Early and sever manifestation of McCune-Albright syndrome with GNAS mutation in the liver tissue

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BACKGROUND

McCune-albright syndrome is classically defined by the clinical triad of fibroud dysplasia of bone(FD), café-au-lait spots and precocious puberty. It is a rare disease with variable presentation caused by somatic gain of function mutation in GNAS gene. It can affects both endocrine and non-endocrine tissue. In addition to precocious puberty, other hyperfunctioning endocrinopathies may be involved including hyperthyroidism, growth excess, cushing syndrome and renal phosphate wasting. Skin and skeletal manifestation is the most non-endocrine pathology, but other systems like gastrointestinal and hepatobiliary systems has been also reported.

OBJECTIVES

To report a case with early and sever manifestation of McCune Albright syndrome where diagnosis was not a straightforward.

CASE DESCRIPTION

We report a 2 years old girl who was presented to medical attention at the age of 16 days with cholestatic jaundice and multiple hyperpigmented skin lesions involving right sided of the face, back, buttocks and thigh. She was found to have hyperthyroidism caused by hyperfunctioning nodule in the the left lobe of the thyroid gland (Figure 1), it was uncontrolled with medical treatment. At 4 months of age, she developed vaginal bleeding with high estradiol level and found to have large left ovarian cyst (Figure2). It was recurrent and difficult to control. At 6 months of age, she developed lesions involving right lobe of the liver (Figure3), these lesions were progressively worsening and required right lobectomy and lesion was confirmed histopathologically to be hepatic adenoma. She had multiple skeletal fracture involving right femur and clavicle which was a results of diffuse polyostotic fibroud dysplasia. Lastely, she shows an evidence of growth acceleration with advanced bone age which was a result of growth hormone excess (Table 1) with normal pituitary MRI

TABLE 1: GROWTH HORMONE SUPPRESSION TEST

Time	GLUCOSE LEVEL MMOI/L	GH LEVEL ug/L
0	3.8	4.45
30 min	12.6	>80
60 min	11.6	>80
90 min	9.2	49.20
120 min	7.2	36.50

RESULTS

GNAs mutation was negative in both peripheral blood and skin tissue samples. Heterozygous GNAS mutation with a change from Arginine (CGT) at codon 201 to Histidine (CAT) was identified in liver tissue sample.

CONCLUSION

To our knowledge, such a sever neonatal form of McCune Albright syndrome is rarely reported in the literature. GNAS mutation should be evaluated in the tissue affected if the blood and skin tests are negative.

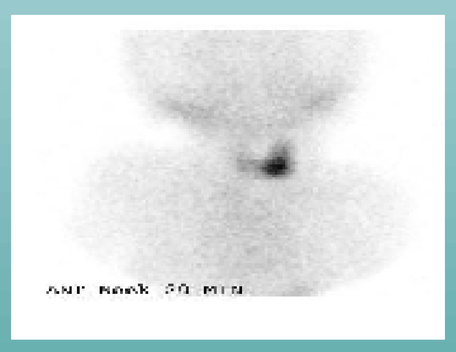

Figure 1 :Thyroid Technetium scan showed increased uptake in the left thyroid lob

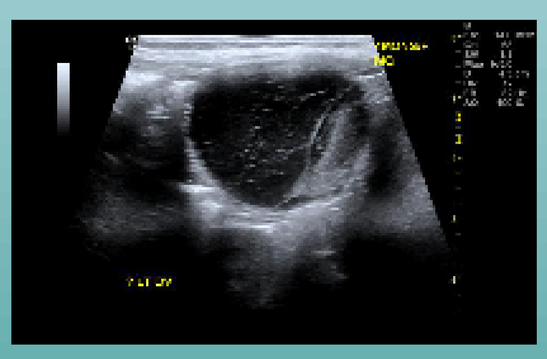

Figure 2: Pelvic ultrasound showed large (4 x 2.2 cm) mlltiseptated cystic structures almost replacing left ovary

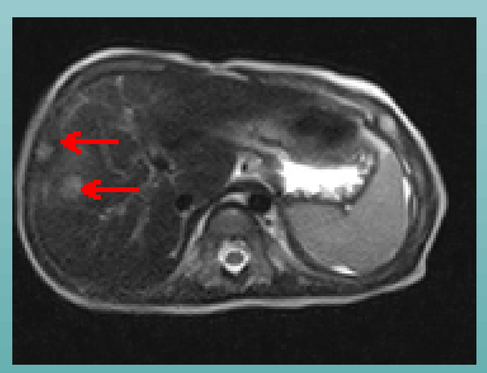

Figure3 : abdominal MRI showed 2 lesion at segment 5 & 8 demonstration high signal intensity at T1

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