

FAMILIAL DYSALBUMINEMIC HYPERTHYROXINEMIA

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- thyroid hormones (mainly T_4) are made by the follicular cells of the thyroid gland and released into the blood (Figure 1)
- most (99.9%) thyroid hormone in the blood is attached (bound) to proteins:
 - thyroxine binding globulin - TBG (~75%)
 - thyroxine binding prealbumin- TBPA (~55%)
 - albumin (~10%)
- only hormone that is not bound to proteins is responsible for its effects in the body
- in this disorder an abnormal type of albumin is made that is able to bind more thyroid hormone than normal leading to misleading test results
- the “apparent” hyperthyroidism is not true and hence the patient should receive no treatment

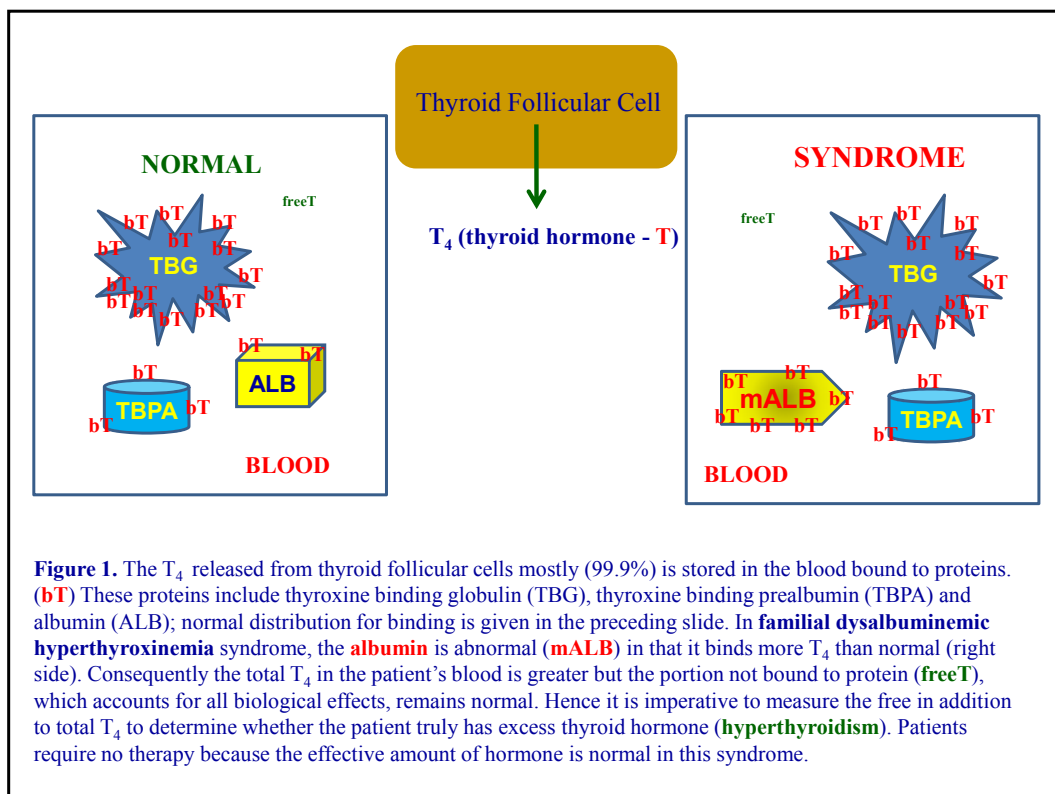


Figure 1. The T_4 released from thyroid follicular cells mostly (99.9%) is stored in the blood bound to proteins. (**bT**) These proteins include thyroxine binding globulin (TBG), thyroxine binding prealbumin (TBPA) and albumin (ALB); normal distribution for binding is given in the preceding slide. In **familial dysalbuminemic hyperthyroxinemia** syndrome, the **albumin** is abnormal (**mALB**) in that it binds more T_4 than normal (right side). Consequently the total T_4 in the patient's blood is greater but the portion not bound to protein (**freeT**), which accounts for all biological effects, remains normal. Hence it is imperative to measure the free in addition to total T_4 to determine whether the patient truly has excess thyroid hormone (**hyperthyroidism**). Patients require no therapy because the effective amount of hormone is normal in this syndrome.