NEAR ADULT HEIGHT ACCORDING TO GENETIC TARGET AND ABSENCE OF CRANIOFACIAL BONE FIBROUS DYSPLASIA IN A GIRL WITH MCCUNE ALBRIGHT SYNDROME AND GROWTH HORMONE EXCESS: 12.6 YEARS FOLLOW-UP.

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BACKGROUND:

McCune Albright Syndrome (MAS) is a rare genetic disease clinically defined by bone fibrous dysplasia (BFD), café-au-lait skin spots and at least one hyperfunction endocrinopathy. Growth Hormone (GH) excess has been described in 20% of patients usually accompanied by hyperprolactinemia (80%). As reported in literature, GH hypersecretion is always associated with craniofacial bone fibrous dysplasia (BFD) and is also accompanied by systemic morbidity. Medical therapy is first line treatment and includes somatostatin analogs, GH receptor antagonist, and dopamine agonists. Treatment goal is to maintain IGF1 Z-score between -2 +1SDS and growth velocity according to Tanner stage, sex and age.

AIM:

Long term outcomes on somatostatin analogue and cabergoline treatment for GH excess in a girl with MAS.

CLINICAL CASE:

FIRST VISIT:

- A 15 year-old girl, without personal and family relevant history, was sent for endocrinological evaluation at 2.72 years, because of tall stature (height +2.57 SDS), high growth velocity (12cm/y) and advanced bone age (4y) (Fig. 1).
- Physical exam showed no pubertal signs and a large café- au- lait skin spot characteristic of MAS (Fig 2).
- GH excess was diagnosed by a paradoxical response on OGTT (GH 13.4ng/ml) and high IGF1 +3.32 SDS (Table 1).
- No other endocrinopathies were detected (Table 2)
- Pituitary MRI was normal.
- Octreotide LAR was started at 0.15mg/kg every 28 days.

Fig.1: Growth Chart at Diagnosis

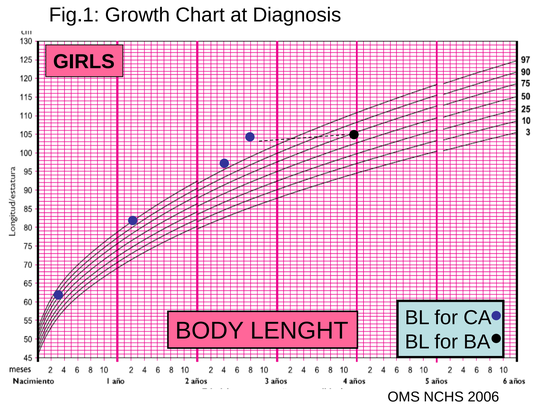


Fig.2 Café-au-lait skin spot

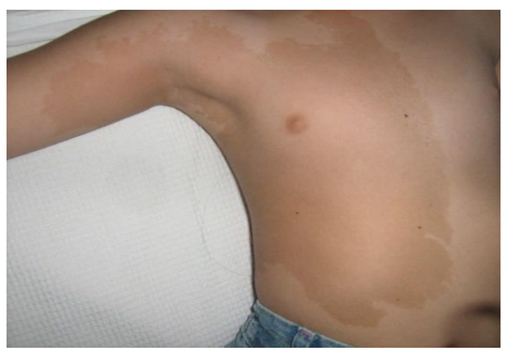


Table 1: OGTT

OGTT	Basal	30´	60´	90´	120´
Glucose mg/dl	81	122	122	119	110
GH ng/ml	1.7	0.95	13.4	3.4	0.99
Insuline µIU/ml	4.2	11.7	100	21.8	20.5

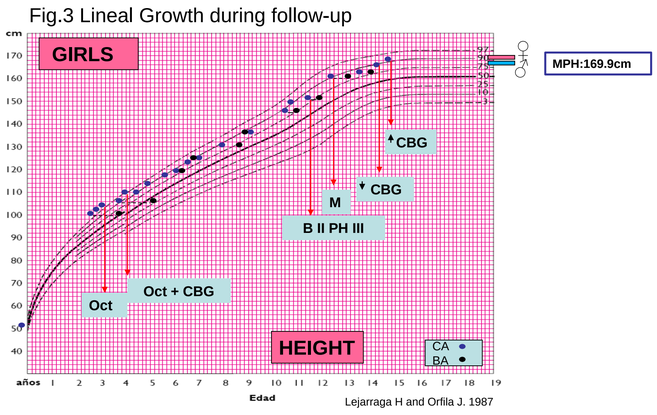
Table 2: Hormonal Data at Diagnosis

	Basal	Reference values
GH (ng/ml)	0.43	
IGF1 (ng/ml) SDS	480 (+2.5)	23 - 56
BP3 (mg/dl)	4.7	1- 4.3
SDHEA (ng/ml)	81	10-200
LH (mIU/ml)	<0.1	0.23 ± 0.88
FSH (mIU/ml)	3.4	2.76 ±1.9
E2 (pg/ml)	<12	<12
PRL (ng/ml)	12.3	< 12
TSH (µIU/ml)	0.85	0.97 - 4.38
fT4L (µg/dl)	1.37	0.93 - 1.94

FOLLOW-UP:

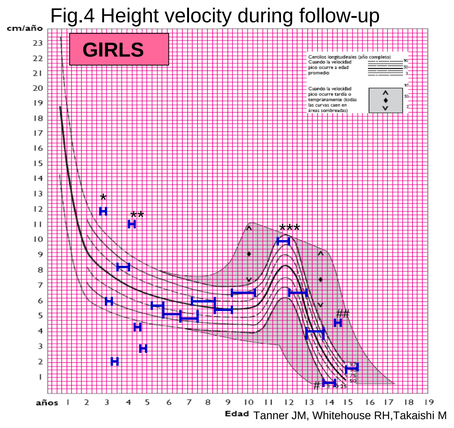
- After 1 year follow-up cabergoline was added at 1mg/week due to the lack of full response on auxological (growth velocity 9cm/y,) and analytical parameters (IGF1 Z-score +1.25 SDS; GH 5ng/ml; PRL 39pg/ml) (Fig. 3 and Fig. 4)
- Under combined treatment all parameters normalized, and remained according to sex, age and Tanner stage (Fig. 5)
- Menarche was presented at normal age (12.56 y) and reached near adult height (169.3cm) according to genetic target (169.9cm) (Fig. 3)
- Once critical growth period was over, and girl reached near adult height, Cabergoline dose was descended in an attempt to evaluate response (GH 0.28ng/ml, IGF1 256ng/ml (-1.23 SDS), PRL <3ng/ml). But due to analitical (IGF1 449.9ng/ml (0.07 SDS), GH 2.96ng/ml and PRL <3ng/ml) and auxological parameters (heigh growth velocity), treatment was restored (Fig.3, Fig.4 and Fig 5).
- During 12.6 years of follow-up, no other endocrinopathies were diagnosed, Pituitary MRI remained normal and no adverse events appeared.
- Craniofacial BFD and long BFD were not found neither in current Head CT nor in 99mTC MDP Bone Scintigraphy. (Fig.6 and 7).

Fig.3 Lineal Growth during follow-up



CA: cronological age; **BA**: bone age; **OCT:** Octeotride; **CBG:** Cabergoline; **B**: Breast Tanner Stage II; **PH**: Pubic Hair Tanner Stage III; **M**: menarche; ♀ Mother Height; ♂ Father Height.

Fig.4 Height velocity during follow-up



* Starts on Octeotride
** Cabergoline was added to treatment
*** Pubertal Spurt (PS)
# Decrease Cabergoline dose
Increase Cabergoline dose

Fig.5: IGF1 SDS during follow-up

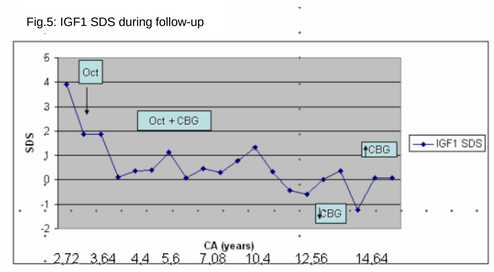


Fig. 5 IGF1 values during follow-up

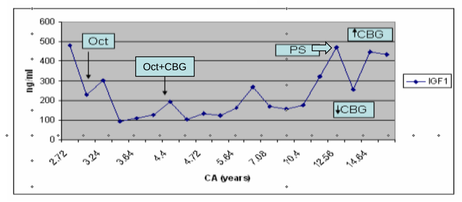


Fig. 6: Current Head CT.

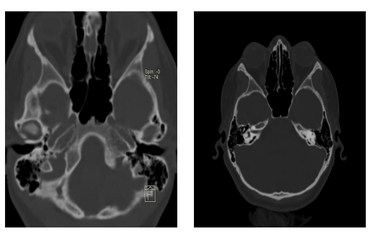
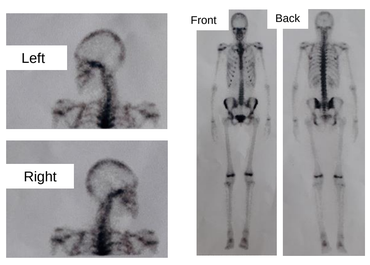


Fig. 7: Current 99mTC MDP Bone Scintigraphy



CONCLUSIONS:

- To our knowledge this is the youngest patient treated with somatostatin analogue and cabergoline. Our data suggest that early treatment with strict compliance could prevent craniofacial complications.
- This clinical case emphasize the effectiveness and safety of an early combined treatment in a girl with MAS and GH excess.
- Further studies with greater number of patients are needed to confirm these conclusions.

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