

AMSER Case of the Month

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14-year-old male presents with concerns of short stature

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Patient Presentation

- 14-year-old male presented with parents to pediatric endocrinologist for concerns of delayed puberty with short stature
- Negative ROS
- Height: 2nd percentile; weight: 35th percentile; BMI: 82nd percentile; Tanner Stage 1 pubic hair

Pertinent Labs / Workup

Lab	Patient Value	Reference Range (Tanner I)
LH	0.7 IU/L	0.02–0.3 IU/L
FSH	5.2 IU/L	0.3–4.6 IU/L
Testosterone	13 ng/dL	<20 ng/dL

- Negative celiac and thyroid antibody screening
- GH stimulation testing showed inadequate response:
 - *Clonidine stimulation test: GH peak level ≤ 2.9 (nml cutoff $\sim 3 - 7$ ng/mL)*
 - *Insulin tolerance test: GH levels remained ≤ 0.1 (nml cutoff ~ 5 ng/mL)*
- Radiographic bone age: 12 years 6 months *versus us* chronologic age: 14 years 3 months

What Imaging Should We Order?

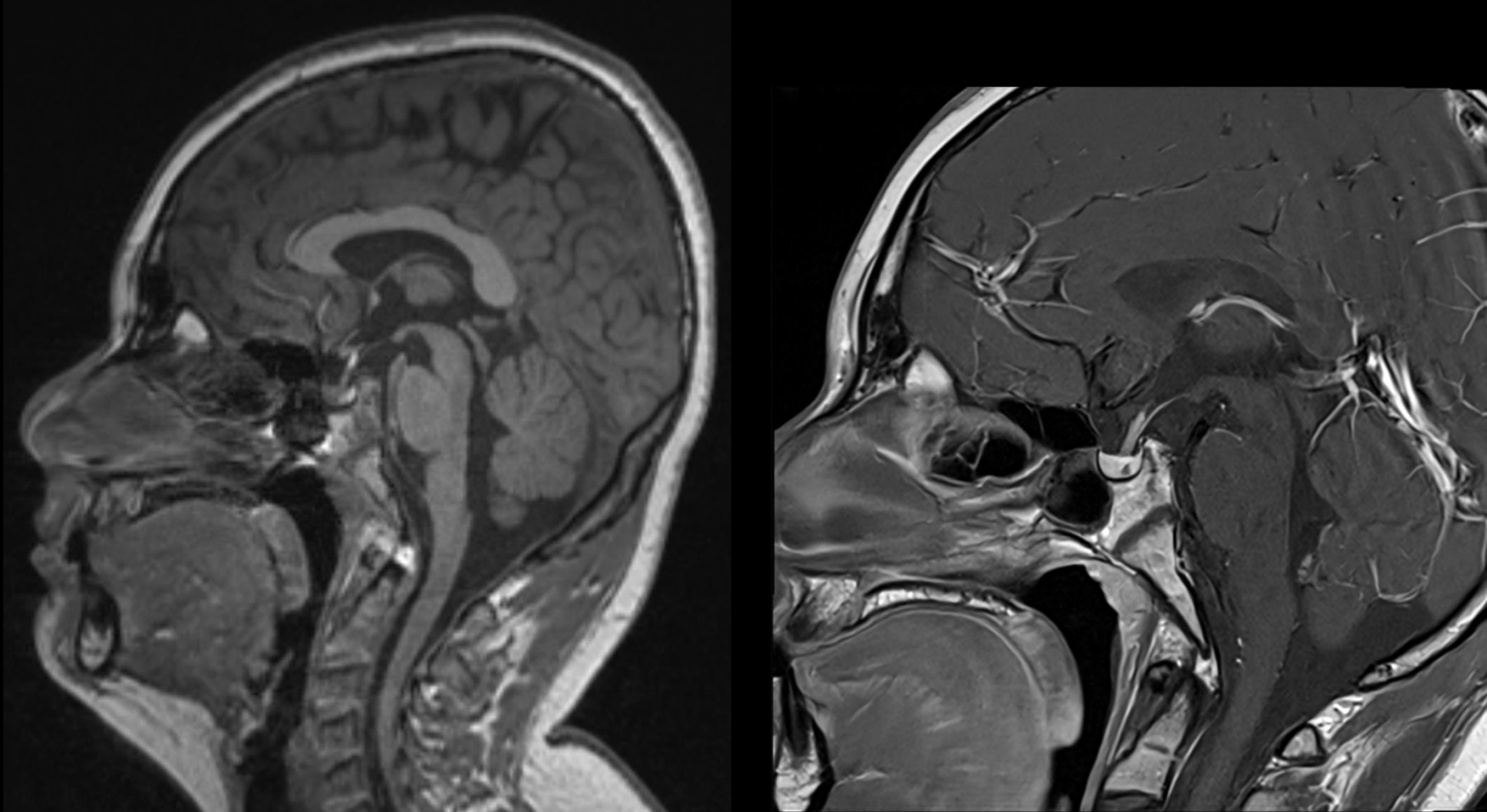
Select the applicable ACR Appropriateness Criteria

Variant: 1 Adult. Suspected or known hypofunctioning pituitary gland (hypopituitarism, growth hormone deficiency, growth deceleration, panhypopituitarism, hypogonadotropic hypogonadism). Initial imaging.

Procedure	Appropriateness Category	Relative Radiation Level
MRI sella without and with IV contrast	Usually Appropriate	○
MRI sella without IV contrast	Usually Appropriate	○
MRI sella with IV contrast	May Be Appropriate	○
CT sella with IV contrast	May Be Appropriate	☢☢☢
Radiography sella	Usually Not Appropriate	☢
Venous sampling petrosal sinus	Usually Not Appropriate	Varies
MRA head with IV contrast	Usually Not Appropriate	○
MRA head without and with IV contrast	Usually Not Appropriate	○
MRA head without IV contrast	Usually Not Appropriate	○
CT sella without and with IV contrast	Usually Not Appropriate	☢☢☢
CT sella without IV contrast	Usually Not Appropriate	☢☢☢
CTA head with IV contrast	Usually Not Appropriate	☢☢☢

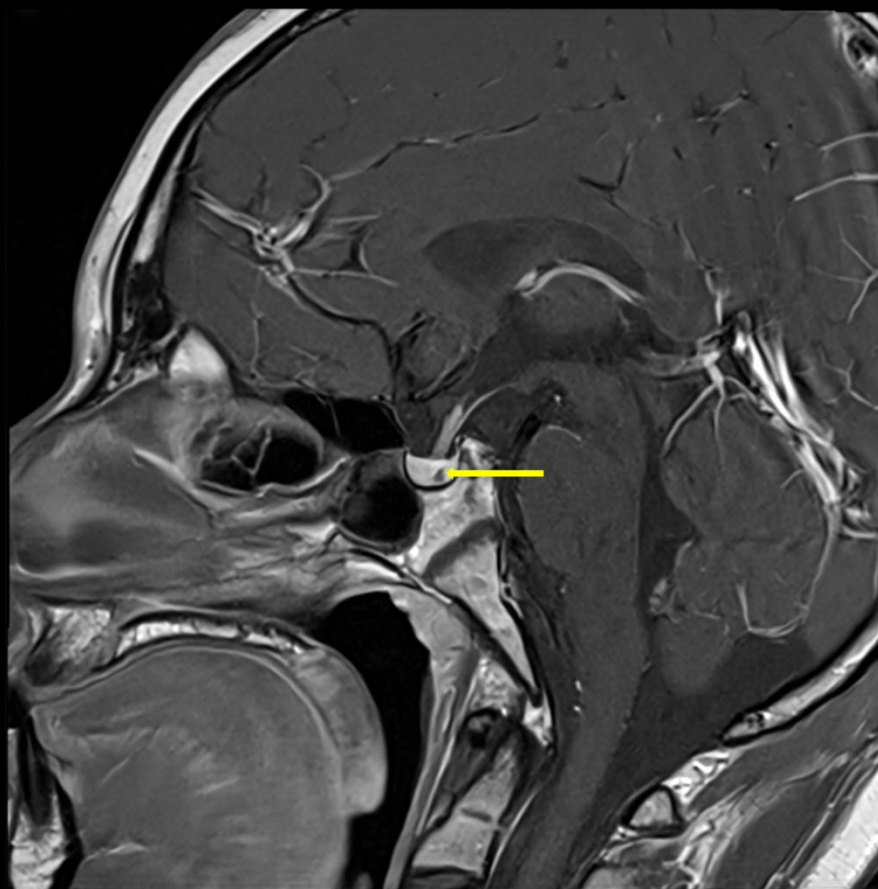
This imaging modality was ordered by the endocrinologist

Findings: (unlabeled)



T1 MPRAGE (left) and T1 post-contrast (right) sagittal MRI sequences of the brain

Findings: (labeled)



- Normal posterior pituitary T1-bright spot (*white*) is present in the posterior sella

- Ectopic T1-bright spot (*red*) is present along the infundibular stalk / median eminence

- Incidental finding of hypoenhancing cystic lesion (*yellow*) in pars intermedia of the anterior pituitary; seen on T1 post-contrast sequence

T1 MPRAGE (left) and T1 post-contrast (right) sagittal MRI sequences of the brain

Final Dx:

Partial Ectopic Posterior Pituitary

with a small pars intermedia cystic lesion likely representing a Rathke cleft cyst

Case Discussion: Pituitary Development

- The pituitary has a dual embryologic origins:
 - Adenohypophysis (anterior pituitary): oral ectoderm rises as the Rathke pouch and eventually forms the pars distalis, pars tuberalis, and pars intermedia of the anterior pituitary. ^[3]
 - Neurohypophysis (posterior pituitary): ventral diencephalon descends as the infundibulum and eventually forms the posterior pituitary tissue. ^[3]
- **Rathke cleft cysts** arise from persistent Rathke pouch remnants near the pars intermedia.
 - Small cysts may be incidental when there is no optic chiasm compression or pituitary stalk deviation. ^[4,9]
- **Partial ectopic posterior pituitary (PEPP)** arises from incomplete neurohypophyseal migration
 - Some posterior pituitary tissue still reaches the posterior sella as expected, while some posterior pituitary tissue remains arrested partway along the infundibular stalk. This is an extremely rare occurrence with only a handful of cases reported worldwide. ^[1,2]
- This patient was found to have developmental abnormalities along both embryological lines.

Case Discussion: Partial Ectopic Posterior Pituitary

- Normal posterior pituitary tissue is intrinsically bright on precontrast T1 MRI. ^[5]
- PEPP manifests two bright spots on pre-contrast T1 MRI:
 - One normal bright spot in the posterior sella
 - An ectopic bright spot along the infundibular stalk or median eminence region. ^[1,2]
- This is much rarer than classic pituitary stalk interruption syndrome (PSIS). ^[1,6]
 - PSIS involves fully ectopic or absent posterior pituitary tissue as well as a thin or absent stalk. It is often associated with anterior pituitary hypoplasia. ^[6]
- Ectopic posterior pituitary tissue is often correlated with anterior pituitary hormone deficiencies. ^[6,7]
 - An intact stalk as seen in PEPP may have milder presentation than PSIS but can still be associated with growth-hormone deficiency. ^[2]

Case Discussion: Management and Follow-Up

- Growth hormone deficiency is diagnosed clinically and biochemically; MRI helps to exclude mass lesion and identify potential structural abnormalities. ^[8]
 - This patient had failed provocative GH stimulation testing and delayed skeletal maturation.
- In this patient's case, MRI showed no sellar mass, optic chiasm compression, cavernous sinus invasion, or stalk deviation.
 - The small pars intermedia cyst can be observed if asymptomatic and noncompressive. ^[9]
- Management will include recombinant growth-hormone therapy and monitoring of his IGF-1 levels and growth velocity ^[7,8] with continued endocrinology follow-up.

References:

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