ENDOCRINE MANIFESTATIONS IN ERDHEIM CHESTER DISEASE

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

<table>
<thead>
<tr>
<th>Hormonal dysfunction</th>
<th>% of patients</th>
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<tbody>
<tr>
<td>Growth hormone deficiency</td>
<td>79%</td>
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<tr>
<td>Testicular deficiency</td>
<td>53%</td>
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<tr>
<td>Diabetes insipidus</td>
<td>33%</td>
</tr>
<tr>
<td>Gonadal function deficiency</td>
<td>22%</td>
</tr>
<tr>
<td>Thyroid deficiency</td>
<td>20%</td>
</tr>
<tr>
<td>Cortisol deficiency</td>
<td>4%</td>
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<tr>
<td>NONE</td>
<td>1%</td>
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</tbody>
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- **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

- ≥ 1 deficit: 91%
- ≥ 2 deficits: 70%
- No anterior pituitary dysfunction: 8.7%
## Recommendations

<table>
<thead>
<tr>
<th>System</th>
<th>Clinical Evaluation</th>
<th>Morphological Evaluation</th>
<th>Hormonal Evaluation</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Pituitary</strong></td>
<td>Search for signs of anterior pituitary deficits</td>
<td>Pituitary MRI</td>
<td>Evaluation of anterior and posterior pituitary functions</td>
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<td></td>
<td>24 hours diuresis and water intake</td>
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<tr>
<td><strong>Gonads</strong></td>
<td>Evaluation of testicular volume and search of palpable testicular nodules</td>
<td>Gonadal sonography If man with testicular infiltration &gt; sperm cryopreservation</td>
<td>Evaluation of gonadal function (ovaries, testicles)</td>
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<tr>
<td><strong>Thyroid</strong></td>
<td>Search of a goitre and of nodules</td>
<td>Thyroid sonography if clinical anomalies</td>
<td>Evaluation of thyroid function</td>
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<tr>
<td><strong>Adrenal</strong></td>
<td>Search of signs of adrenal deficiency</td>
<td>Abdominal or adrenal CT scan</td>
<td>Evaluation of adrenal function</td>
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<tr>
<td><strong>Breast</strong></td>
<td>Search for lumps† †</td>
<td>Mammography +/- mammary sonography if presence of clinical lumps</td>
<td>-</td>
</tr>
<tr>
<td><strong>Metabolism</strong></td>
<td>Blood pressure Electrocardiogram</td>
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<td>Blood glucose Lipid profile</td>
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