P3-811
The authors have nothing to disclose

Cushing's syndrome (CS) due to ectopic ACTH secretion by a germline tumor in the cross-tail area in a 7 months old female infant

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Background

Germ cell tumors are derived from primary germ cells-gonocytes which migrate from the yolk-sac along the embryonic axis and retain the possibility of multi-directional differentiation, hence the large variety of growth and potential location throughout the body. Ectopic ACTH syndrome is very rarely seen in infancy, usually occurring in older children.

Case presentation

We present a rare case of Cushing's syndrome in an infant with a tumor in the cross-tail area. A female infant was born by Caesarean section (BW 4280 g) with congenital anal atresia and a large tumor surrounding the cross-tail region. CT image identified an abnormal heterogeneous pelvic mass (dimensions 76mm x 49mm x 38mm) below the sacrum. On day 1 of life, a sigmoid colostomy was established and at age 1 week, part of the tumor with the coccyx was removed. Control CT abdomen and pelvis scan showed residual tumor (27 x 21 x 28 mm). Histopathology showed a grade 3 teratoma immaturum. **Alpha-fetoprotein (AFP) pre-surgery was 59,000 ng/ml and post-surgery 6,339ng/ml (normal range 500ng/ml).** There were no metastases.

Results

For 3 months, the child remained in good condition (AFP normalization, stabilization of the tumor), then there was an increase of AFP, beta-HCG levels, and an increase in tumour size on imaging. Chemotherapy - 3 blocks VBP (vinblastine, bleomycin, cisplatin) normalized AFP and decreased tumor size. At age 7 months the child had increased appetite, weight gain (>97thc), Cushingoid appearance, hypertension (BP 210/160 mm Hg), hypokalemia (2.85 mmol/l), hypercortisolemia (09,00h 1794 nmol/, 13.00h; 1794 nmol/l), increased ACTH (121 pg/ml) and LDH (1,005 U/L).

Dexamethasone suppression test showed absent cortisol suppression: 1,054 nmol/l (basal), 1,056 nmol/l (post-dex). Imaging studies (CT CNS, chest, adrenal scintigraphy with octreotide) excluded metastases. Immunohistochemical staining of the tumor was positive for ACTH in cancer cells. Subsequent chemotherapy: VIP (etoposide, ifosfamide, cisplatin) did not contribute to the regression of the tumor mass or normalization of biochemical and hormonal parameters. Ketoconazole, metyrapone, anti-hypertensive (metocard, captopril, ebrantil, aldactone) therapy induced only temporary, control of hypercortisolism (09.00h cortisol 1453 nmol/l, ACTH 700 pg/ml) and BP.

The child was operated twice in Department of Surgery Memorial Health Institute in Warsaw, Poland at age 12 and 14 months. A significant part of the tumor was removed at surgery. Currently, she requires small doses of metyrapone due to fluctuation of ACTH and cortisol concentrations.

Germ cell tumors

* Benign and malignant tumors are rare and comprise 3% of all cancers in childhood.

- 3 peaks of morbidity:
- up to 2 years of age -tumors in cross-tail area,
- after 6 years of age- ovarian tumors,
- after 14 years of age- testicular tumors.

Location	Frequency (%)
Cross-tail area	19
OVARIES	30
TESTES	17
CNS	20
mediastinum	4
retroperitoneal area	3
OTHER	5

GROUP 1	GROUP 2	GROUP 3
Gonadoblastoma	Yolk sac tumor	Ca embryonale
Germinoma - dysgerminoma -seminoma	Polyembrioma	Teratoma immaturum
	Chorioncarcinoma	Teratoma adultum

THE PATIENT AT AGE 6 MONTHS

THE PATIENT AT AGE 7 MONTHS

THE PATIENT AT AGE 8 MONTHS AFTER 3 WEEKS OF KETOCONAZOLE THERAPY

THE PATIENT AT AGE 17 MONTHS AFTER 2 OPERATIONS

Conclusion

An extremely rare cause of CS due to ectopic ACTH syndrome is described in a female infant.

ESPE 2015 811-P3

Misc 1

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DOI: 10.3252/pso.eu.54espe.2015

Poster presented at: 54th Annual Meeting 1-3 October ESPE 2015 BARCELONA European Society for Paediatric Endocrinology

PosterSessionOnline.com