PITUITARY DYSFUNCTION WITH ASSOCIATED LESIONS IN THE HYPOHYALAMO-PITUITARY REGION: HISTIOCYTOSIS OR DYSGERMINOMA?

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Introduction-Aim

Patients who present with clinical and laboratory findings of pituitary dysfunction and whose MRI findings reveal increased pituitary size or thickening of pituitary stalk, pose a diagnostic challenge. Differential diagnosis mainly includes dysgerminoma, histiocytosis and hyphopitidis. A non-invasive approach is often non-diagnostic. The aim is to present two patients with similar clinical picture and positive MRI findings in whom transphenoidal biopsy of the pituitary lesion, led to prompt accurate diagnosis and further therapeutic management.

Patient-1

Prepubertal girl 8 years of age, presented with the main complaint of slow growth rate.

PMH: Polyuria, Polydipsia, for three years, growth rate < 3 cm/yr

Physical exam: HT: 132 cm (<3rd Mile), Wt: 25 Kg (25th %ile), HC: 52 cm

She had no dysmorphic features. Thyroid gland was not palpable, Breasts, Axillary, Pubic Hair: Tanner I.

Laboratory investigations: CBC: NL, UA: S.g. 2005; BUN: 33 Cr: 0.5 Na: 145 K: 3.9 Ca: 9.8 P: 3.9 AP: 180 T4: 85 µg/dl PRL: 82 IGF-1: 34 ng/ml BA: 7 yrs. PEAK GH: 23 ng/ml Peak cortisol: 8.5 µg/dl At time point 0 cortisol: 6.5 µg/dl ACTH: 14 Water deprivation test: 12 h plasma Osm: 297 mOsm Urine Osm: 89 mOsm

Post Minirin Plasma Osm: 307 Urine Osm: 260 Skeletal survey, chest x-ray, abdominal U/S and hematologic indices for histiocytosis were negative. Serum and CSF levels of HCG and α-FP were negative. Cytology of CNS was also negative.

Diabetes insipidus, growth hormone deficiency and cortisol insuficiency.

MRI

Transphenoidal biopsy with excision of the lesion revealed dysgerminoma.

The patient responded to two cycles of Carboplatin/Etoposide. She also received limited field RT (COG AN50232 Protocol).

Patient-2

15-5 yrs old adolescent girl, presented for investigation of secondary amenorrhea and an episode of fainting.

PMH: Diabetes insipidus, treated with Minirin

Physical exam: HT: 171 cm (75th %ile), Wt: 67.5 Kg (75-90 %ile)

She had no dysmorphic features. Thyroid gland was palpable Breasts, Axillary, Pubic Hair: Tanner V.

Laboratory investigations: CBC: NL, UA: S.g. 2005; BUN: 23 Cr: 0.8 Na: 137 K: 4.0 Plasma osmolality: 290 mOsm Urine osmolality: 496 mOsm T4: 70 µg/dl TSH: 0.103 µIU/ml PRL: 40.4 ng/ml PEAK GH: 1.57 ng/ml E2 ≤ 12 pg/ml, FSH: 2.4 IU/ml, LH 1.1 IU/ml, Prl: 40.4 ng/ml Peak cortisol: 21.2 µg/dl At time o cortisol: 18.35 µg/dl Skeletal survey chest x-ray, abdominal U/S and hematologic indices for histiocytosis were negative. Serum and CSF levels of HCG and α-FP were negative. Cytology of CNS was also negative.

Hypogonadotrophic hypogonadism, central hypothyroidism, diabetes insipidus, growth hormone deficiency hyperprolactinemia nad normal cortisol secretion

Management

Transphenoidal biopsy with excision of the lesion revealed dysgerminoma.

The patient received chemotherapy consisting of Carboplatin/Etoposide. She also received limited field RT (COG AN50232 Protocol).

Conclusions

Tissue biopsy provides the definitive diagnosis and the transphenoidal approach appears to be optimal for certain patients.

Inconclusive imaging appearance, negative markers and slow progression cannot exclude the diagnosis of dysgerminoma.