Multiple endocrine neoplasia syndrome

MEN = Multiple Endocrine Neoplasia Syndrome - this includes three syndromes, the main components of which are tumors of the endocrine glands.

MEN 1 syndrome (Wermer syndrome)

Definition and epidemiology

An autosomal dominantly inherited syndrome with a prevalence of 2-20 cases per 100,000 population, characterized mainly by primary hyperparathyroidism, tumors of the endocrine part of the pancreas and tumors of the pituitary gland.

Etiopathogenesis

Defects (deletion, mutation) gene of the menin protein. This gene is a tumor suppressor gene. It is located on the 11th chromosome.

Clinical picture

1. **Primary hyperparathyroidism** – occurs within the syndrome with a probability of 95-100% and is also the first manifestation.
2. **Tumors of the endocrine pancreas** – can be a functional or hormonally active - most often gastrinoma (60 %) or insulinoma (20 %).
3. **Pituitary tumors** – almost always **adenomas** . They can be non-functional, in the case of activity it is almost always a prolactinoma.

MEN 1 can include: adenomas of the adrenal glands, carcinoid, subcutaneous lipomas, angiofibromas on the face, collagenomas, ...

Diagnostics

Proof of at least two basic components of the syndrome, proof of diagnosis based on molecular biological examinations.

Therapy

It consists in the therapy of individual components.

Prognosis

Increased morbidity and mortality depend on early diagnosis and treatment of individual components.

MEN 2A syndrome (Sipple syndrome)

Definition and epidemiology

An autosomal dominantly inherited syndrome with a prevalence of 1–5 cases per 100,000 population. The components of the syndrome are: medullary carcinoma of the thyroid gland, pheochromocytoma, primary hyperparathyroidism.

Etiopathogenesis

Mutation of the RET proto-oncogene located on the 10th chromosome.

Clinical picture

1. Medullary carcinoma of the thyroid gland – usually manifests first, is highly malignant.
2. **Primary hyperparathyroidism** – manifests in approximately 25%.
3. Pheochromocytoma.

Differential diagnosis

Differentiation from other tumor syndromes. We will confirm the diagnosis by showing a mutation of the RET proto-oncogene.

Therapy

It consists in the therapy of individual components. Dispensary is important and preventive total thyroidectomy is indicated for offspring.

Prognosis

MEN 2A is a severe syndrome with increased morbidity and mortality. Early diagnosis, therapy of individual components and also which components of the syndrome are manifested are important.

MEN 2B syndrome

Definition and epidemiology

An autosomal dominantly inherited syndrome with a prevalence of 1–3 cases per 100,000 population. Components of the syndrome are: medullary carcinoma of the thyroid gland, pheochromocytoma, mucosal neuromas.

Etiopathogenesis

Mutation of the RET proto-oncogene located on chromosome 10.

Clinical picture

1. **Medullary thyroid carcinoma** – present in 100% of cases.
2. **Pheochromocytoma** – present in 50% of cases, often bilateral.
3. **Mucous neuromas** – in 100% of cases, marfanoid habitus (70%) and intestinal ganglioneuromas.

Diagnostics

Evidence of individual components of the syndrome and evidence of a RET proto-oncogene mutation.

Therapy

To prove the syndrome, preventive total thyroidectomy is indicated even before the manifestation of cancer, also for offspring. The therapy of individual components is also used, dispensary with active search and early diagnosis is important.

Prognosis

MEN 2B is a severe syndrome with increased morbidity and mortality. Early diagnosis, therapy of individual components and also which components of the syndrome are manifested are important.

Links

- ws:MEN syndrom

related articles

- Medullary carcinoma of the thyroid gland
- Pheochromocytoma

Source

- ČEŠKA, Richard – TESAŘ, Vladimír, et al. *Interna.* 1. edition. Praha : Triton, 2010. ISBN 978-80-7387-423-0.