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A novel frameshift mutation causes early termination of the TBG protein and comp

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Two Novel Mutations in the Serpina7 Gene Are Associated ...

www.ncbi.nlm.nih.gov › ... › Eur Thyroid J › v.4(Suppl 1); 2015 Sep

Thyroxine-binding **globulin** (TBG) is the main transport **protein** for T 4 in blood. Until now, 22 mutations leading to **complete TBG deficiency** (TBG-CD) have been reported. **TBG deficiency** is usually suspected when discrepant free and total TH are found. As the Serpina7 gene is located on the X ...

Cited by: 1 **Author:** Lars C. Moeller, Yaw Appiagyei-Dankah, ...
Publish Year: 2015

A Novel Mutation Causing Complete Thyroxine-Binding ...

https://www.researchgate.net/publication/12264456_**A_Novel_Mutation**...

In this study we **report a novel** single **mutation** causing **complete TBG deficiency** due to a deletion of the last base of codon 38 (exon 1), which led to a **frame shift** resulting in a premature stop at ...

(PDF) Complete thyroxine-binding globulin (TBG) deficiency ...

https://www.researchgate.net/publication/13516826_**Complete**...

PDF | Fourteen T4-binding **globulin** (TBG) variants have been identified at the gene level. They are all located in the coding region of the gene and 6 produce **complete deficiency** of TBG (TBG-CD).

Thyroxine-Binding Globulin Deficiency - Medscape

misc.medscape.com/pi/android/medscapeapp/html/A125764-business.html

A novel mutation causing **complete deficiency** of thyroxine binding **globulin**. Clin Endocrinol (Oxf). 1997 Jul. 47(1):1-5. [View Abstract] Carvalho GA, Weiss RE, Refetoff S. **Complete** thyroxine-binding **globulin** (TBG) **deficiency** produced by a **mutation** in acceptor splice site causing **frameshift** and **early termination** of translation (TBG-Kankakee).

Thyroxine-Binding Globulin Deficiency: Overview, Molecular ...

https://**emedicine.medscape.com**/article/125764-overview

Sep 14, 2017 - **A novel mutation** causing **complete deficiency** of thyroxine binding **globulin**. Clin Endocrinol (Oxf). 1997 Jul. 47(1):1-5. . Carvalho GA, Weiss RE, Refetoff S. **Complete** thyroxine-binding **globulin** (TBG) **deficiency** produced by a **mutation** in acceptor splice site causing **frameshift** and **early termination** of translation (TBG-Kankakee).

Name of Journal: *World Journal of Clinical Cases*

Manuscript NO: 48393

Manuscript Type: CASE REPORT

Novel frameshift mutation causes early termination of the thyroxine-binding globulin protein and complete thyroxine-binding globulin deficiency in a Chinese family: A case report

Ping-Ping Dang, Wei-Wei Xiao, Zhong-Yan Shan, Yue Xi, Ran-Ran Wang, Xiao-Hui Yu, Wei-Ping Teng, Xiao-Chun Teng

Abstract

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First report of inherited thyroxine-binding globulin ...

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First **report** of inherited thyroxine-binding **globulin deficiency in** Iran caused by a known de novo **mutation** in SERPINA7 Fahimeh Soheilipour , a Hassan Fazilaty , b, 1 Fatemeh Jesmi , c William A. Gahl , d, e and Babak Behnam b, d, e, ⁎, 2

Cited by: 1 **Author:** Fahimeh Soheilipour, Hassan Fazilaty, Fa...
Publish Year: 2016

Two Novel Mutations in the Serpina7 Gene Are Associated ...

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Publish Year: 2015

Two novel mutations in the gene encoding thyroxine ...

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Two **novel** mutations in the gene encoding thyroxine-binding **globulin** (TBG) as a cause of **complete** TBG **deficiency** in Taiwan Article in Clinical Endocrinology 58(4):409-14 · April 2003 with 14 Reads

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Two **Novel Mutations** in the Serpina7 Gene Are **Associated** with Complete **Deficiency of Thyroxine-Binding Globulin Lars** C. Moeller , a, * Yaw Appiagyei-Dankah , c Birgit Köhler , b Heike Biebermann , b Onno E. Janssen , a and Dagmar **Führer** a

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Thyroxine-binding Globulin

Thyroxine-binding globulin (TBG) is a globulin that binds thyroid hormones in circulation. It is one of three transport proteins (along with transthyretin and serum albumin) responsible for carrying the thyroid hormones thyroxine (T_4) and triiodothyronine (T_3) in the bloodstream. Of these three proteins, TBG has the highest affinity for T_4 and T_3 but is present in the lowest concentration. Despite its low concentration, TBG carries the majority of T_4 in the blood plasma. Due to the very low concentration of T_4 and T_3 in the blood, TBG is rarely more than 25% saturated with its ligand. Unlike transthyretin and albumin, TBG has a single binding site for T_4/T_3. TBG is synthesized primarily in the liver as a 54-kDa protein. In terms of genomics, TBG is a serpin; however, it has no inhibitory function like many other members of this class of proteins.

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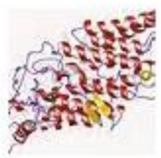
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