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CENTER FOR ANATOMY AND CELL BIOLOGY

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CC:

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Ward of Paediatric and Adolescent Medicine
Dr. Mika-Michaela Rappold
1100 Wien, Kundratstrasse 3

Vienna, 05 February 2024

Person concerned: **HEINER, Bruno, date of birth: 11 October 2016**

Molecular genetic examination on the dystrophin gene (OMIM * 300377)

Starting clinical diagnosis:	Duchenne muscular dystrophy (OMIM #310200)
Question:	Mutation in the dystrophin gene
Examination material:	obtained from blood treated with EDTA
Sample received:	29 January 2024
Examination methods:	Multiplex ligation-dependent probe amplification (MLPA, P034-B2, P035-B1)
Sequences examined:	Muscle promoter, brain promoter, all 79 exons of the dystrophin gene

Result and summary

In the dystrophin gene (DMD) of **patient HEINER, Bruno**, a **deletion present in the hemizygotic form in exon 46-47 was established** using the MLPA method [NM_004006.2:c.(6614+1_6615-1)_(6912+1_6913-1)del]. The detected deletion disrupts the RNA reading frame ("out-of-frame"), thus **formally conforms to a Duchenne type dsytrophinopathy**.

Therefore the established molecular genetic finding conforms to the dystrophinopathy occurring in the starting clinical diagnosis; it is most likely a Duchenne type (OMIM # 310200).

It is recommended that the mother as well as the family shall be visited in the near future by a conductor.

Sincerely,

signature (sgd.) of — *signature (sgd.) of*

Dr. Reginald E. Bittner, university professor — Dr. Wolfgang. M. Schmidt, associate professor

Pursuant to Section 69 of the Genetic Engineering Act, the interpretation of this finding or the results of the genetic analysis shall be performed by a specialist trained in human/medical genetics or by a specialist in the field of indication as part of a detailed consultation.

The use of these data for scientific or expert purposes requires the permission of the Neuromuscular Research Laboratory of the Center for Anatomy and Cell Biology of the Medical University of Vienna.