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Manuscript Type: CASE REPORT

Compound heterozygous mutations in the NEU1 gene in type 1 sialidosis: Case report and literature review

Clinical and genetic features of type 1 sialidosis

Abstract

BACKGROUND

Type 1 sialidosis, also known as cherry-red spot-myoclonus syndrome, is a rare autosomal recessive lysosomal storage disorder presenting in the second decade of life with the most common symptoms of myoclonus, ataxia and seizure and has rarely been reported in Chinese mainland.

CASE SUMMARY

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Type 1 Sialidosis Patient With a Novel Deletion Mutation ...

https://www.ncbi.nlm.nih.gov/pubmed/30635863

Type 1 sialidosis (OMIM#256550) is a rare autosomal recessive lysosomal storage disease caused by a mutation in the NEU1 (OMIM * **608272**) gene. In this study, we aimed to review the previous reports of type 1 sialidosis and compare those with the first case of type 1 sialidosis in Korea.

Cited by: 7 **Author:** Jong Hyeon Ahn, Ah Reum Kim, Chung Lee,...

Publish Year: 2019

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The patient's gene analysis identified compound heterozygous mutation in the NEU1 that is shown to be associated with the sialidosis type 1. In this very rarely seen case, we found volume ...

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Type 1 Sialidosis Patient With a Novel Deletion Mutation ...

https://link.springer.com/article/10.1007/s12311-019-1005-2

Jan 11, 2019 · Recent advances in next-generation sequencing technologies have uncovered the genetic backgrounds of various diseases. Type **1** sialidosis (**OMIM#256550**) is a rare autosomal recessive lysosomal storage disease caused by a mutation in the NEU1 (OMIM * 608272) gene. In this study, we aimed to review the previous reports of type 1 sialidosis and compare those with the first case of type ...

Cited by: 7 **Author:** Jong Hyeon Ahn, Ah Reum Kim, Chung Lee,...

Publish Year: 2019

Type 1 sialidosis presenting with ataxia, seizures and ...

https://europepmc.org/article/MED/30023283

Jan 12, 2018 · **Type 1 Sialidosis** Patient With a Novel Deletion **Mutation in the NEU1 Gene**: **Case Report**

Novel mutations in the neuraminidase-1 (NEU1) gene in two ...

https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3612311

Sialidosis is a rare, autosomal recessive lysosomal storage disorder resulting from deficiency of neuraminidase-1 (also known as lysosomal sialidase) - the enzyme responsible for catalyzing the hydrolysis of the terminal sialic acid residues of **sialylated glyco-conjugates**. Deficiency of the enzyme leads to progressive intracellular accumulation of sialylated glycopeptides and oligosaccharides.

Cited by: 9 **Author:** Prajnya Ranganath, Vishakha Sharma, Sum...
Publish Year: 2012

Type 1 Sialidosis Patient With a Novel Deletion Mutation ...

https://pubmed.ncbi.nlm.nih.gov/30635863

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Cited by: 7 **Author:** Jong Hyeon Ahn, Ah Reum Kim, Chung Lee,...
Publish Year: 2019

Child Neurology: Type 1 sialidosis due to a novel mutation ...

https://n.neurology.org/content/90/13/622

Mar 27, 2018 · **Sialidosis** is an autosomal **recessive lysosomal storage disorder** caused by **homozygous or compound heterozygous mutations in the NEU1 gene on chromosome 6p21.33**, encoding sialic acid cleaving enzyme sialidase 1 or neuraminidase 1.

Cited by: 8 **Author:** Akilandeswari Aravindhan, Aravindhan Vee...
Publish Year: 2018

Clinical and electrophysiological characteristics of a ...

https://www.sciencedirect.com/science/article/pii/S0929664619303328

Jan 01, 2020 · **Sialidosis is a rare autosomal recessive lysosomal storage disease caused by a mutation in the Neuraminidase 1 (NEU1) gene encoding the sialic acid-cleaving enzyme neuraminidase, which leads to decreased enzymatic activity and accumulation of sialyloligosaccharides in tissues**.1, 2 Sialidosis can be subdivided into two clinical types according to age of onset and severity. 3 Type 1 ...