**Supplementary table 1. Complications during pregnancy in pregnancies among GNE myopathy patients including pre-onset pregnancies**

|  |  |  |  |
| --- | --- | --- | --- |
| Complications | General population  (Japanese) | Pre- and post-onset pregnancies  (n=771)) | P value |
|  | **%** | **% [95% CI]** | **p** |
| Hyperemesis gravidarum |  | 84.4 [74.6-91.7] |  |
| Threatened abortion | 11.9[18] | 19.5 [11.3-30.1] | 0.05 |
| Threatened premature delivery | 19.2[18] | 10.4 [4.6-19.4] | 0.06 |
| Hypertensive disorder of pregnancy | 3.3[18] | 1.3 [0.03-7.0] | 0.73 |
| Preeclampsia |  | 1.3 [0.03-7.0] |  |
| Gestational diabetes mellitus | 2.7[18] | 1.3 [0.03-7.0] | 0.73 |
| Abruptio placentae | 0.5 [19] | 0.0 [0.0-4.7] | 1.0 |
| Placenta previa | 0.7[19] | 1.3 [0.03-7.0] | 0.37 |
| Blood type incompatibility |  | 0.0 [0.0-4.7] |  |
| Others |  | 2.6 [0.3-9.1] |  |
|  | (edema, prolapsed uterus) |  |

1. Analyzed 77/81 pregnancies due to missing data (no response).

**Supplementary table 2. Outcomes of pregnancy in pregnancies among GNE myopathy patients including pre-onset pregnancies**

|  |  |  |  |
| --- | --- | --- | --- |
| Outcomes | General population  (Japanese) | Pre- and post-onset pregnancies  (n=791)) | P value |
|  | **%** | **% [95% CI]** | **p** |
| Early miscarriage | 16-18[20]2) | 8.9 [3.6-17.4] |  |
| Late miscarriage | 0.0 [0.0-4.6] |
| Abortion |  | 1.3 [0.03-6.9] |  |
| Stillbirth | 0.3[19] | 0.0 [0.0-4.6] | 1.0 |
| Delivery |  | 89.9 [81.0-95.5] |  |
| Preterm3) | 6.9[19] | 2.5 [0.3-8.8] | 0.24 |
| Term3) | 92.9[19] | 95.8 [88.1-99.1] | 0.49 |
| Post-term3) | 0.2[19] | 1.3 [0.03-6.9] | 0.14 |

1. Analyzed 79/81 pregnancies due to missing data (no response).
2. Percentage of miscarriages, including both early and late miscarriages at maternal age up to 31 years old. Prevalence will increase with age.
3. Percentage among deliveries including pre-onset deliveries.

**Supplementary table 3. Complications during delivery in deliveries among GNE myopathy patients including pre-onset deliveries**

|  |  |  |  |
| --- | --- | --- | --- |
|  | General population  (Japanese) | Pre- and post-onset deliveries  (n=71) | P value |
|  | **%** | **% [95% CI]** | **p** |
| Type of delivery | | | |
| Vaginal delivery | 81.9[18] | 90.1 [80.7-95.9] | 0.088 |
| Cesarean section (CS) | 18.1[18] | 8.5 [3.2-17.5] | 0.016\* |
| Emergency CS | 13.71) | 2.8 [0.3-9.8] | 0.004\* |
| (fetal distress, prolonged labor) | |
| Previous CS | 8.1[18] | 2.8 [0.3-9.8] | 0.12 |
| Breech presentation | 2.4[21] | 4.2 [0.9-11.9] | 0.25 |
| Beginning of vaginal delivery2) | | | |
| Spontaneous labor |  | 69.5 [56.1-80.8] |  |
| PROM | 10-20[22]3) | 11.9 [4.9-22.9] |  |
| Bleeding |  | 3.4 [0.4-11.7] |  |
| Induced labor |  | 15.3 [7.2-27.0] |  |
| Fetal abnormality |  | 1.7 [0.04-9.1] |  |
| Maternal abnormality |  | 3.4 [0.4-11.7] |  |
| Overdue pregnancy |  | 6.8 [1.9-16.5] |  |
| Scheduled |  | 3.4 [0.4-11.7] |  |
| Outcomes of vaginal delivery4)5) | | | |
| No assistance |  | 78.1 [66.0-87.5] |  |
| Vacuum extraction | 7.6[18] | 12.5 [5.6-23.2] | 0.13 |
| Manual fundal pressure | 11.2[23] | 12.5 [5.6-23.2] | 0.69 |
| Forceps delivery | 0.3[18] | 0.0 [0.0-5.6] | 1 |
| Management | | | |
| Labor augmentation6) |  | 30.5 [19.2-43.9] |  |
| Blood transfusion | 0.5[17] | 1.4 [0.04-7.6] | 1 |
| Number of fetuses | | | |
| Singletons | 98.1[17] | 100.0 [95.1-100.0] | 0.41 |
| Complications after delivery | | | |
| Maternity blues |  | 2.7 [0.3-9.4] |  |
| Postnatal depression |  | 0.0 [0.0-4.9] |  |
| Deep vein thrombosis |  | 0.0 [0.0-4.9] |  |
| Intrauterine infection |  | 1.4 [0.0-7.3] |  |
| Mastitis |  | 8.1 [3.0-16.8] |  |
| Prolapsed uterus |  | 0.0 [0.0-4.9] |  |

PROM: Premature rupture of the membrane.

1) Analyzed 3109 deliveries of Tokyo Medical and Dental University from 2013-2019.

2) Analyzed 59/71 vaginal deliveries due to missing data (no response).

3) Frequency among vaginal deliveries and CS.

4) Analyzed 64/71 vaginal deliveries due to missing data (no response).

5) Multiple choices allowed.

6) Analyzed 59/71 deliveries due to missing data (no response).

**\*p<0.05**

**Supplementary table 4. Newborn outcomes in deliveries among GNE myopathy patients including pre-onset deliveries**

|  |  |  |  |
| --- | --- | --- | --- |
|  | General population  (Japanese) | Pre- and post-onset deliveries  (n=71) | |
| Characteristics at birth | | | |
|  | **Average** | **Average [SD]** | **Median [Min-Max]** |
| Body weight [kg]1) | 2987 [447][18] | 3003.6 [344.6] | 2937 [2218-3800] |
| Height [cm]2) | B: 49.0, G: 48.5[26] | 49.2 [1.7] | 49.0 [45.5-53.0] |
|  | **%** | **% [95% CI]** | **p** |
| Low birth weight (<2500 g)1) | 9.1[18] | 2.9 [0.4-10.2] | 0.09 |
| Complications3)4) | | | |
|  | **%** | **% [95% CI]** | |
| No complications |  | 75.4 [63.5-84.9] | |
| Prolonged jaundice |  | 4.4 [0.9-12.2] | |
| Intracranial hemorrhage |  | 2.9 [0.4-10.1] | |
| Vomiting |  | 1.4 [0.04-7.8] | |
| Others |  | 1.4 [0.04-7.8]  (hypospadias, ptosis) | |

B: boy, G: girl

1) Analyzed 68/71 newborns due to missing data (no response).

2) Analyzed 65/71 newborns due to missing data (no response).

3) Analyzed 69/71 newborns due to missing data (no response).

4) Multiple choices allowed.