



Université catholique de Louvain

Cliniques universitaires Saint-Luc

association sans but lucratif



Centre de Génétique Humaine UCL : Prof Miikka Vikkula

Clinical booklet

Tuberous sclerosis

Index patient

last name

Patient's first name

Date of birth (DD/MM/YYYY)

Sex: Female or male

Ethnic origin e: Caucasian /Asian/ north Africa/South Africa /

Please notify country and city of origin

Name of the referring geneticist :

Adress

E-mail

Phenotype index patient

Dermatological :

examination done / no done

If done normal/ abnormal

If abnormal please comment :

Ophthalmological : examination done / no done

If done normal/ abnormal

If abnormal , please comment :

Brain/Cerebral (imaging) : : examination done / no done

If done normal/ abnormal

If abnormal please comment :

Heart/Cardiac : ultrasound examination done / no done

If done normal/ abnormal

If abnormal, please comment:

Kidney/Renal : ultrasound exam done / no done

If done normal/ abnormal

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If abnormal , please comment :

Bone cyst : examination done / no done

If done normal/ abnormal

If abnormal, please comment :

Possible familial presentation based on,parents ? grand-parents history ?

If it's , please mention the family tree and related affected (?)

Conclusion based on hereabove/available information : Sporadic form Familial form

Natural history

Prenatal Echographic anatomic anomalies

First trimester : increased nuchal lucency : Yes/ no

Second trimester : Intra cardiac rhabdomyoma Yes / No

Growth retardation ? : if yes , please specify : centile and age at onset

Oligoamnios / polyhydramnios

Hyperechoic Kidneys/ presence of cyst(s)

Third trimester : Growth retardation

Cerebral anomalies ; if yes, **please comment**

On ultrasound or fetal IRM

Other anomalies ; if yes , **please comment**

Postnatal

Prematurity (birth < 37 weeks) yes / no

Weight in grams Percentile

Height in cm Percentile

Head circumference (OFC) Percentile

Dermatological evaluation

Wood's light Examination

Hypo/Amelanotic spots : Yes / No / not done

Angiofibroma : Yes / No / not done If yes : no treatment / surgery

Front plaque : Yes / No / not done

Shagreen plaque : Yes / No / not done

Ophthalmological evaluation

Retinal hamartoma : Yes / No If yes , age at diagnosis

Neurological evaluation

Delayed psychomotor development

If yes , please clarify : Sitting at (age in month)

Walking at (age in month)

Mental retardation

If yes :

IQ and age at of the time of testing

Special education ? if yes, please comment

Personality Disorder

Please comment or use the DSMIV classification
Epilepsy
Age at onset
Presence of Infantile spasms ? yes/no/ do not know
Controlled epilepsy vs refractory to treatments

Epilepsy requiring surgery Yes /No / not applicable

Cerebral imaging (MRI)

Anomalies : if yes , subependymal nodules/tuber/abnormal migration

Renal evaluation

Angiomyolipoma : yes /no/ not screened
If yes , size in cm at the moment of the diagnostic
Proteinuria : yes /no/not screened
Hematuria : yes / no / not screened
Polykystosis : Yes / no /not screened
renal failure : yes / no / not screened

Cardiac evaluation(postnatal)

rhabdomyoma If yes , please note how many
therapy

Index case patient : living/dead
If dead , please notify: age and cause

Diagnostic criteria Adapted from Yates *Europ J Hum Genet* 2006; 14: 1067

	Major criteria	Minor criteria
Dermatological	1) facial angiofibroma or frontal plaque 2) fibroma of tongue (non traumatic) 3) ≥3 hypomelanotic macules (amelanotic spots) 4) "shagreen" plaque	Ski lesions "in confetti"
Ophtalmological	Multiple retinal nodular hamartoma	amelanotic retinal pellet
Cerebral	1) cortical tuber 2) subependymal nodule 3) giant cells astrocytoma (subependymal)	abnormal migration of white matter (neuronal migration anomaly)
Cardiaque	Rhabdomyoma (single or numerous)	
Renal	Renal angiomyolipoma or lymphangiomyomatosis	Multiple cyst
Dentition / oral cavity		1) enamel tooth anomalies (multiple pits) 2) gingival fibroma

Mis en forme : Anglais
(Royaume-Uni)

Digestif		colon hamartomatous polyposis
Bone		Bone cyst
Vascular		Hamartoma (other than renal)

Confirmed clinical diagnosis iff there's two major criteria or 1 major criteria and 2 minor criteria

Probable diagnosis iff there 's 1 major criteria and 1 minor criteria

Possible diagnosis iff there's 1 major criteria