Centre de Référence des Maladies Endocriniennes Rares de la Croissance

Institut E3M Endocrinologie Maladies Métaboliques et Médecine interne

ENDOCRINE MANIFESTATIONS IN ERDHEIM CHESTER DISEASE

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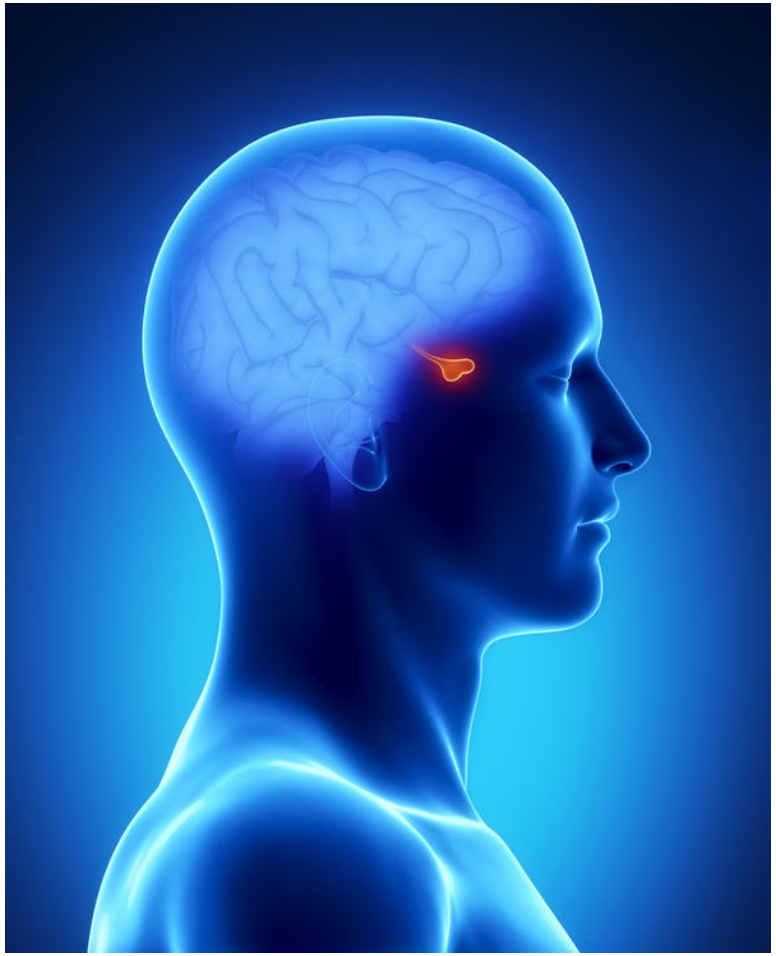
Annual International ECD Patient and Family Gathering

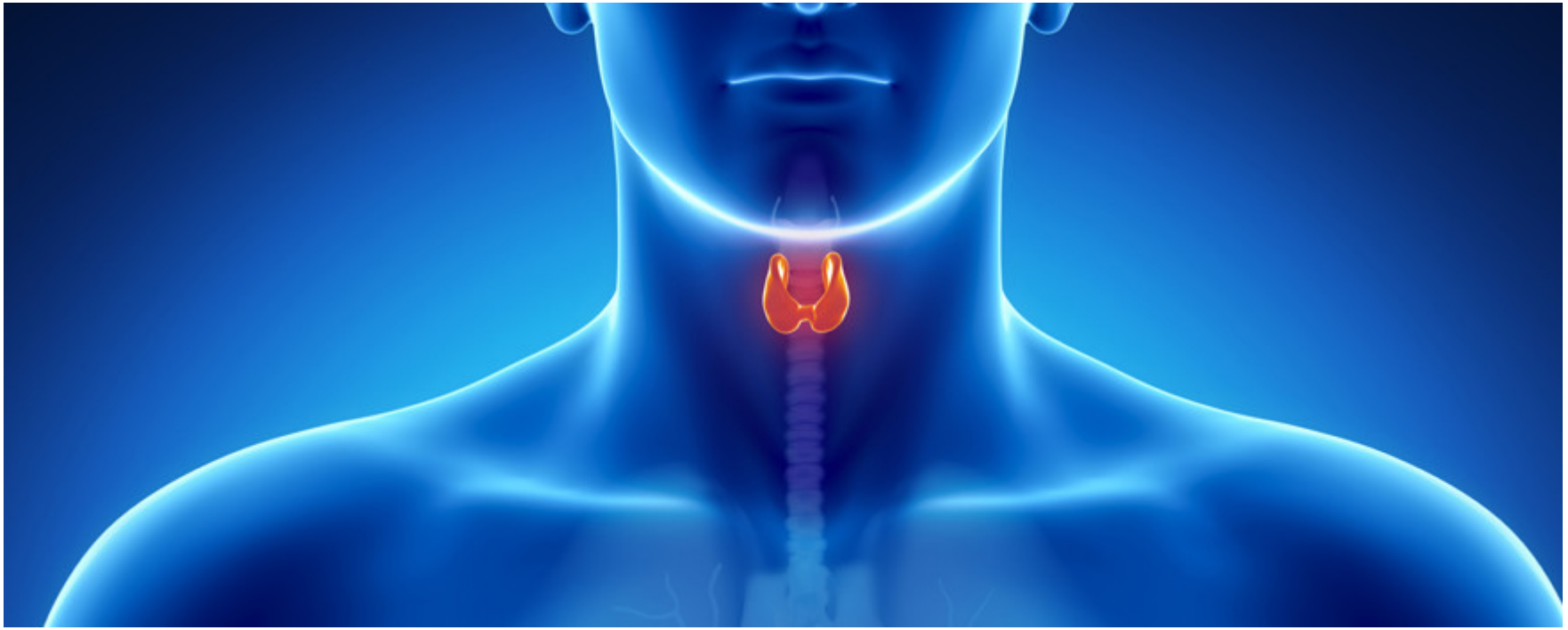
September 16th 2016

Pituitary gland



- The **anterior pituitary** regulates other glands in the body and controls most of the hormonal secretions:
  - **Thyroid**
  - **Adrenals**
  - **Ovaries, testicles**
  - **Breasts**
  - **Growth**
- The **posterior pituitary** produces the **anti-diuretic hormone**
  - A lack in this hormone induces enhanced diuresis which in turn drives to an important thirst and a need to drink in order to maintain normal hydration
  - This deficit is called **diabetes insipidus**

Thyroid gland



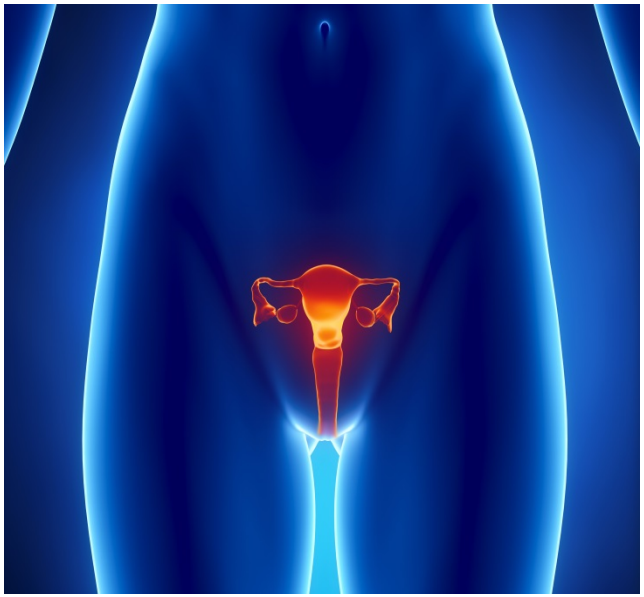
- The **thyroid gland** secretes hormones that regulate many **metabolic processes**, including **growth** and **energy expenditure**

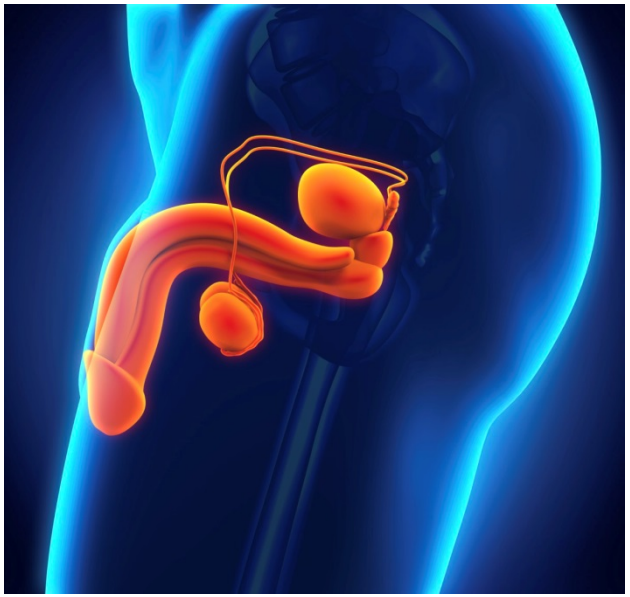
Adrenal glands

- **Adrenal glands produce several major hormones** important for
  - coping with **physical stresses** to body
  - maintaining adequate **blood pressure** control, **blood volume** and **salt retention** by the body

- These hormones are
  - **Cortisol** and **aldosterone** which regulate blood pressure, salt retention and general well being
  - Mildly potent **male hormones**
  - **Adrenaline** and **noradrenaline**

Gonads = ovaries / testicles





- **Gonads** have 2 functions
  - Secretion of the **sexual hormones:** estradiol and progesterone in women, testosterone in men
  - Production of **gametes** to ensure **reproduction: oocytes** in women and **spermatozoids** in men

ECD and endocrine manifestations

- All the glands can be infiltrated by the histiocytosis
  - Pituitary (24%)
  - Testicles (29%)
  - Adrenals (39%)
  - Thyroid
  - Breast

- Hormonal dysfunctions are **very frequent** and have **important implications**
  - Diabetes insipidus
  - Fatigue, headrush
  - Excess body weight
  - Low muscular strength
  - Impotency, infertility
  - Depression, mood changes
  - Increase in cardiovascular risk
  - Bone demineralization

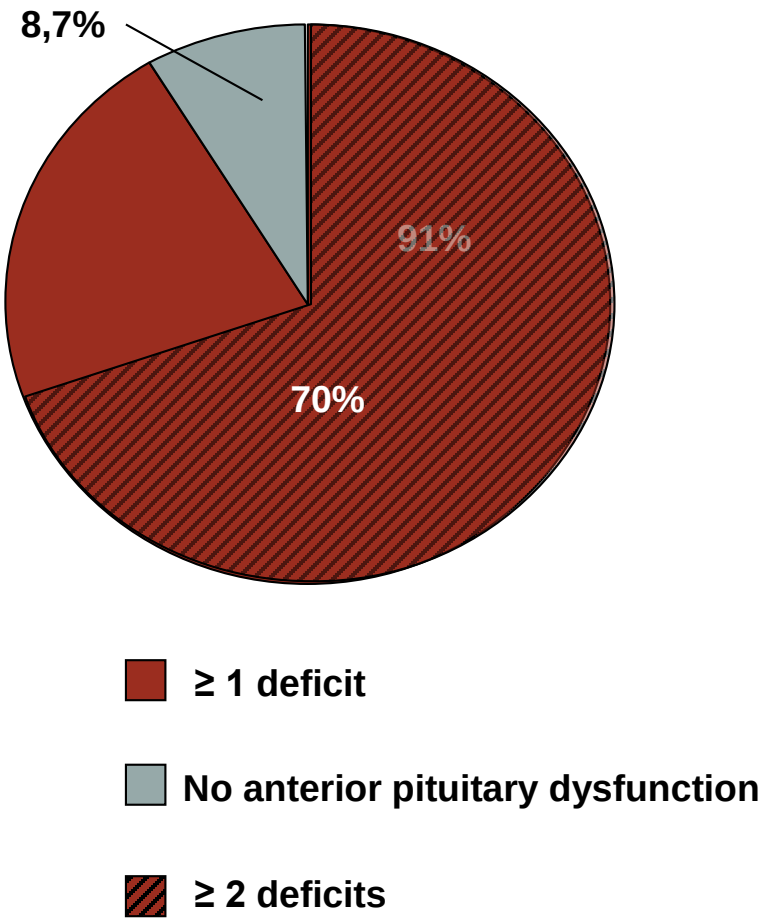
Endocrine manifestations

Hormonal dysfunction	% of patients
Growth hormone deficiency	79%
Testicular deficiency	53%
Diabetes insipidus	33%
Gonadal function deficiency	22%
Thyroid deficiency	20%
Cortisol deficiency	4%
NONE	1%

- **Diabetes insipidus**
  - is often one of the first signs of ECD and the first endocrine manifestation
  - is permanent
- **New deficits** can appear during follow-up
- Men can have **alteration in sperm count**, so if there might be a parental project, **conservation of sperm** as soon as possible is mandatory

Results

Anterior pituitary deficits



Recommendations

	CLINICAL EVALUATION	MORPHOLOGICAL EVALUATION	HORMONAL EVALUATION
PITUITARY	**Search for signs of anterior pituitary deficits** **24hours diuresis and water intake**	**Pituitary MRI**	**Evaluation of anterior and posterior pituitary functions**
GONADS	Evaluation of **testicular volume** and search of palpable **testicular nodules**	**Gonadal sonography** If man with testicular infiltration > **sperm cryopreservation**	**Evaluation of gonadal function (ovaries, testicles)**
THYROID	Search of a **goitre** and of **nodules**	**Thyroid sonography** if clinical anomalies	**Evaluation of thyroid function**
ADRENAL	Search of signs of **adrenal deficiency**	**Abdominal or adrenal CT scan**	**Evaluation of adrenal function**
BREAST	Search for **lumps**	**Mammography +/- mammary sonography** if presence of clinical lumps	**-**
METABOLISM	**Blood pressure** **Electrocardiogram**	**-**	**Blood glucose** **Lipid profile**