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Review Article

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Retrospective analysis of Dyshormongenetic Goitre

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Abstract:

Dyshormonogenetic goitre is a rare thyroid entity which occurs due to enzymatic deficiency in the physiological process of thyroxin synthesis resulting in goitre formation. This has to be differentiated from iodine deficiency goitres for their similarity in clinical presentation, hormonal profile and on scintigraphy studies. This differentiation is vital for the reason that Dyshormonogenetic goitre (DHGG) needs to be treated with thyroxin while Iodine deficiency disorder (IDD) requires simple dietary iodine supplementation.

Key Words

Dyshormonogenetic goitre, Hypothyroidism, Perchlorate discharge test, Pendred syndrome, Iodine deficiency disorder, Tc99 scan

Materials and Methods

52 DHGG patients were identified and diagnosed out of 2364 patients treated between December 2001 to December 2011 in our department a referral centre for whole of South India for thyroid disorders. Details collected include age, sex, grade of goitre, nodularity, associated deafness, The tests done include Thyroid function tests, Perchlorate discharge test, USG neck, Scintigraphy study, Fine needle aspiration cytology & histopathological examination. In these 52 patients, fifty patients were between 15 and20 years of age; 30 patients were males and 22 patients were females. Siblings belonging to 3 different families showed features of DHGG. All the 52 presented with grade III goitres. All patients had hypothyroidism at the time of presentation. USG showed nodularity in all cases. 1 Patient presented with mental retardation. This was present in the families were siblings were affected. One patient had associated deafness, Pendred syndrome. None of them had malignancy.I-131 scan was done in 35 patients and Tc 99 scan was done in 17 patients. Scintigraphy study showed increased uptake. I131 scan also showed low liver counts. In order to differentiate DHGG from IDD perchlorate discharge test and 24 hour urinary iodine measurements were done. 24 hour urinary iodine was above 200 ug/day. These specific investigations revealed the defect to be at the organification level. None of these patients showed iodine deficiency status thus

clearly showing the distinction from iodine deficiency goitre.

All 52 patients were subjected to Total thyroidectomy after one month preparation with thyroxin. Thyrotherapy not only corrects the hypothyroid status but also helps to reduce the vascularity of the gland.

Discussion:

In DHGG there is defect in the hormone synthesis ([1]) indicated by low T3, T4 and high TSH. Scintigraphy shows high uptake ([2]) except DHGG due to trapping defect. There are three important steps in thyroxin synthesis: 1) Trapping - Iodide is trapped by the thyroid gland 2) Organification – trapped iodide is converted to iodine and with thysine Monoiodothyrosine (MIT) and diiodothyrosine (DIT) are formed. 3) Coupling - MIT and DIT couple to form T3 and T4.Trapping defect DHGG has low uptake in scintigraphy studies. It also shows a low salivary: plasma radioiodide ratio of 1:1 (Normal: 10:1) ([3]). Organification defect is confirmed by perchlorate discharge test ([4]). This is done in the following way; radiotracer is given and an uptake test is done, following which 1 Gm of potassium perchlorate is given orally and an uptake is repeated after 2 hours. A fall in uptake between 10-20% indicates organification defect. Coupling defect will show MIT and DIT in plasma whereas it is normally absent. They are usually present in the first or second decade with or without a family history. When DHGG runs in families it is associated with mental retardation ([5]) and Pendred syndrome ([6, 7]). DHGGs are due to TG gene mutation ([8]) or DHGG presents with huge goiters ([9]), soft and highly vascular.

Differential diagnosis of IDD should be thought of when there is hypothyroidism with increased uptake by scintigaphy studies. IDD has a low 24 hour urinary iodine level. Aa a result of long standing hypothyroidism with sustained high levels of TSH in DHGG the thyroid gland shows a highly cellular picture in FNAC and a HPE of such a gland shows papillary proliferation with papillary fronds, nuclear atypia and minimal amount of colloid and hence it is mistaken for thyroid cancer ([10]). There is increased incidence of malignancy due to overexpression of EGF and EGF-R m RNAs in DHGG ([11]).

Conclusion:

Whenever Dyshormonogenetic goitres present as diffuse goitres they must be treated with thyrotherapy. Only when they present as nodular goitres surgery is indicated. Carcinomatous change can occur if DHGGs are not treated with thyrotherapy. Surgery of choice is total thyroidectomy. After thyroidectomy they require lifelong replacement with thyroxine.

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Images:

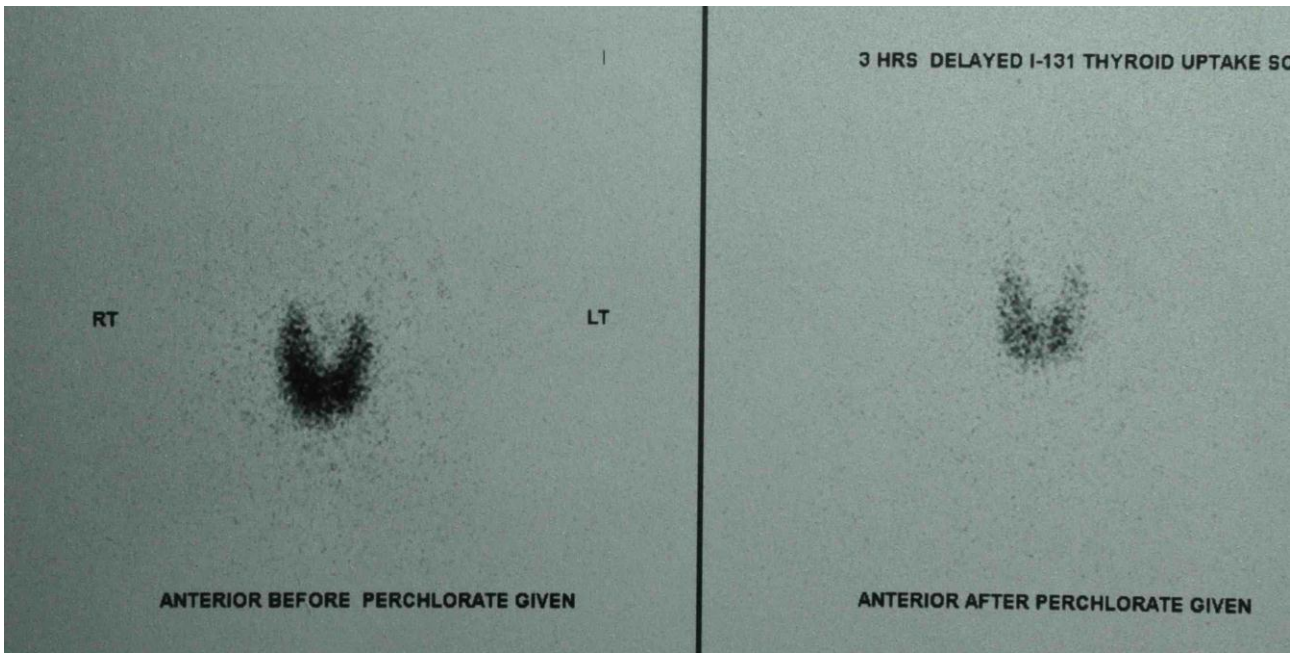

Image showing perchlorate discharge test