'Doctor Google' ending the diagnostic odyssey in lysosomal storage disorders: parents using internet search engines as an efficient diagnostic strategy in rare diseases
Bouwman, M.G.; Teunissen, Q.G.A.; Wijburg, F.A.; Linthorst, G.E.

Published in:
Archives of disease in childhood

DOI:
10.1136/adc.2009.171827

Citation for published version (APA):

General rights
It is not permitted to download or to forward/distribute the text or part of it without the consent of the author(s) and/or copyright holder(s), other than for strictly personal, individual use, unless the work is under an open content license (like Creative Commons).

Disclaimer/Complaints regulations
If you believe that digital publication of certain material infringes any of your rights or (privacy) interests, please let the Library know, stating your reasons. In case of a legitimate complaint, the Library will make the material inaccessible and/or remove it from the website. Please Ask the Library: http://uba.uva.nl/en/contact, or a letter to: Library of the University of Amsterdam, Secretariat, Singel 425, 1012 WP Amsterdam, The Netherlands. You will be contacted as soon as possible.
‘Doctor Google’ ending the diagnostic odyssey in lysosomal storage disorders: parents using internet search engines as an efficient diagnostic strategy in rare diseases

Machtelt G Bouwman,1 Quirine G A Teunissen,1 Frits A Wijburg,1 Gabor E Linthorst2

ABSTRACT
The expansion of the internet has resulted in widespread availability of medical information for both patients and physicians. People increasingly spend time on the internet searching for an explanation, diagnosis or treatment for their symptoms. Regarding rare diseases, the use of the internet may be an important tool in the diagnostic process. The authors present two cases in which concerned parents made a correct diagnosis of a lysosomal storage disorder in their child by searching the internet. These cases illustrate the utility of publicly available internet search engines in diagnosing rare disorders and in addition illustrate the lengthy diagnostic odyssey which is common in these disorders.

INTRODUCTION
The expansion of the internet has resulted in widespread public access to both scientific and non-scientific information. When confronted with a health complaint, people increasingly spend time on the internet searching for a diagnosis or treatment. Many physicians nowadays are confronted with patients or parents who present potential diagnoses and treatments for their complaints, which were retrieved from the internet. This behaviour undoubtedly influences patient/physician relationship. The suggested diseases or remedies found on the internet may not be relevant to the patient’s health issues, presenting rare diagnoses or treatment strategies that do not fall within the realm of conventional medicine for more common complaints and disorders. But sometimes these internet searches may reveal the correct diagnosis for rare diseases, which were not considered by the patients’ physician. Here, we describe two cases where, after a long doctor’s delay, concerned parents diagnosed a lysosomal storage disorder (LSD) in their child by searching the internet.

CASE A
An 11-year-old boy was referred to our metabolic centre after his parents had suggested a diagnosis of Fabry disease to their paediatrician. From the age of 5 years he had suffered from severe episodic pains in the hands and feet, especially during exercise and fever. In addition, he had unexplained recurrent fever, joint aches and a skin rash on hands and feet. Three different paediatricians were consulted: a general paediatrician and two specialists in periodic fever syndromes and immunology, respectively. This led to the consideration of the following diagnoses: growing pains, juvenile idiopathic arthritis, hyper-IgD syndrome, tumour necrosis factor receptor-1-associated periodic syndrome and familial Mediterranean fever. Extensive laboratory tests were performed, but none of these diagnoses could be confirmed. Because symptoms persisted, the concerned parents initiated a search on internet, using ‘Google’ as search engine. They used the following search terms (in Dutch): unexplained recurrent fever, pain in feet and skin rash. Browsing the internet for several hours, they found a picture of a skin rash in a patient with Fabry disease. They immediately recognised the skin rash as similar to their child’s, and after studying all available information, they concluded that their child might suffer from Fabry disease. On physical examination, angiokeratoma were present on the hands, feet, knees and hips (figure 1A). The consulted ophthalmologist found cornea verticillata. The diagnosis of the LSD Fabry disease was confirmed by enzyme analysis. There

Figure 1 (A) Patient A: angiokeratoma on the palmar side of the hands. Right hand ulnar side and left hand ulnar side and thenar. (B) Bowed fingers on both hands on patient B.
were no signs of renal and cardiac involvement, as often seen at this age. He was started on enzyme replacement therapy (ERT) within the scope of an early intervention trial (FIELD study, NCT00701415).

Fabry disease is an X linked LSD caused by a deficiency of the enzyme α-galactosidase A (αGal-A). The main substrate of αGal-A, globotriaosylceramide (Gb-3), accumulates in endothelial cells and other cell types. In childhood, Fabry disease is characterised by severe episodic pains in the hands and feet (acroparasthesia), anhidrosis and angiokeratoma. In adult life, disease progression causes renal, cardiovascular and neurological morbidity, and a reduced life-span. Since 2001, therapy has become available as ERT. Treatment with ERT reduces tissue Gb-3 and may stabilise renal disease and other disease features.3

CASE B

A boy, aged 16 months, was referred to a metabolic centre, after his mother suspected that he might have mucopolysaccharidosis type I (MPS I). Born prematurely at 33 weeks (1790 g) from non-consanguineous parents, the boy needed continuous pulmonary airway pressure during his first 5 days of life. He failed his neonatal hearing tests. In the following 3 months, he was admitted several times because of excessive crying.

At the age of 3 months, he was seen because of macrocephaly. A mild hydrocephalus without signs of high pressure was diagnosed on MRI. A wait-and-see policy was adopted. At the age of 5 months, a one-sided inguinal hernia was corrected.

The child was referred to a clinical geneticist at the age of 7 months, primarily for investigation of myotonic dystrophy type I, an autosomal dominant disorder that had been diagnosed in his father. Apart from the possibility of myotonic dystrophy, the geneticist was also asked to consider other diagnoses, in view of his symptoms. No alternative diagnosis was suggested by the clinical geneticist.

The child had recurrent upper airway infections during his first year of life and was noted to snore significantly during sleep. At the age 10 months, an adenectomy was performed, grommets were placed, and he received hearing aids. His parents noticed kyphosis, changing facial features, bowed fingers (figure 1B), an umbilical hernia and slowing of psychomotor development. This latter observation resulted in cognitive testing, which revealed a mild hydrocephalus without signs of high pressure was diagnosed on MRI. A wait-and-see policy was adopted. At the age of 5 months, a one-sided inguinal hernia was corrected.

The child was referred to a clinical geneticist at the age of 7 months, primarily for investigation of myotonic dystrophy type I, an autosomal dominant disorder that had been diagnosed in his father. Apart from the possibility of myotonic dystrophy, the geneticist was also asked to consider other diagnoses, in view of his symptoms. No alternative diagnosis was suggested by the clinical geneticist.

The child had recurrent upper airway infections during his first year of life and was noted to snore significantly during sleep. At the age 10 months, an adenectomy was performed, grommets were placed, and he received hearing aids. His parents noticed kyphosis, changing facial features, bowed fingers (figure 1B), an umbilical hernia and slowing of psychomotor development. This latter observation resulted in cognitive testing, which revealed a mild hydrocephalus without signs of high pressure was diagnosed on MRI. A wait-and-see policy was adopted. At the age of 5 months, a one-sided inguinal hernia was corrected.

The child was referred to a clinical geneticist at the age of 7 months, primarily for investigation of myotonic dystrophy type I, an autosomal dominant disorder that had been diagnosed in his father. Apart from the possibility of myotonic dystrophy, the geneticist was also asked to consider other diagnoses, in view of his symptoms. No alternative diagnosis was suggested by the clinical geneticist.

The child had recurrent upper airway infections during his first year of life and was noted to snore significantly during sleep. At the age 10 months, an adenectomy was performed, grommets were placed, and he received hearing aids. His parents noticed kyphosis, changing facial features, bowed fingers (figure 1B), an umbilical hernia and slowing of psychomotor development. This latter observation resulted in cognitive testing, which revealed a mild hydrocephalus without signs of high pressure was diagnosed on MRI. A wait-and-see policy was adopted. At the age of 5 months, a one-sided inguinal hernia was corrected.

The child was referred to a clinical geneticist at the age of 7 months, primarily for investigation of myotonic dystrophy type I, an autosomal dominant disorder that had been diagnosed in his father. Apart from the possibility of myotonic dystrophy, the geneticist was also asked to consider other diagnoses, in view of his symptoms. No alternative diagnosis was suggested by the clinical geneticist.

The child had recurrent upper airway infections during his first year of life and was noted to snore significantly during sleep. At the age 10 months, an adenectomy was performed, grommets were placed, and he received hearing aids. His parents noticed kyphosis, changing facial features, bowed fingers (figure 1B), an umbilical hernia and slowing of psychomotor development. This latter observation resulted in cognitive testing, which revealed a mild hydrocephalus without signs of high pressure was diagnosed on MRI. A wait-and-see policy was adopted. At the age of 5 months, a one-sided inguinal hernia was corrected.

The child was referred to a clinical geneticist at the age of 7 months, primarily for investigation of myotonic dystrophy type I, an autosomal dominant disorder that had been diagnosed in his father. Apart from the possibility of myotonic dystrophy, the geneticist was also asked to consider other diagnoses, in view of his symptoms. No alternative diagnosis was suggested by the clinical geneticist.

The child had recurrent upper airway infections during his first year of life and was noted to snore significantly during sleep. At the age 10 months, an adenectomy was performed, grommets were placed, and he received hearing aids. His parents noticed kyphosis, changing facial features, bowed fingers (figure 1B), an umbilical hernia and slowing of psychomotor development. This latter observation resulted in cognitive testing, which revealed a mild hydrocephalus without signs of high pressure was diagnosed on MRI. A wait-and-see policy was adopted. At the age of 5 months, a one-sided inguinal hernia was corrected.
REFERENCES


'Doctor Google' ending the diagnostic odyssey in lysosomal storage disorders: parents using internet search engines as an efficient diagnostic strategy in rare diseases

Machtelt G Bouwman, Quirine G A Teunissen, Frits A Wijburg, et al.

Arch Dis Child 2010 95: 642-644 originally published online April 23, 2010
doi: 10.1136/adc.2009.171827

Updated information and services can be found at:
http://adc.bmj.com/content/95/8/642.full.html

These include:

References
This article cites 13 articles, 4 of which can be accessed free at:
http://adc.bmj.com/content/95/8/642.full.html#ref-list-1

Article cited in:
http://adc.bmj.com/content/95/8/642.full.html#related-urls

Email alerting service
Receive free email alerts when new articles cite this article. Sign up in the box at the top right corner of the online article.

Topic Collections
Articles on similar topics can be found in the following collections

Editor's choice (1159 articles)
Metabolic disorders (12299 articles)

Notes

To request permissions go to:
http://group.bmj.com/group/rights-licensing/permissions

To order reprints go to:
http://journals.bmj.com/cgi/reprintform

To subscribe to BMJ go to:
http://group.bmj.com/subscribe/