

Best Practice Meeting for the Molecular Diagnosis of FSHD
Leiden University Medical Center
Leiden, The Netherlands
June 9, 2010

08.30 - 08.50	<i>Registration and coffee</i>
08.50 - 09.00	Welcome S. van der Maarel
	Chair: P. Lunt
09.00 - 9.40	Session 1 Southern blotting based methods Definition of the mutation in FSHD and the molecular diagnosis by Southern blotting R. Lemmers
09.40 - 10.00	Non-radioactive probe labeling Southern blotting S. O'Shea
10.00 - 10.30	Prenatal diagnosis FSHD B. Bakker
10.30 - 11.00	<i>Coffee break</i>
	Session 2 Other methods
11.00 - 11.40	Molecular diagnosis by long range PCR Y. Hayashi
11.40 - 12.00	Molecular diagnosis by molecular combing N. Levy
12.00 - 13.00	<i>Lunch</i>
	Chair: S. van der Maarel
	Session 3 Experiences molecular testing
13.00 - 13.30	Large molecular diagnosis study in the UK M. Upadhyaya
13.00 - 14.00	Homogenization of genetic testing, the Italian experience R. Tupler
14.00 - 14.30	Summary questionnaire experience labs worldwide P. Lunt
14.30 - 15.00	Cost-Benefit Analysis (4qA/4qB or D4Z4 analysis) B. Bakker
15.00 - 15.30	<i>Coffee break</i>
	Chair: R. Frants
15.30 - 16.00	FSHD clinical uncertainties G. Padberg
16.00 - 16.30	General discussion
16.30 - 17.00	Consensus molecular diagnosis FSHD
17.00 - 17.30	Flowchart molecular diagnosis FSHD
17.30 - 18.00	Closing