

University of Groningen

Myoclonus

Zutt, Rodi

IMPORTANT NOTE: You are advised to consult the publisher's version (publisher's PDF) if you wish to cite from it. Please check the document version below.

Document Version

Publisher's PDF, also known as Version of record

Publication date:

2018

[Link to publication in University of Groningen/UMCG research database](#)

Citation for published version (APA):

Zutt, R. (2018). Myoclonus: A diagnostic challenge [Groningen]: Rijksuniversiteit Groningen

Copyright

Other than for strictly personal use, 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), unless the work is under an open content license (like Creative Commons).

Take-down policy

If you believe that this document breaches copyright please contact us providing details, and we will remove access to the work immediately and investigate your claim.

Downloaded from the University of Groningen/UMCG research database (Pure): <http://www.rug.nl/research/portal>. For technical reasons the number of authors shown on this cover page is limited to 10 maximum.

MYOCLONUS
A DIAGNOSTIC CHALLENGE

Rodi Zutt

ISBN: 978-94-034-0383-0

Printed by: Ipskamp printing

Publication of this thesis was financially supported by University of Groningen, Research School of Behavioural and Cognitive Neurosciences, HagaZiekenhuis van Den Haag, MedCaT BV, Chipsoft, ABN-AMRO, Merz Pharma Benelux BV en Ipsen Farmaceutica BV.



rijksuniversiteit
 groningen

Myoclonus

A diagnostic challenge

Proefschrift

ter verkrijging van de graad van doctor aan de
Rijksuniversiteit Groningen
op gezag van de
rector magnificus prof. dr. E. Sterken
en volgens het besluit van het College voor Promoties.

De openbare verdediging zal plaatsvinden op
woensdag 7 februari 2018 om 14:30 uur.

door

Rodi Zutt

geboren op 1 februari 1984
te Alkmaar

Promotor

Prof. dr. M.A.J. de Koning-Tijssen

Copromotores

Dr. T.J. de Koning

Dr. J.W. Elting

Beoordelingscommissie

Prof. dr. J.G. van Dijk

Prof. dr. D.S. Verbeek

Prof. dr. M. Vidailhet

Paranimfen

Marenka Smit

Kathryn J. Peall

Vivat, crescat, floreat

'Leef, groei en bloei'

In dierbare herinnering aan mijn vader
Gerard Zutt (1950-2011),
van wie ik veel heb geleerd en
die mij deze levensspreuk meegaf.

CONTENT

CHAPTER 1	INTRODUCTION AND AIMS	1
1.1	DEFINITION AND CLASSIFICATION	2
1.2	EPIDEMIOLOGY	2
1.3	CLINICAL PRESENTATION	2
1.4	MYOCLONUS ASSIGNED TO ITS ANATOMICAL CLASSIFICATION	4
1.5	DIFFERENTIAL DIAGNOSIS	12
1.6	TREATMENT	13
1.7	AIMS OF THE THESIS	16
1.8	REFERENCES	19
CHAPTER 2	A NOVEL DIAGNOSTIC APPROACH TO PATIENTS WITH MYOCLONUS	25
2.1	ABSTRACT	26
2.2	INTRODUCTION	27
2.3	CLINICAL APPROACH TO MYOCLONUS	28
2.4	FROM DIAGNOSIS TO TREATMENT	44
2.5	CONCLUSIONS	46
2.6	SUPPLEMENTARY APPENDIX 1	47
2.7	SUPPLEMENTARY TABLE 1 - COMPREHENSIVE OVERVIEW OF GENES ASSOCIATED WITH MYOCLONUS	50
2.8	REFERENCES	59
CHAPTER 2A	UNUSUAL COURSE OF LAFORA DISEASE	65
CHAPTER 3	DISTRIBUTION AND CO-EXISTENCE OF MYOCLONUS AND DYSTONIA AS CLINICAL PREDICTORS OF <i>SGCE</i> MUTATION STATUS: A PILOT STUDY	75
3.1	ABSTRACT	76
3.2	INTRODUCTION	77
3.3	METHODS	78
3.4	RESULTS	79
3.5	DISCUSSION	85
3.6	CONCLUSION	87
3.7	REFERENCES	88
CHAPTER 4	THE PRESENCE OF DEPRESSION AND ANXIETY DO NOT DISTINGUISH BETWEEN FUNCTIONAL MYOCLONIC JERKS AND CORTICAL MYOCLONUS	91
4.1	ABSTRACT	92
4.2	INTRODUCTION	93
4.3	METHODS	94
4.4	RESULTS	95
4.5	DISCUSSION	98
4.6	REFERENCES	100
CHAPTER 5	MYOCLONUS SUBTYPES IN TERTIARY REFERRAL CENTER CORTICAL MYOCLONUS AND FUNCTIONAL JERKS ARE COMMON	103
5.1	ABSTRACT	104

5.2	INTRODUCTION	105
5.3	METHODS	105
5.4	RESULTS	109
5.5	DISCUSSION	116
5.6	REFERENCES	119
CHAPTER 6	ELECTROPHYSIOLOGICAL TESTING AIDS DIAGNOSIS AND SUBTYPING OF MYOCLONUS	123
6.1	ABSTRACT	124
6.2	INTRODUCTION	125
6.3	METHODS	127
6.4	RESULTS	130
6.5	DISCUSSION	139
6.6	REFERENCES	141
CHAPTER 7	IMPROVING NEUROPHYSIOLOGICAL BIOMARKERS FOR FUNCTIONAL MYOCLONIC MOVEMENTS	145
7.1	ABSTRACT	146
7.2	INTRODUCTION	147
7.3	METHODS	148
7.4	RESULTS	150
7.5	DISCUSSION	158
7.6	REFERENCES	160
CHAPTER 8	DISCUSSION AND CONCLUDING REMARKS	163
8.1	DEVELOPMENT OF A NOVEL DIAGNOSTIC ALGORITHM FOR PATIENTS WITH MYOCLONUS	165
8.2	THE IMPORTANCE OF CLINICAL PHENOTYPING IN DIAGNOSIS AND CLASSIFICATION OF MYOCLONUS	165
8.3	THE ROLE OF ELECTROPHYSIOLOGICAL TESTING TO AID DIAGNOSIS AND SUB-CLASSIFICATION OF MYOCLONUS	166
8.4	THE CONTRIBUTION OF NOVEL ELECTROPHYSIOLOGICAL TECHNIQUES TO DIAGNOSTIC TESTING	168
8.5	FUTURE PERSPECTIVES	168
8.6	CONCLUSION	170
8.7	REFERENCES	172
CHAPTER 9	NEDERLANDSE SAMENVATTING	175
9.1	KLINISCHE DIAGNOSTIEK MYOCLONUS	177
9.2	NEUROFYSIOLOGISCHE DIAGNOSTIEK MYOCLONUS	179
9.3	TOEKOMSTPERSPECTIEVEN	180
CHAPTER 10	DANKWOORD ACKNOWLEDGEMENTS	183
CHAPTER 11	CURRICULUM VITAE	189

