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Letter to the Editor

Obstetric risk in patients with myopathy due to *MATR3* mutations

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ABSTRACT

Background: The obstetric risk associated with myopathy due to *MATR3* mutations is unknown.
Methods: Eight women with the *MATR3* p.S85C mutation were recruited. Information on pregnancy, outcome, and effect on muscular function was analysed retrospectively using a pregnancy and delivery questionnaire. The data were compared with information from the German perinatal quality survey.
Results: All eight women responded. Their muscular symptoms started between the ages of 36 and 56. Sixteen pregnancies and twelve deliveries could be analysed. Two women had a voluntary abortion after their deliveries for other medical reasons. One woman reported a miscarriage in the first trimester. Five women had pregnancies and deliveries without complications. One woman twice had labour weakness requiring forceps delivery. Another patient twice had a preterm dilatation of the cervical os and forceps deliveries. One of her children had foetal distress and was born preterm and with low birth weight. No perinatal childhood death was reported. No women described muscular symptoms before or during their pregnancies.
Conclusions: Pregnancies in matrin 3 myopathy typically occur several years before the onset of myopathy. No increase in the incidence of foetal distress or miscarriage was found. However, late pregnancies (e.g. in the 5th decade) should be regarded as pregnancies at risk.

Dear Sirs,

Myopathy due to mutations in *MATR3*, which encodes the nuclear matrix protein matrin 3, is a newly described rare myopathy with late onset (usually after the fourth decade) and predominantly distal paresis of legs and arms (Welander type), sometimes with the phenotype of a motor neuron disorder [1,3,4]. No information is currently available to guide obstetricians and neurologists in counselling women with matrin 3 myopathy about becoming pregnant and giving birth. We used a pregnancy and delivery questionnaire to retrospectively collect data about pregnancy and birth outcomes from 8 patients with matrin 3 myopathy due to the *MATR3* p.S85C mutation diagnosed at the Department of Neurology at Martin-Luther-University Halle-Wittenberg, Germany [2]. An informed consent form was signed by all participants. The study was approved by the Local Ethics Committee of the Martin-Luther-University Halle (Saale).

Sixteen pregnancies and twelve deliveries could be analysed (Table 1). None of the women had a chronic disorder at the time of their pregnancies. Two women had chosen a voluntary abortion (P1 at age 25, P4 at age 26) for medical reasons (i.e. due to the complications in the previous pregnancies and deliveries). In both cases the voluntary abortion was of the 3rd pregnancy. P2 had a voluntary abortion at age 29 for private reasons (i.e. reasons other than medical ones).

The majority of the women became pregnant two or even three decades prior to the occurrence of functionally disabling symptoms which can be attributed to the myopathy. In all but one woman the myopathy-related symptoms occurred in the 5th decade, i.e. at the end of the reproductive age. None of the women reported an abnormal weakness during the pregnancies, but one (P1) reported labour weakness during both her deliveries and some difficulties in the recovery after the delivery. In other words, all women were asymptomatic for myopathy at the time of their pregnancies. However, the effect of late-onset symptoms on pregnancy and delivery could become increasingly relevant because the mean age at first pregnancy in woman in Germany has been rising over the last decades. In 2010 the mean age during the 3rd pregnancy was 32.4 years in 2010 [5]. Limitations of this study include the small number of patients and a possible recall bias. The data are nevertheless important given the paucity of information currently available.

Informed consent

All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the Helsinki Declaration of 1975, as revised in 2000 [5]. Informed consent was obtained from all patients for being included in the study.

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Table 1
Demographic data and data regarding pregnancies and deliveries in 8 woman with *MATR3* p.S85C mutation.

Patient	Age (years)	Age at onset (years)	Age at pregnancy (years)	Pregnancy duration (weeks), gender, birth weight (g)	Complications during pregnancy and delivery	Miscarriage
1.	69	47	19	37, M, 2850	Forceps delivery	No
			23	37, F, 2700	Forceps delivery	
2.	46	36	25	36, M, 3020	No	No
3.	50	47	20	40, F, 3800	No	No
			25	39, F, 2800	No	
4.	56	53	21	31, M, 1700	Preterm labour, preterm dilatation of the cervical os, forceps delivery, hypoxia of the newborn	No
			23	38, M, 3850	Preterm dilatation of the cervical os and cervical cerclage at week 28, forceps delivery	
5.	72	42	22	39, F, 3500	No	1st trimester
			30	40, F, 4000	No	At 26 years
6.	60	56	19	40, M, 3000	No	No
7.	55	52	21	40, M, 2930	No	No
8.	74	52	20	40, F, 3500	No	No

Disclosure

F.H. received lecturer honoraria and travel fees from Biomarin, Genzyme, and Astellas Inc. J.B. and T.M. declare that they have no conflict of interest.

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