CORRESPONDENCE

Exome Sequencing in Children

Undiagnosed Developmental Delay and Neurological Illness

by Elisa A. Mahler, Dr. med. Jessika Johannsen, Dr. med. Konstantinos Tsiakas, Dr. med. Katja Kloth, Dr. med. Sabine Lüttgen, Prof. Dr. med. Chris Mühlhausen, Bader Alhaddad, Dr. med. Tobias B. Haack, PD Dr. med. Tim M. Strom, Dr. rer. nat. Fanny Kortüm, Prof. Dr. med. Thomas Meitinger, Prof. Dr. med. Ania C. Muntau, Prof. Dr. med. René Santer, Prof. Dr. med. Christian Kubisch, Dr. med. Davor Lessel, PD Dr. med. Jonas Denecke, and Dr. med. Maja Hempel in issue 12/2019

Billing-Related Problems

This article may create the impression in readers that in the presence of a “global developmental disorder,” individual out-of-hospital exome sequencing is covered by German health insurance under EBM fee schedule code number 11514 (1). This assumption is incorrect. In the preamble to chapter 11 of the EBM (the uniform value scale), section 4, the prerequisite for insurance coverage is suspicion of a rare disorder with a maximum prevalence of five in 10 000. Regarding the prevalence of a “global developmental disorder,” the authors name a rate of 1% (100 in 10 000). The compulsory provisions of the EBM rule out human genetic investigation of disorders occurring at such a low prevalence. Encouraging the parents of children with disabilities to apply for healthcare services that aren’t covered by health insurance providers’ defined services, will inevitably lead to disappointment. It is regrettable that readers’ expectations regarding what is covered by the EBM may have been raised as a result of the incorrect explanation/information in the article. Unfortunately the article did not include any mention of support for relevant diagnostic evaluation from the innovation fund of the Federal Joint Committee (G-BA) in the context of the TRANSLATE-NAMSE project; this project runs outside the EBM.

DOI: 10.3238/arztebl.2019.0487a

References

1. Mahler E A, Johannsen J, Tsiakas K, et al.: Exome sequencing in children—undiagnosed developmental delay and neurological illness. *Dtsch Arztebl Int* 2019; 116: 197–204.

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In Reply:

We thank Dr. von Zastrow for his correspondence, especially since he highlights the lack of clarity around the funding of exome sequencing.

It was not the intention of our article to explore the funding options for exome sequencing (1).

We are, however, perfectly aware that for many colleagues who provide healthcare to patients with suspected genetic disorders, this is a question of central importance.

It is correct that the EBM covers fee schedule code number 11514 under the proviso that genetic diagnostic evaluation was undertaken if a rare disorder was suspected. Undoubtedly, those genetic disorders underlying the clinical characteristic of global developmental disorder are rare. This is shown in our study, which found 20 different genetic diagnoses in 21 patients whose disorder was diagnosed; many international studies support this finding. The requirement of a suspected rare disorder is thus met. In spite of this, an application for reimbursement under EBM fee schedule number 11514 will usually be refused by statutory health insurers. We did mention this in our article (1).

The presented study did not take recourse to the time-limited national innovation fund project TRANSLATE-NAMSE. This project is not the answer to the need for creating a funding structure for indicated exome sequencing that is secure in the long term.

In our opinion, what is urgently required is the creation of appropriate structural and financial conditions so as to enable patients with undiagnosed diseases to access interdisciplinary treatment, including comprehensive phenotyping and wide-ranging genetic diagnostic evaluation. In many places, centers for rare diseases already provide the clinical structures. However, funding for diagnostic evaluation has not yet been secured.

DOI: 10.3238/arztebl.2019.0487b

References

1. Mahler E A, Johannsen J, Tsiakas K, et al.: Exome sequencing in children—undiagnosed developmental delay and neurological illness. *Dtsch Arztebl Int* 2019; 116: 197–204.

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Conflict of interest statement

The authors of both contributions declare that no conflict of interest exists.