FAMILIAL DYSALBUMINEMIC HYPERTHYROXINEMIA

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- Thyroid hormones (mainly T4) 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.