RETT SYNDROME

Request form
And clinical informations

Medical secretariat :
Phone : +33 1 34 40 20 80
Fax : +33 1 34 40 20 85
e-mail : smedical@lab-cerba.com
website: www.lab-cerba.com

The L**aboratory is authorized according to the french legislation to perform genetic testing**

PATIENT	REFERRING PHYSICIAN
First Name..... Last Name..... Address..... .... Date of birth/Gender :.....	Signature :

REQUESTED TEST	INDICATION	
	☐ Male patient	☐ Female patient
☐ *MECP2* gene analysis	☐ Classical Rett syndrome ☐ Atypical Rett syndrome ☐ X-linked mental retardation (Xq28) ☐ Neonatal encephalopathy ☐ Syndromic mental retardation (hypotony, ataxia, spasticity….)	☐ Classical Rett syndrome ☐ Atypical Rett syndrome ☐ Angelman syndrome ☐ Autistic syndrome ☐ Other non specific mental retardation ☐ Familial study : mother of an affected girl ☐ Familial study : asymptomatic sister of an affected girl
☐ *CDKL5* gene analysis	☐ Severe/early onset epileptic encephalopathy ☐ Atypical Rett syndrome (infantile seizure onset)	☐ Severe/early onset epileptic encephalopathy ☐ Atypical Rett syndrome (infantile seizure onset)
☐ *FOXG1* gene analysis	☐ Classical Rett syndrome ☐ Congenital variant of Rett syndrome +/- MRI abnormalities	☐ Classical Rett syndrome ☐ Congenital variant of Rett syndrome +/- MRI abnormalities

CLINICAL INFORMATIONS (according to Neuln et al Ann Neurol 2010 ;68:944-950)

Exclusion criteria and Required criteria for typical or classic RTT

No	Yes		
☐	☐	Normal prenatal and perinatal criteria	
☐	☐	No brain injury secondary to another etiology	
☐	☐	Normal psychomotor development in first six months of life	
☐	☐	Partial or complete loss of acquired purposeful hand skills	Age at onset :
☐	☐	Partial or complete loss of acquired spoken language	Age at onset :
☐	☐	Stereotypic hand movements	Age at onset :
☐	☐	Gait abnormalities : Impaired (dyspraxia) or absence of ability	Age at onset :

Other criteria

No	Yes	
☐	☐	Breathing disturbances when awake
☐	☐	Bruxism when awake
☐	☐	Impairment sleep pattern
☐	☐	Abnormal muscle tone - Spasticity
☐	☐	Peripheral vasomotor disturbances
☐	☐	Scoliosis - kyphosis
☐	☐	Growth retardation
☐	☐	Small cold hands and feet
☐	☐	inappropriate laughing or screaming spells
☐	☐	Diminished reponse to pain
☐	☐	Intense eye communication – “eye pointing”
☐	☐	Epilepsy
☐	☐	Infantile spasms
☐	☐	Hypotonia
☐	☐	Cerebral abnormalities at MRI
☐	☐	Testicular atrophy
☐	☐	Recurrent infections
☐	☐	Communication dysfunction, automutilation
☐	☐	Postnatal deceleration of head growth
☐	☐	Microcephaly

OTHER INFORMATIONS AND PEDIGREE :