Igenomix India Pvt. Ltd.
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PGD TEST REPORT

IGENOMIX ID:		Report Date:	
Patient name:		DOB:	**02/02/1989**
Partner name:		DOB:	**15/06/1988**
Clinic:			
Physician:		Date Received:	
Specimen Type:	**TROPHECTODERM**	DNA Code:	**3476**
Indication:	**NEMALINE MYOPATHY**	Female Mutation:	**c.24559C>T**
Gene:	**NEB**	Male Mutation:	**c.6712dupA**

RESULTS:

Embryo ID	c.24559C>T mutation At risk patient haplotype	c.6712dupA mutation At risk partner haplotype	Results
1	**Absent**	**Absent**	**Normal**
2	**Absent**	**Absent**	**Normal**
3	Present	Present	Abnormal
4	Present	Present	Abnormal
5	**Present**	**Absent**	**Normal carrier**

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METHODS:

Combined direct and indirect genetic study of the NEB gene by fluorescent PCR. Direct: PCR amplification of c.24559C>T mutation-containing gene region and minisequencing and c.6712dupA mutation-containing gene region; Indirect: PCR amplification of polymorphic markers linked to NEB gene (D2S2277, D2S2275, D2S2236 and D2S2299). Fragment analysis of PCR products by capillary electrophoresis (CE) through AB 3130 (ThermoFisher).

NOTES:

The accuracy of this PGD exceeds 98%. In case of pregnancy, prenatal control or diagnosis is recommended to confirm the genetic diagnosis and to detect the presence of potential chromosomal abnormalities that may not analyzed by this test. The presence of allele drop out and/or maternal/external contamination cannot be fully ruled out in this analysis. Chromosomal abnormalities have not been directly analyzed in this diagnosis.

Approved by

Sample name, PhD	**Sample name, PhD**
Biologist	Laboratory Manager

This test was performed by IGENOMIX S.L. in its laboratory located in Parque Tecnológico de Paterna, Calle Narcís Monturiol Estarrio, nº 11 Parcela B, Building Europark, 46980 Paterna (Valencia), Spain.