

**SERVICE DE GÉNÉTIQUE MOLÉCULAIRE,
PHARMACOGÉNÉTIQUE ET HORMONOLOGIE**

**SERVICE DE GÉNÉTIQUE MOLÉCULAIRE,
PHARMACOGÉNÉTIQUE ET HORMONOLOGIE**

Clinical information Card : Calcium and phosphorus metabolism

Chef de Service :

Pr Anne GUIOCHON-MANTEL

**GÉNÉTIQUE MOLÉCULAIRE ET
PHARMACOGÉNÉTIQUE**

Dr Jérôme BOULIGAND – MCU-PH
☎ :01.45.21.35.88

Dr Bruno FRANCOU - PHC
☎ :01.45.21.38.45

Courriel : prenom.nom@aphp.fr

Patient label	Prescriber (Senior mandatory)
	First and last name: _____ Hospital: _____ Department: _____ _____ Address: _____ _____ Phone: _____ Fax: _____ Email: _____

1/ Extracted patient:

Is the individual affected? YES – NO

Is he/she the propositus? YES – NO

If not, what is the family relationship with the propositus?

Propositus identity: First and last name - gene and mutation

In which laboratory was the first analysis made?

2/ Family history

Are there similar symptoms in one or more relatives? YES – NO

Family tree at the back of the page indicating the propositus and relatives (First and last name, date of birth) and their state (affected/ not affected)

3/ Genetic hypothesis

Do you suspect a specific gene? :GNAS/ PHEX/ SHOX/ FGFR3/ Other:

OR

Do you suspect methylations alterations?

Do you suspect PHP1b? Silver-Russellsyndrome? Beckwith-Wiedemannsyndrome?

OR

Suspected diagnosis (written out):

4/ Circle the matching item(s):

Diagnostic framework: Rickets – Hypophosphatemia – Disorder of Vitamin D metabolism – Hypophosphatasia – Primary hyperparathyroidism – Familial hypocalciuric hypercalcemia – Hypercalciuria/ Nephrocalcinosis – Hypoparathyroidism – iPPSD (pseudo-hypoparathyroidism – Acrodysostosis) – Achondroplasia – Dyschondroplasia – TrichoRhynoPhalangeal syndrome – Osteopetrosis – Osteogenesis imperfecta

Clinical signs and symptoms: Short stature – Obesity – Brachymetacarpals – Subcutaneous ossifications – No subcutaneous ossifications – Calcifications of the cerebral ganglia – Lithiasis – Bone abnormality (AHO?) – Recurrent fractures – Dental abnormality – Cryptorchidism – Hormonal resistance – Other

Biological parameters: Please write ↗ or ↘ or N (Normal) in the interpretation cell) *Join a report if possible*

	Calcemia	Phosphatemia	Calciuria	Phosphatasia	PTH	1,25 OHD	25 OHD	TSH	Creatinine urine
Value									
Units	mmol/L	mmol/L	mmol/L	mmol/L	pg/mL	pg/mL	ng/mL	mUI/L	mmol/L
Normal range									
Interpretation									

Other relevant information: