Name of Journal: *World Journal of Clinical Cases*

Manuscript NO: 59597

Manuscript Type: CASE REPORT

Adult onset type 2 familial hemophagocytic lymphohistiocytosis with *PRF1* c.65delC/c.163C>T compound heterozygous mutations: A case report

Xin-Yi Liu, Yan-Bo Nie, Xue-Jing Chen, Xiao-Hui Gao, Li-Jia Zhai, Feng-Ling Min

Abstract

BACKGROUND

Familial hemophagocytic lymphohistiocytosis (FHL) is a primary immunodeficiency disease caused by gene defects. The onset of FHL in adolescents and adults may lead clinicians to ignore or even misdiagnose the disease. To our knowledge, we are the first to describe in detail the clinical features of type 2 FHL (FHL2) with compound

22,400 Results Any time

(PDF) Type 2 familial hemophagocytic lymphohistiocytosis ...

https://www.researchgate.net/publication/326615341...

Familial hemophagocytic lymphohistiocytosis type 2 (FHL2), caused by perforin 1 (**PRF1**), is a genetic disorder of lymphocyte cytotoxicity that usually presents in the first 2 years of life and has ...

Hemophagocytic Lymphohistiocytosis, Familial - ...

https://www.ncbi.nlm.nih.gov/books/NBK1444/?report=printable

Mar 22, 2006 · **Familial hemophagocytic lymphohistiocytosis** (FHL) is characterized by proliferation and infiltration of hyperactivated macrophages and T-lymphocytes manifesting as acute illness with prolonged fever, cytopenias, and hepatosplenomegaly. **Onset** is typically within the first months or years of life and, on occasion, in utero, although later childhood or **adult onset** is more common than ...

Cited by: 9 **Author:** Kejian Zhang, Alexandra H Filipovich, Jud...
Publish Year: 2013

Familial hemophagocytic lymphohistiocytosis in an adult ...

https://www.researchgate.net/publication/6833083_Familial_hemophagocytic...

Since the discovery of perforin gene **mutations** in **familial hemophagocytic lymphohistiocytosis** (FHL) **type 2**, heterogeneous features in FHL2 patients have been identified in a **report** of Feldmann et ...

Adult-onset Primary Hemophagocytic Lymphohistiocytosis ...

https://www.researchgate.net/publication/338723687...

Genetic testing identified **compound heterozygous mutations** in **PRF1** (Patients 1 and 2) and UNC13D (Patient 3), with no evidence of systemic disease except decreased NK-cell function.

Primary hemophagocytic lymphohistiocytosis in adults: the ...

https://link.springer.com/article/10.1186/s13023-017-0753-7

Jan 22, 2018 · **PRF1** had **compound heterozygous** pathogenic variants, including a **heterozygous** missense pathogenic variant at c.172 T > C and a non-frameshift pathogenic variant at c.1083_1094del. Family surveys of P03 showed that P03 had suspected a positive family history of HLH, as two elder siblings of P03 died from unexplained fevers in their childhoods.

Cited by: 10 **Author:** Zhili Jin, Yini Wang, Jingshi Wang, Jia Zh...
Publish Year: 2018

23,500 Results Any time

Hemophagocytic Lymphohistiocytosis, Familial - GeneReviews ...

https://www.ncbi.nlm.nih.gov/books/NBK1444/?report=printable

Mar 22, 2006 · **Familial hemophagocytic lymphohistiocytosis** (FHL) is characterized by proliferation and infiltration of hyperactivated macrophages and T-lymphocytes manifesting as acute illness with prolonged fever, cytopenias, and hepatosplenomegaly. **Onset** is typically within the first months or years of life and, on occasion, in utero, although later childhood or **adult onset** is more common than ...

Cited by: 9 **Author:** Kejian Zhang, Alexandra H Filipovich, Judit...
Publish Year: 2013

(PDF) Type 2 familial hemophagocytic lymphohistiocytosis ...

https://www.researchgate.net/publication/326615341...

Familial hemophagocytic lymphohistiocytosis type 2 (FHL2), caused by perforin 1 (**PRF1**), is a genetic disorder of lymphocyte cytotoxicity that usually presents in the first **2** years of life and has ...

Familial hemophagocytic lymphohistiocytosis in an adult ...

https://www.researchgate.net/publication/6833083_Familial_hemophagocytic...

Since the discovery of perforin gene **mutations** in **familial hemophagocytic lymphohistiocytosis** (FHL) **type 2**, heterogeneous features in FHL2 patients have been identified in a **report** of Feldmann et ...

Primary hemophagocytic lymphohistiocytosis in adults: the ...

https://link.springer.com/article/10.1186/s13023-017-0753-7

Jan 22, 2018 · **PRF1** had **compound heterozygous** pathogenic variants, including a **heterozygous** missense pathogenic variant at c.172 T > C and a non-frameshift pathogenic variant at c.1083_1094del. Family surveys of P03 showed that P03 had suspected a positive family history of HLH, as two elder siblings of P03 died from unexplained fevers in their childhoods.

Cited by: 11 **Author:** Zhili Jin, Yini Wang, Jingshi Wang, Jia Zhan...
Publish Year: 2018

Late-onset cases of familial hemophagocytic ...

https://www.researchgate.net/publication/6538572_Late-onset_cases_of_familial...

Familial hemophagocytic lymphohistiocytosis type 2 (FHL2), caused by perforin 1 (**PRF1**), is a genetic disorder of lymphocyte cytotoxicity that usually presents in the first **2** years of life and has ...

1 2 3 4 5

Search Tools

Turn off Hover Translation (关闭

22,600 Results Any time Open links in new tab

(PDF) Type 2 familial hemophagocytic lymphohistiocytosis ...

https://www.researchgate.net/publication/326615341...

Familial hemophagocytic lymphohistiocytosis type 2 (FHL2), caused by perforin 1 (**PRF1**), is a genetic disorder of lymphocyte cytotoxicity that usually presents in the first **2** ...

Hemophagocytic Lymphohistiocytosis, Familial - GeneReviews ...

https://www.ncbi.nlm.nih.gov/books/NBK1444/?report=printable

Mar 22, 2006 · **Familial hemophagocytic lymphohistiocytosis** (FHL) is characterized by proliferation and infiltration of hyperactivated macrophages and T-lymphocytes manifesting as...

Cited by: 9 **Author:** Kejian Zhang, Alexandra H Filipovich, J...
Publish Year: 2013

Familial hemophagocytic lymphohistiocytosis in an adult ...

https://www.researchgate.net/publication/6833083_Familial_hemophagocytic...

Since the discovery of perforin gene **mutations** in **familial hemophagocytic lymphohistiocytosis** (FHL) **type 2**, heterogeneous features in FHL2 patients have been identified in a **report** ...

The risk of hemophagocytic lymphohistiocytosis in ...

https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3624940

Apr 11, 2013 · Introduction. Hermansky-Pudlak syndrome **type 2** (HPS2) is a rare autosomal-recessive disorder of lysosomal trafficking caused by **mutations** in the gene encoding the β3A...

Cited by: 80 **Author:** Birthe Jessen, Sebastian F N Bode, San...
Publish Year: 2013

Late-onset cases of familial hemophagocytic ...

https://www.researchgate.net/publication/6538572_Late-onset_cases_of_familial...

Familial hemophagocytic lymphohistiocytosis type 2 (FHL2), caused by perforin 1 (**PRF1**), is a genetic disorder of lymphocyte cytotoxicity that usually presents in the first **2** ...

Primary hemophagocytic lymphohistiocytosis in adults: the ...

https://link.springer.com/article/10.1186/s13023-017-0753-7

Jan 22, 2018 · **PRF1** had **compound heterozygous** pathogenic variants, including a **heterozygous** missense pathogenic variant at c.172 T > C and a non-frameshift pathogenic variant at...

Perforin gene mutations in adult-onset hemophagocytic ...

Search Tools

Turn off Hover Translation (关闭取词)