

A boy with Combined Pituitary Hormone Deficiency and Agenesis of Right Internal Carotid Artery: a rare association or a simple coincidence?

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Background

Congenital Combined Pituitary Hormone Deficiency (CPHD) may be associated with pituitary/extra pituitary abnormalities. Well-known causes are mutations in pituitary transcription factor genes. Agenesis of internal carotid artery (ICA) is a rare vascular anomaly that has been associated with CPHD.

Case presentation

The patient is a 2-yr old male, born at term to non-consanguineous parents, birth wt 3,2 kg, length 47 cm, no gestational or perinatal complications. Shortly after birth he developed symptomatic episodes of hypoglycemia with capillary glucose between 33 to 39 mg/dL. Insulin was not measured. He was treated with IV glucose infusion (16mg/kg/min) until oral intake increased and IVF could be discontinued. He was discharged on the 6th day with q3h-breastfeeding.

On follow-up, he showed poor growth, dropping below the 3rd percentile for wt and length, and micropenis and cryptorchidism were noticed. Hormonal work up at 5 months revealed glucose 69 mg/dL, IGF-1<15 ng/mL (n.v.27-114), IGFBP3: 583 ng/mL (n.v.600-2900), LH 0.4mIU/mL (n.v.3-22), testosterone 9 ng/dL (n.v.72-340), FT4 0.7ng/ml (n.v.0.6-1.2), TSH 4.73 mIU/L (n.v.0.8-6.), cortisol 21,4 ug/dl. The diagnosis was CPHD (GH + gonadotrophin deficiency).

A cranial/sellar MRI showed an empty sella, interrupted pituitary stalk and absence of the right ICA with a complex arterial anastomosis supplying the middle cerebral artery on the right side (**Figure 1**).

At 9 months the patient was started on rhGH and growth velocity doubled. He reached the 10th pc after 14 months of treatment (**Figure 2**).

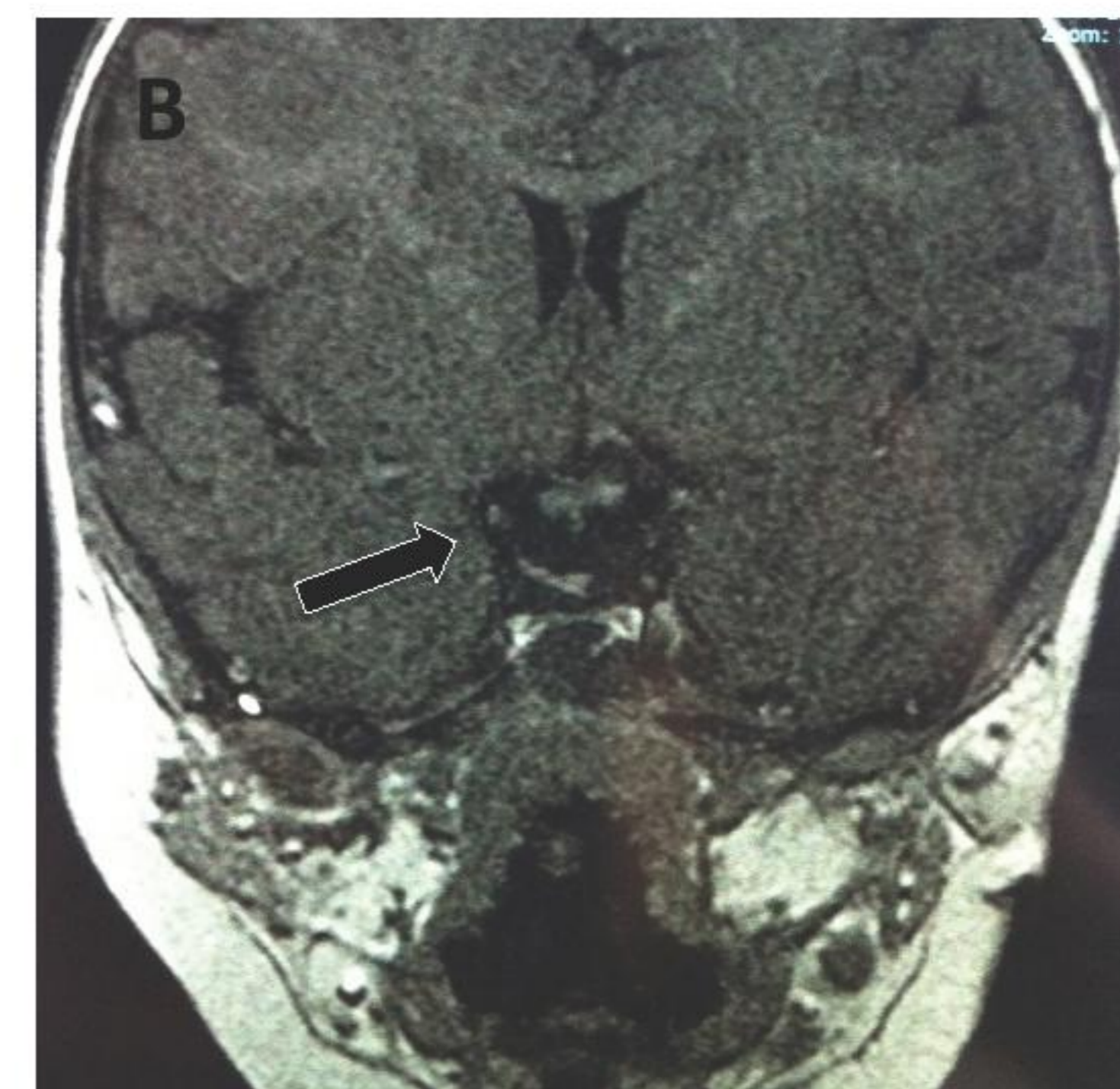
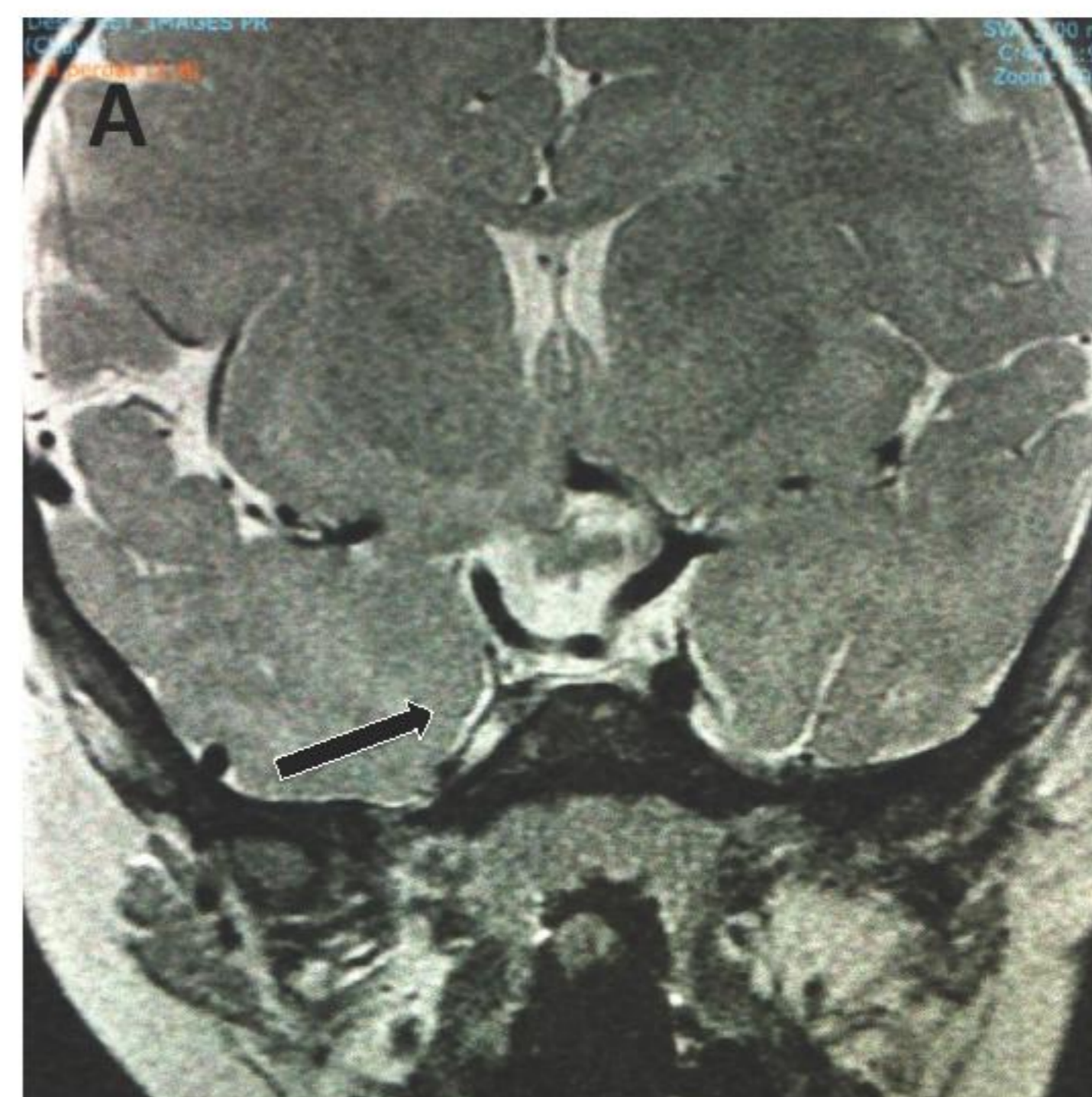


Figure 1: A) T2 weighted coronal view: absence of right carotid artery
B) T1 weighted coronal view: anterior pituitary hypoplasia and interrupted stalk

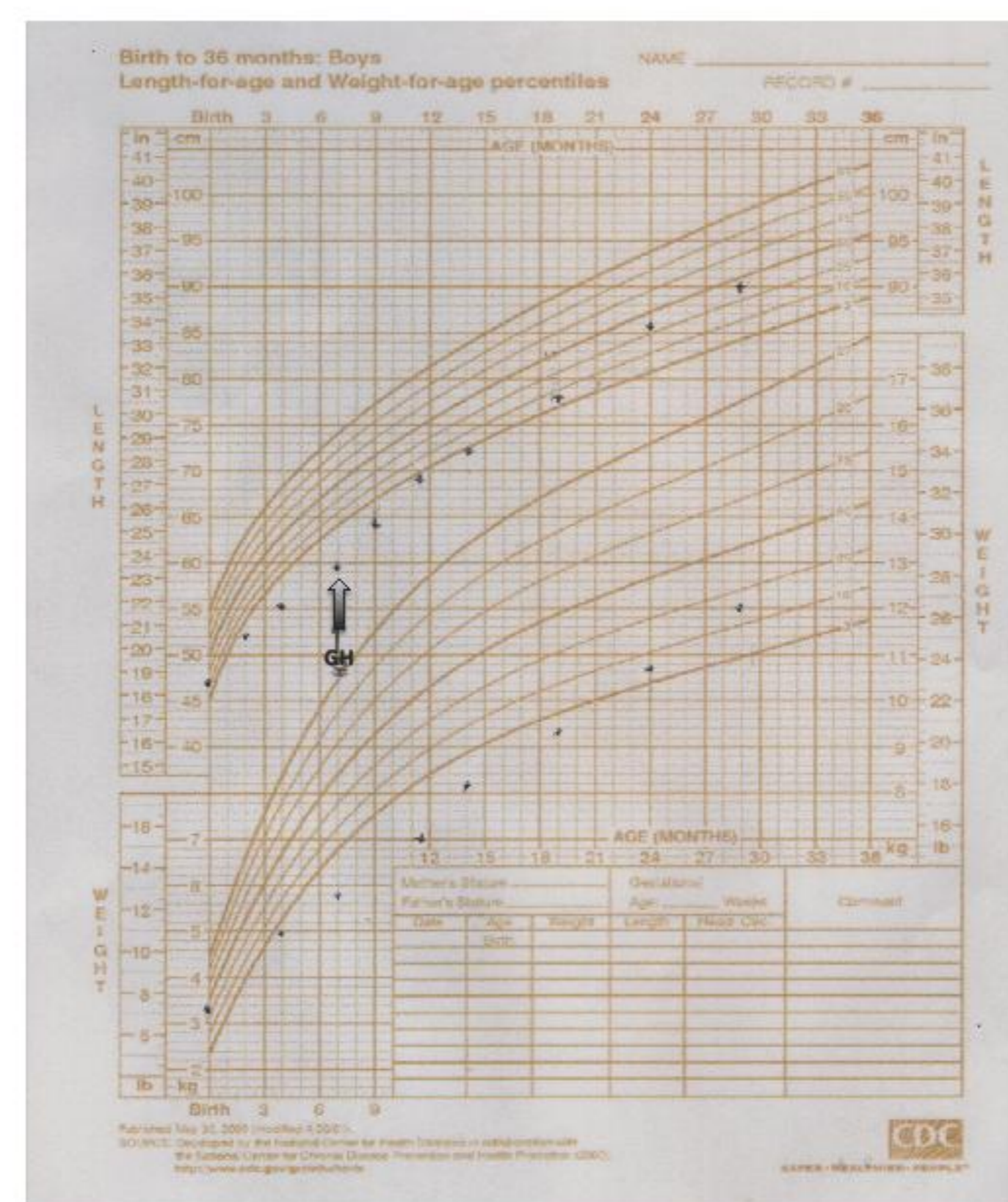


Figure 2: Growth curve

Conclusion

CPHD and ICA are rare disorders individually and even rarer together. Underlying mechanisms for this combination are unclear and may involve a single molecular abnormality that could primarily cause ICA and secondarily affect the blood supply of the developing pituitary, or affect both vascular and pituitary development simultaneously.

This eleventh case of CPHD+ICA here described, together with the other ten already described, may stimulate the molecular research for the cause of this new clinical entity.

References

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