HIPOTIROIDISM LA COPII

☆ congenital : agenezie ty, ectopie ty, mutatii genetice pe enzimele formarii hh ty/ captarii iodului. 1:3500 nn,

Hipotiroidism secundar al nn (afectare hipofizara sau hiootalamica) 1 :60.000-140.000

Approximately 75% of infants with congenital hypothyroidism have defects in thyroid gland development, 10% have hereditary defects in thyroid hormone synthesis or uptake, 5% have secondary (pituitary) or tertiary (hypothalamus) hypothyroidism, and 10% have transient hypothyroidism.

Thyroid dysgenesis: Defective thyroid gland development accounts for most instances of congenital hypothyroidism. Thyroid dysgenesis occurs sporadically in most cases but is occasionally familial because of mutations or deletions of genes (*TSHR*, *PAX8*, *NKX2-1*, *FOXE1*, and *NKX2-5*) that are involved in fetal thyroid formation. Thyroid dysgenesis ranges in severity from thyroid aplasia or hypoplasia to functional ectopic thyroid tissue. Approximately 40-60% of infants with thyroid gland dysgenesis have some functioning tissue. Laboratory and imaging studies facilitate the determination of the degree of dysgenesis. Thyroid agenesis is suggested by a low serum T4 level with an elevated serum TSH level and undetectable serum thyroglobulin. Newborns with ectopic or hypoplastic thyroid glands manifest low serum T4, elevated serum TSH, and measurable levels of circulating thyroglobulin. Imaging aids in confirming the diagnosis of aplastic, hypoplastic, or ectopic thyroid.

Familial thyroid dyshormonogenesis: Rare autosomal recessive inborn errors of thyroid hormone synthesis, secretion, or uptake also cause congenital hypothyroidism. The following 8 inborn errors have been identified:

* Failure to respond to TSH secondary to defective activation of the thyroid receptor and related cyclic adenosine monophosphate (cAMP) signal transduction pathway
* Defect in trapping of iodide secondary to sodium-iodide symporter failure
* Defective oxidation of iodide to iodine secondary to thyroid peroxidase deficiency
* Defective coupling of iodotyrosines
* Deiodination defects
* Defective thyroglobulin synthesis
* Defective proteolysis of thyroglobulin
* Release of T3 and T4 into the circulation

Partial peripheral resistance to thyroid hormones (autosomal dominant defect): Patients relate a family history of goiter with euthyroidism or hypothyroidism in the face of elevated serum levels of T4 or T3 but nonsuppressed serum TSH concentrations.

Hypopituitarism

Transplacental passage of maternal TSH-binding inhibitory antibodies: This can cause transient neonatal hypothyroidism. In mothers with autoimmune thyroiditis, immunoglobulin G (IgG) antithyroid antibodies can be transmitted across the placenta. These antibodies block binding of TSH to its receptor on the fetal thyroid. The half-life of these antibodies is approximately 1 week, and this form of congenital hypothyroidism usually resolves within 2-3 months of life. Although these infants are asymptomatic, they require thyroid hormone replacement until the pituitary-thyroid axis recovers. Monitoring the infant's serum titer of maternal antibodies is unnecessary, although monitoring serum TSH values is essential for guiding therapy.

Maternal exposure to radioiodine: The fetal thyroid is able to trap iodide by 70-75 days' gestation. Hypothyroidism can develop if the mother is exposed to radioiodine to treat Graves disease or thyroid carcinoma.

Goitrogens: These include iodides found in certain asthma medications, amiodarone, neonatal exposure to iodine-containing antiseptics, propylthiouracil, or methimazole.

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☆ dobandit, in principal autoimun, prin deficit de iod, rar tiroidita subacuta, trt cu litiu, amiodarona, thioamine. Dupa iradiere ( pt alte cancere pana la 30% la 1-10 ani ), euthyoid sick syndrome, histiocitoza X si alte boli infiltative sau tezaurismoze, decompensarea unei tiroide ectopice sau cu un defect de sinteza hh, posttiroidectomie

Clinic

☆ nn cu hipoty congenital:

gestatie prelungita, greutate mare la nastere,

constipatie de la nastere, inclusiv la eliminarea de merconiu

icter prelungit

dificultati de alimentare

plans caracteristic, ragusit, respiratie zgomotoasa

hipotermie

bradicardie, macroglosie, edeme pretibial, umbra cardiaca marita dat pericarditei, hernie ombilicala, fontanele mari

ecg : microvoltaj, bradicardie

☆ hipoty dobandit

scaderea vitezei de crestere cu creste moderata in greutate,

VO intarziata

semne cuatante si neurologice simklare cu adultul, apnee de somn

pseudopubertate precoce izosexuala

gusa

DIAGNOSTIC

TSH - crescut in hipoty primar

- low normal sau low in cel secunar (rar defect izolat de TSH) sau tertiar. ! si la alte semne de afectare hpf: micropenis, defecte de linie mediana, hipoglicemie nn.

TT4 sau fT4 - scazut. ! la deficitul de TBG (T. binding globuline) 1:3000 nn, care modifica doar TT4.

Scintigrama ( de obicei cu Tc99 pt ca nu se organifica si radiatia e mai mica) pt localizarea unei ty ectopice sau pt a confirma un defect de organificare - captare normala dar timp de injumatatire ty mai scurt ca se pierde.

TRATAMENT

screeningul pozitiv trebuie confirmat, nu este 100% sigur asa ca dc are semne de hypoty mai bine repet.

Trt se initiaza imediat dupa recoltarea de sange la nn pt a evita tulburarile de dezvoltare neurologica determinate de inceperea trt dupa 6 sapt de viata. Si trt pana la 2 ani fara pauza. Monitorizez lunar pana cand fT4/ TT4 se normalizeaza apoi la 3 luni pana la 3 ani apoi la 6 luni.

! TSH ul poate ramane crescut cateva luni dupa ce am reglat fT4.

Initiez la nn cu 10-15 mmg/kgc/zi, mai putin dc e

1-5 ani: 100 μg/M2 or 4–6 μg/kg

6-10 ani: 3–4 μg/kg

11 ani si dupa: 2–3 μg/kg

HIPOTY SUBCLINIC

Efecte adverse pe dezvoltarea neurologica - nu exista studii clare, aparent scaderea cap de concentrare.

Risc mai mare de HTA, LDLc crescut

La cei cu DZ1 rism mai mare de hipoglicemii si asociat cu BMI mai mare

!!! Obezitate cu talie inalta si TSH <7 de obicei este efectul obezitatii, nu cauza

In general hypoty subclinica fara autoimunitate este tranzitorie, mai putin daca anticorpii sunt pozitivi sau TSH initial >10.

La copii sub 1 luna la care TSHul nu se normalizeaza se recomanda trt pana la 3 ani, cand se poate tenta o pauza.

la copii peste 3 ani cu hypoty subclinica si anticorpi negativi monitorizez anual (hh si ATPO, ATGL) pentru ca de obicei nu evolueaza.

daca au anticorpii pozitivi, monitorizez la 6- 12 luni, chiar mai frecvent daca TSH> 10 si decid sa nu tratez de la inceput