

BACKGROUND

Reports of Cushing syndrome during the first month of life are rare; Mortality is high, despite medical (metopyrone) or surgical (adrenalectomy) treatment.

OBJECTIVE : To report a new neonatal case of Cushing due to Mc Cune Albright syndrome (MAS)

CASE REPORT



Figure 1 : Clinical Manifestations

Table 1 : Biologicals parameters at diagnosis

Calcémie	3,55
Phosphore	0,77
Mg	0,55
Glycémie	11,5
Lactates	3,4
ASAT	45
ALAT	68
Bilirubine	127
GGT	258
TA (mmhg)	140/78
FC (/min)	143

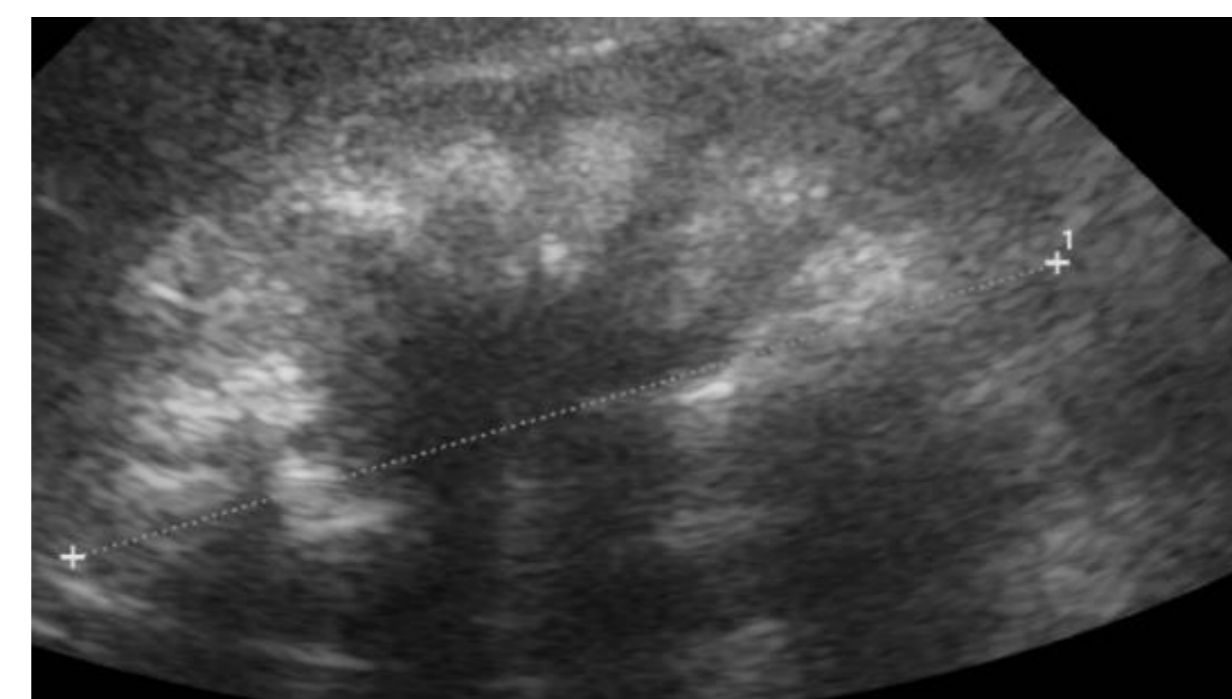


Figure 2 : Nephrocalcinosis

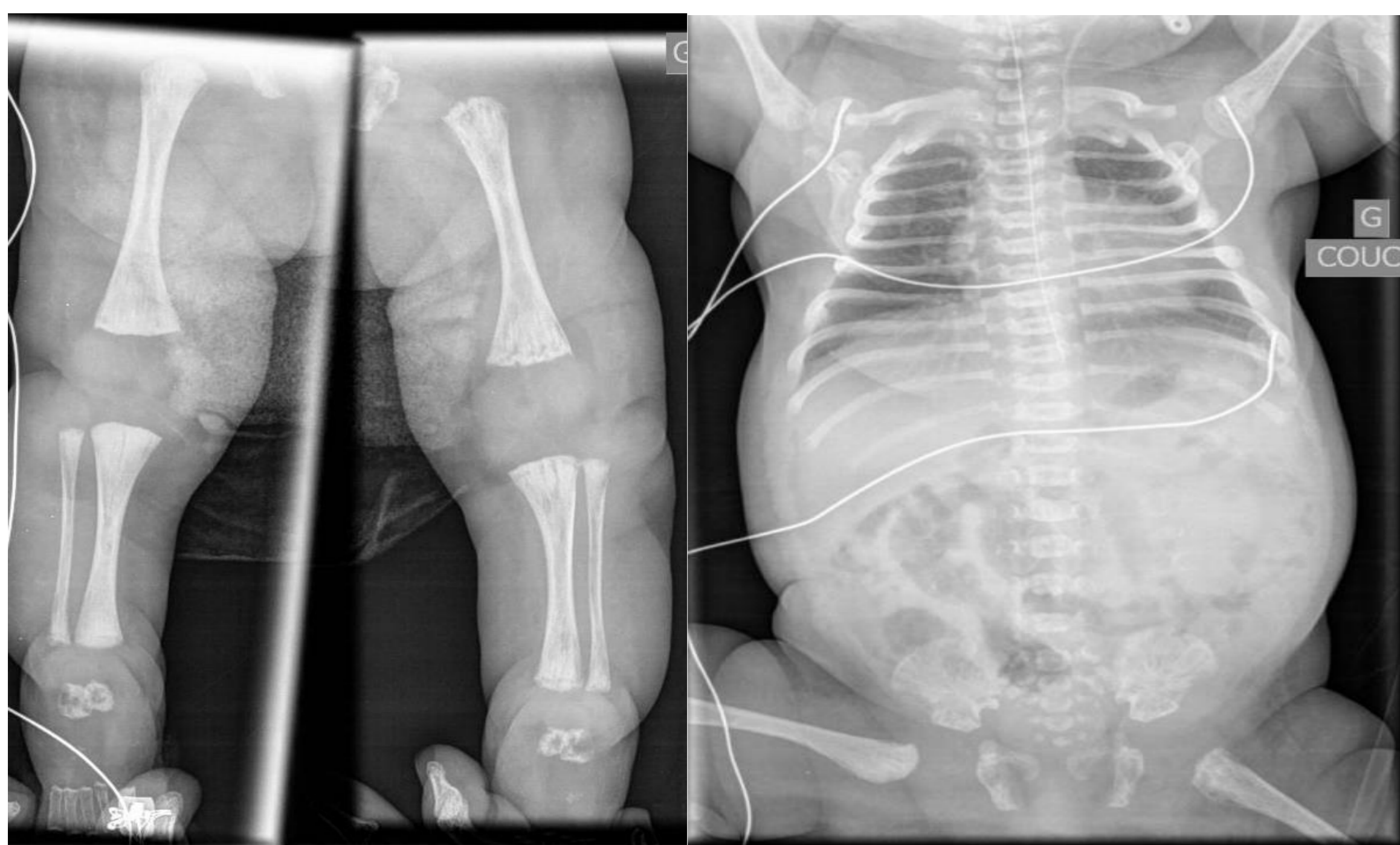


Figure 3 : Bone disostosis

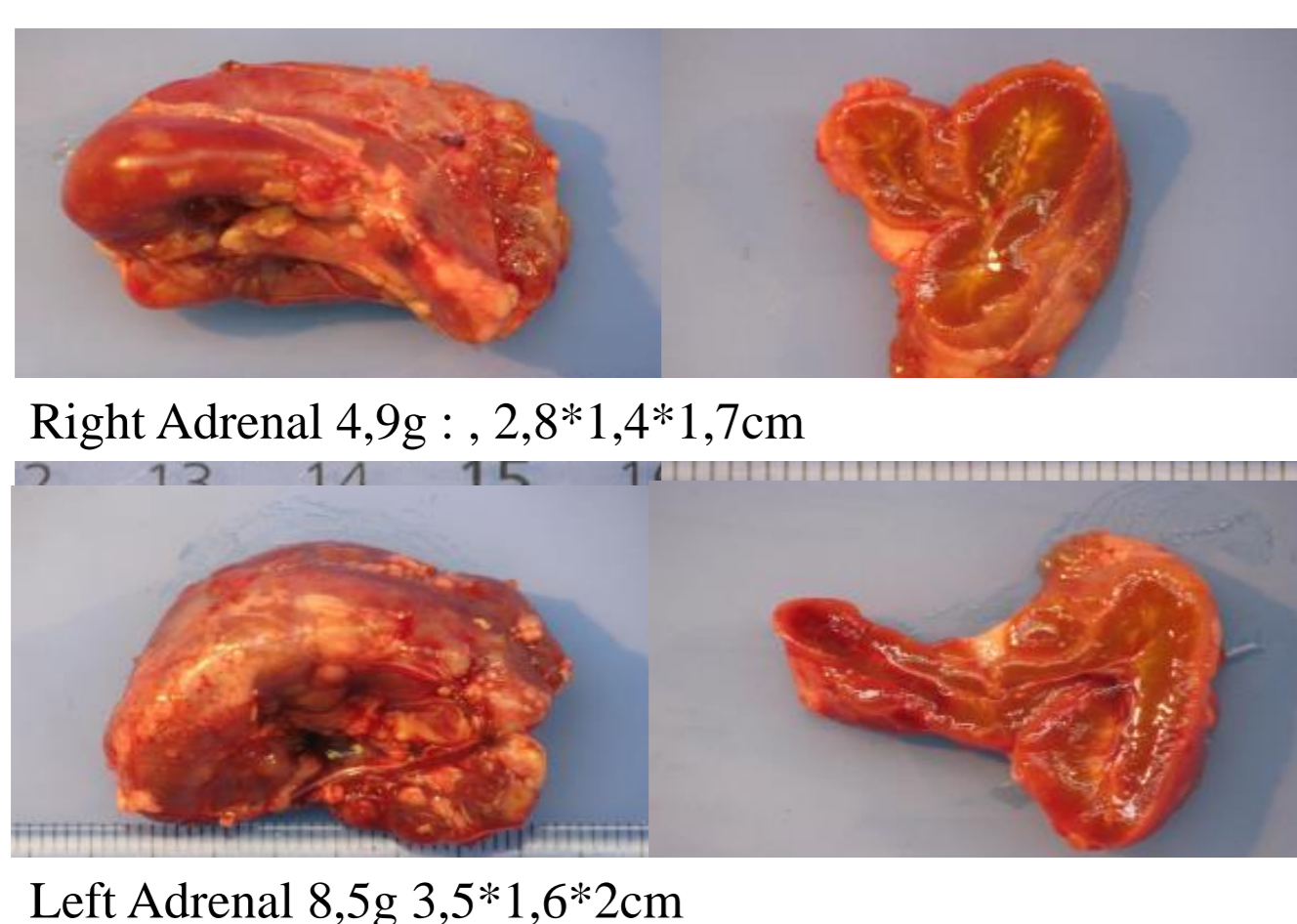


Figure 4 : Bilateral Adrenalectomy

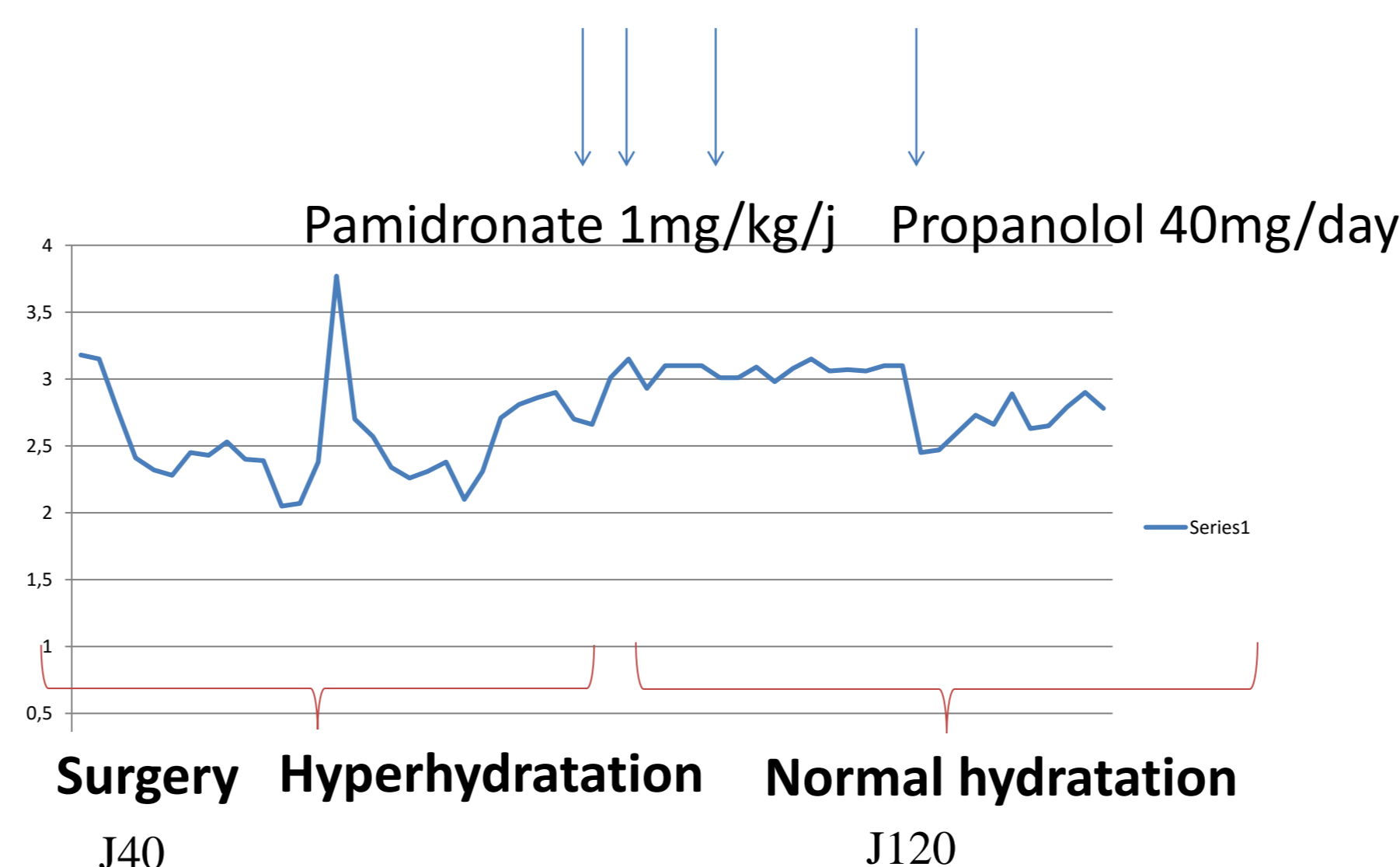


Figure 5 : Evolution of calcemia



Figure 6 : Large café-au-lait spots

Although a healthy baby at age 10 days, a new born girl presented with sudden manifestations within the following 2 weeks : facial truncal plethora (Figure 1), severe hypertension, cardiomyopathy with ventricular hypertrophy, hyperglycemia, elevated transaminases, major hypercalcemia with hypercalciuria (Table 1) and nephrocalcinosis (Figure 2), bone dysostosis (left femur, forearms) (Figure 3), and large bilateral adrenal hyperplasia.

Laboratory data confirmed adrenocorticotrophic hormone-independent Cushing's syndrome with plasma cortisol 975 ng/ml.

The baby girl underwent bilateral adrenalectomy at 40 days (Figure 4).

Medical treatment included glargine insuline (3-4 units/d) and propranolol (40mg/d), which allowed the control of hyperglycemia, hypercalcemia (Figure 5), hypertension and cardiomyopathy. The baby received 10mg/d hydrocortisone, 50ug/d fludrocortisone and 1g/d ClNa.

Transaminases normalized, while gamma-GT remained > 1000. Mild hyperthyroidism (T3L : 7,5 pmol/l) (treated with 5mg/d carbimazole), failure to thrive, large-café-au-lait spots appeared during the 3rd month of life (Figure 6).

A mosaic activating GNAS gene mutation was found on DNA extracted from blood and the adrenals.

Conclusion :

Although the interpretation of mosaic multi-organ involvement is difficult in a sick baby, current disease evolution supports the efficacy of propranolol to control diverse aspects of Gs-alpha hyperactivity.

