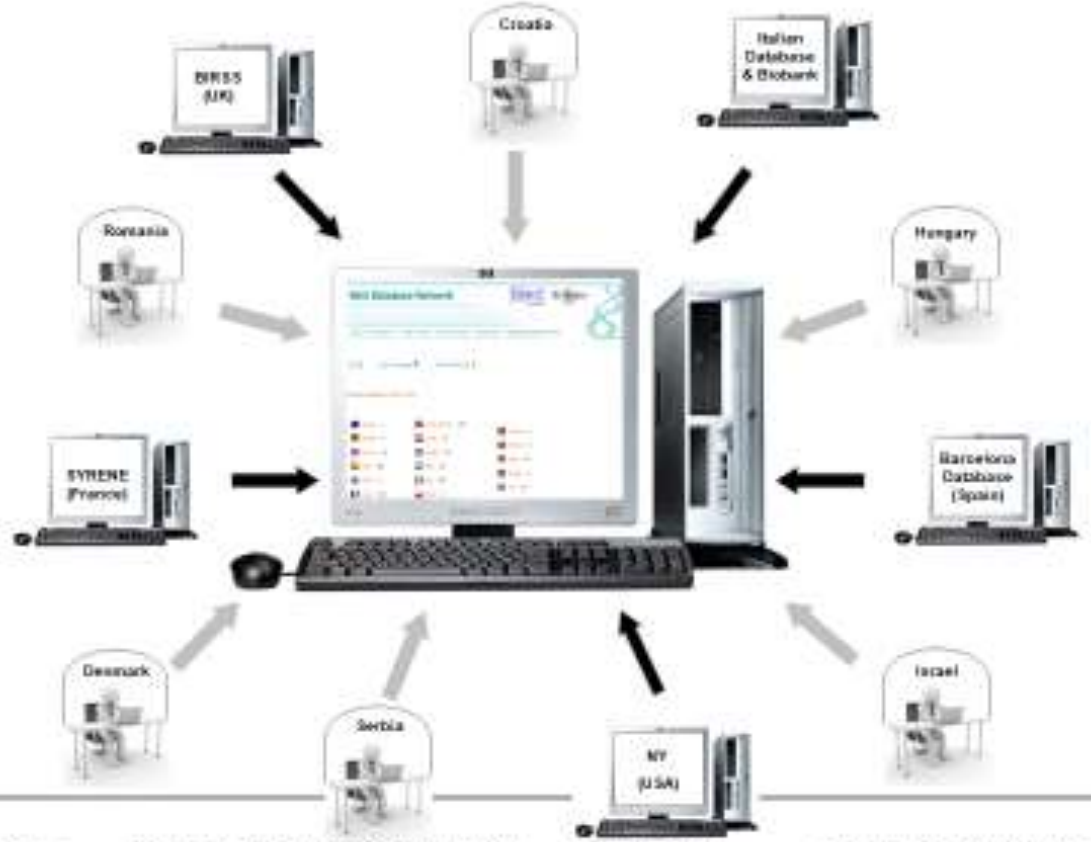
ILIAD Rare Diseases patient registry: an International Library of Intellectual disability and Anomalies of Development

Rett Syndrome Networked Database



Barcelona Rett Database		British Isles Rett Syndrome Survey (BIRSS)		and Biobank	SYndrome REtt NEtwork (SYRENE)		Rett Database Network
Stagnation head	Acquired microcephaly	OFC fall	OFC at present	Head	PC deceleration of head growth	PC at evaluation	Head score
Yes	Yes	1 = yes, any evidence of fall from original centile	percentile ≤ 3	2 = postnatal microcephaly	yes	percentile ≤ 3	2 = postnatal microcephaly
Yes	No		percentile > 3	1 = deceleration of head growth	yes	percentile > 10	1 = deceleration of head growth
					yes	percentile 3–10	
No	No	2 = no evidence of fall from original centile		0 = no deceleration	no		0 = no deceleration

OFC: Occipitofrontal circumference.

Rett Database Network

EuroRETT

RettSearch

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Number of patients in archive: 2116

Australia 1	France 252	Italy 713	Serbia 51
Croatia 29	Germany 0	Poland 0	Spain 434
Czech Republic 0	Hungary 92	Portugal 0	Sweden 0
Denmark 64	India 3	Romania 17	United Kingdom 255
Finland 0	Israel 93	Russia 16	USA 96

Clinical informations

From Diagnosis

Classical Rett

Atypical:
Early seizures variant
Congenital variant
Preserved speech variant

Rett-like phenotypes

.. To Core Data set (1)

Head
Weight
Height

Sitting
Walking
Hand skills
Speech
Phrase
Regression
Hand stereotypy
Intellectual disability

.. To Core Data set (2)

Gastrointestinal disturbances
Feeding
Sphincter control

Breathing disorders
Cold extremities

Epilepsy

Scoliosis
Cyfosis

Detailed genetic data

HYPOTHESIS OF AGGREGATED DATA INTEGRATION

RSND

ILIAD

1) Familial cases usefull for better understand disease molecular mechanism (to data 30)
   as in ILIAD «famBool»

2) Single items (core data set) for each gene to obtain descriptive data
   and genotype – phenotype correlations as in ILIAD «HPO»
   Ex. Microcephaly (postnatal/progressive) (430 *MECP2*, 20 *CDKL5*, 30 *FOXG1)*
   Intellectual disability (severe, moderate/mild, profound)
   Epilepsy (260 *MECP2*, 60 *CDKL5*, 15 *FOXG1)*

3) Epilepsy related therapy adopted as in ILIAD «ATC»

2) Single items (core data set) for each gene to obtain descriptive data and genotype – phenotype correlations as in ILIAD «HPO»

Ex. Microcephaly (postnatal/progressive) (430 *MECP2*, 20 *CDKL5*, 30 *FOXG1)*
Intellectual disability (severe, moderate/mild, profound)
Epilepsy (260 *MECP2*, 60 *CDKL5*, 15 *FOXG1)*

Rett Syndrome Networked Database	ILIAD Registry	
Postnatal Microcephaly	Postnatal Microcephaly	HPO
Hand stereotypy	Stereotypy hand wringing	HPO
Never spoken	Absent speech	HPO
Speech (more than 10 words at age of 5)	Poor speech	HPO
Never learned to walk/loss of ability to walk	Inability to walk	HPO
Epilepsy	Epilepsy	HPO
Myoclonic epilepsy	Myoclonic epilepsy	HPO
Scoliosis	Scoliosis	HPO
Kyphosis	Kyphosis	HPO
Intellectual disability QI>40	Intellectual disability moderate	HPO
Intellectual disability severe	Intellectual disability severe	HPO
Intellectual disability profound	Intellectual disability profound	HPO
Gastroesophageal reflux	Gastroesophageal reflux	HPO
Breathing disorders (Apnea, Hyperventilation)	Apnea	HPO
	Hyperventilation	HPO
Regression	Developmental regression	HPO
	Motor regression	HPO
Genu valgu/pes planus	Genu valgu/pes planus	HPO
Autism	Autism	HPO