**Analysis of Birth Data**

**Student Name**

**Advisor Name**

## Introduction

Several things can be attributable to congenital anomalies. Some are errors in the infant's physical structure. Others could be genetic, but many congenital anomalies have unknown causes. Other studies suggest that there is an association between maternal age and congenital anomalies [1,2,3]. The purpose of this project is to further study potential associations between certain characteristics (in particular maternal age) and the occurrence of congenital anomalies. This study uses a large data set to examine the relationship (after controlling for confounding variables) between maternal age and the presence of each of the following congenital anomalies: Down syndrome, Omphalocele/Gastroschisis, Polydactyly/Syndactyly/Adactyly, and musculoskeletal anomalies (not including club foot, diaphragmatic hernia, and cleft lip/palate). Down syndrome (also known as trisomy 21) is where the 21$^{st}$ chromosome is copied and causes abnormalities in the development of the infant. Omphalocele/Gastrosis are congenital anomalies in which the infant is born with a hole in their abdominal wall. Polydactyly, Syndactyly, and Adactyly are extra fingers (or toes), fingers fused together, and missing fingers, respectively.

## Study Population

Data for this study was obtained from the Howard W. Odum Institute for Research in Social Science. The data were collected by the North Carolina State Center for Health Statistics and consisted of all births in North Carolina recorded in 2009. The dataset contained 129,444 observations and 125 unique variables measuring characteristics of both the mother and the infant. For this study, an observation was excluded from the analysis if it was missing information on either maternal age or occurrence of any of the following: Down syndrome, Omphalocele/Gastroschisis, Polydactyly/Syndactyly/Adactyly, or other musculoskeletal anomalies. In total, 126,251 observations were used in the analysis. Table 1 shows how the characteristics in the models are distributed by age. The table splits the subjects into three maternal age groups: 11-19, 20-29, and 30 years or greater.

**Table1:** Demographics (Characteristics of Subjects)

| Characteristics | Maternal age (years old) | | |
|---|---|---|---|
| | 11-19 | 20-29 | 30+ |
| No. of live birth | 14,020 | 67,916 | 44,315 |
| Maternal race (%) | | | |
| *White* | 58.84 | 69.73 | 77.30 |
| *Black* | 37.19 | 25.83 | 16.71 |
| *Other than black or white* | 3.97 | 4.44 | 5.99 |
| Unmarried (%) | 85.77 | 47.14 | 19.93 |
| Tobacco use (%) | 11.73 | 12.38 | 6.06 |
| Alcohol use (%) | 0.21 | 0.33 | 0.47 |
| Kotelchuck index (%) | | | |
| *Inadequate* | 16.50 | 9.64 | 5.35 |
| *Intermediate* | 8.99 | 8.33 | 6.13 |
| *Adequate* | 36.96 | 39.86 | 40.53 |
| *Adequate Plus* | 37.55 | 42.17 | 47.99 |
| Male infant (%) | 51.35 | 50.97 | 51.57 |

A majority of live births came from the 20-29 age group. As age increases, the proportion of white mothers increases. The proportion of married mothers increases as maternal age group increased. Kotelchuck index ratings improve as maternal age increases. Proportions for tobacco and alcohol use were highest for the 20-29 age group.

**Table 2:** Rates of congenital anomalies

| | Maternal Age (years old) | | |
|---|---|---|---|
| Rate per 10,000 live births | 11-19 | 20-29 | 30+ |
| Down Syndrome | 1.43 | 0.04 | 7.45 |
| Omphalocele/Gastrosis | 9.99 | 0.04 | 0.68 |
| Polydactyly/Syndactyly/Adactyly | 16.41 | 0.11 | 9.25 |
| Other musculoskeletal anomalies | 32.1 | 20.91 | 6.32 |

**Statistical Analysis**

SAS Version 9.2 [6] was used to fit logistic regression models in order to estimate adjusted odds ratios (and associated 95% confidence intervals) for the presence of congenital anomalies associated with maternal age. Logistic regression is used to predict outcomes for a categorical variable. In this case we have a binomial response, because an infant either has a congenital anomaly or it does not. Maternal age is a continuous variable, and can be used to

estimate the likelihood of the infant having a congenital anomaly. Logistic regression can also use categorical predictor variables if desired. A separate model was fit for each of the following congenital anomalies: Down syndrome, Omphalocele/ Gastroschisis, Polydactyly/Syndactyly/ Adactyly, and other musculoskeletal anomalies. All of the models included covariates to adjust for maternal race, marital status, tobacco and alcohol use, the Kotelchuck index, and the infant's gender. The Kotelchuck index is a rating based on how early and how frequently the mother received prenatal care during her pregnancy.

**Results**

**Table 3:** Odds ratios and 95% confidence intervals of congenital anomalies in association with maternal age

| Anomalies | Maternal age (years old) |
|---|---|
| Down syndrome | 1.15* (1.1, 1.21) |
| Omphalocele/Gastrosis[a] | 0.87* (0.81,0.92) |
| Polydactyly/Syndactyly/Adactyly[b] | 0.99 (0.96, 1.02) |
| Other musculoskeletal anomalies[c] | 0.95* (0.93, 0.98) |

* Maternal age is significant at the 0.05 level

Significance of Covariates:

[a] Kotelchuck index significant at the 0.05 level, smoking and drinking at 0.10 level

[b] Race significant at the 0.05 level

[c] Race, marital status, smoking, and Kotelchuck index significant at the 0.05 level

These results show that there is a significant association between maternal age and certain congenital anomalies. After adjusting for confounding factors, a one-year increase in maternal age is associated with a 15% increase in the odds of Down syndrome, a 13% decrease in the odds of Omphalocele/Gastroschisis, and a 5% decrease in the odds of other musculoskeletal anomalies.

With Polydactyly/Syndactyly/Adactyly, maternal age was significant at the 0.05 level when confounding factors are not included in the model, but it was non-significant in the full model. Race was the only factor that was significant at a 0.05 level for the

Polydactyly/Syndactyly/ Adactyly model. Kotelchuck index was significant for Omphalocele/Gastrosis at the 0.05 level.

**Discussion**

This study supports the results of other work and provides significant evidence of an association between maternal age and the occurrence of various congenital anomalies. While the odds of an infant having Down syndrome appear to increase with advanced maternal age, the odds of an infant being affected by Omphalocele/Gastroschisis or Polydactyly/Syndactyly/ Adactyly actually decrease with maternal age and are highest with teen pregnancies. It's possible that the different congenital anomalies have an opposite relationship with maternal age due to whether the anomaly is attributed to a chromosomal error or not. The increased risk of other congenital anomalies for teenage mothers could occur due to malnourishment. Other research suggests that Down syndrome is less likely at a younger maternal age because of the fertility of the egg.

It should be noted that maternal age was a significant predictor for Polydactyly/ Syndactyly/Adactyly when there were no other variables in the model (the odds of an infant having this anomaly were much higher for young mothers); however, after adjusting for the confounding variables, the effect of maternal age was no longer significant.

In the model for predicting other musculoskeletal anomalies, maternal age, smoking, marital status, maternal race, and the Kotelchuck index were all significant covariates. Figure 1 (below) illustrates how maternal age and these covariates affect the predicted probability of having musculoskeletal anomalies. In Figure 2, the predicted probability curve increases in a J-shape as maternal age increases. This shape is similar to other researchers' findings [3]. Confounding factors were not significant in changing the likelihood of Down syndrome, and therefore the predicted probabilities of the subjects with different demographics were similar.

The shape in Figure 3 is similar to that in Figure 2, but instead of the likelihood greatly increasing as maternal age increases, likelihood decreases greatly. This result agrees with other studies [1]. Predicted probabilities and confidence intervals are nearly identical between demographics. Figure 4 shows the likelihoods are nearly horizontal, showing maternal age doesn't have a significant effect on Polydactyly/Syndactyly/Adactyly. Race was the only significant factor in the model.

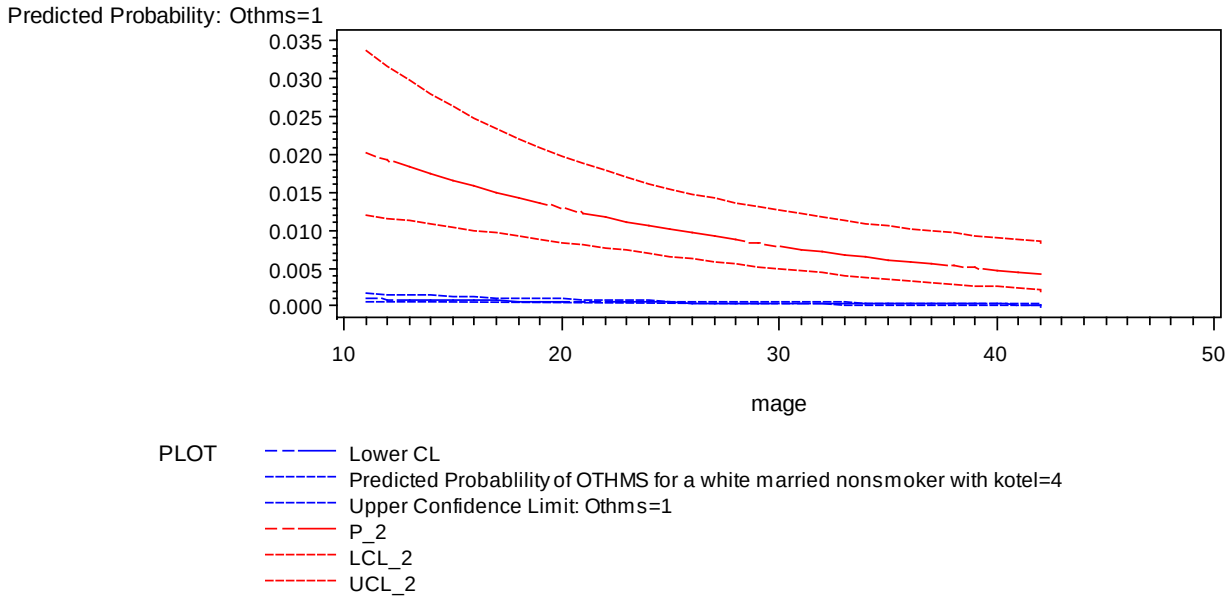


**Figure 1:** Predicted probabilities (with 95% confidence intervals) of a musculoskeletal anomaly for a baby born to a white, married, non-smoker with a Kotelchuck index rating of adequate plus (shown in blue) vs. a baby born to a black, single, smoker with a Kotelchuck index rating of inadequate (shown in red).

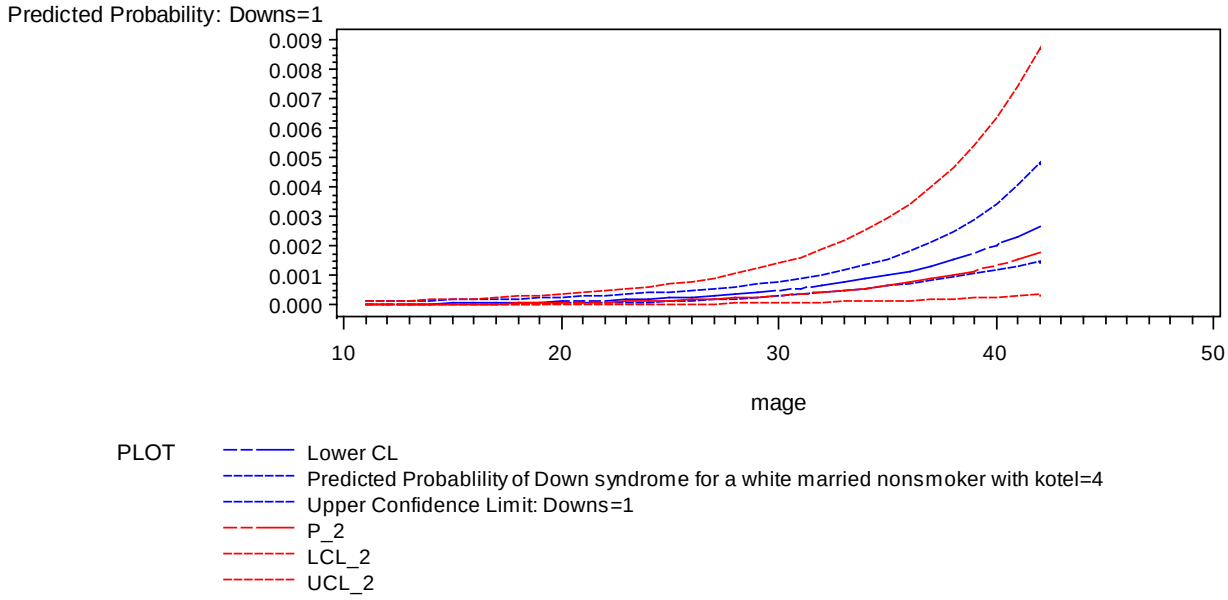


**Figure 2:** Predicted probabilities (with 95% confidence intervals) of Down syndrome.

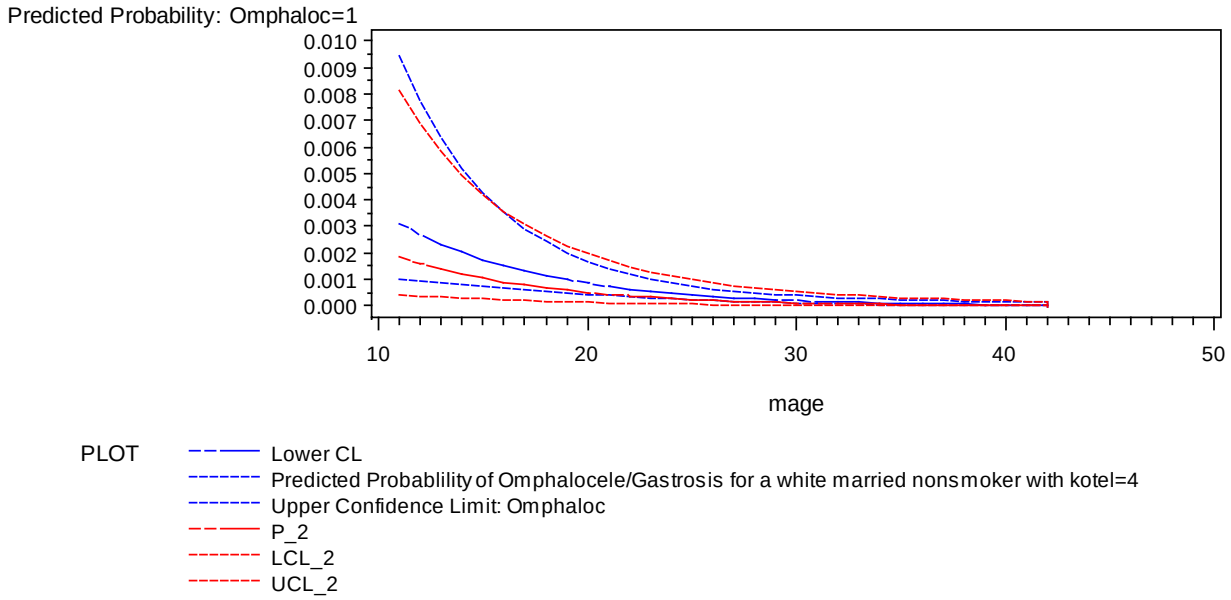


**Figure 3:** Predicted probabilities (with 95% confidence intervals) of Omphalocele/Gastrosis.

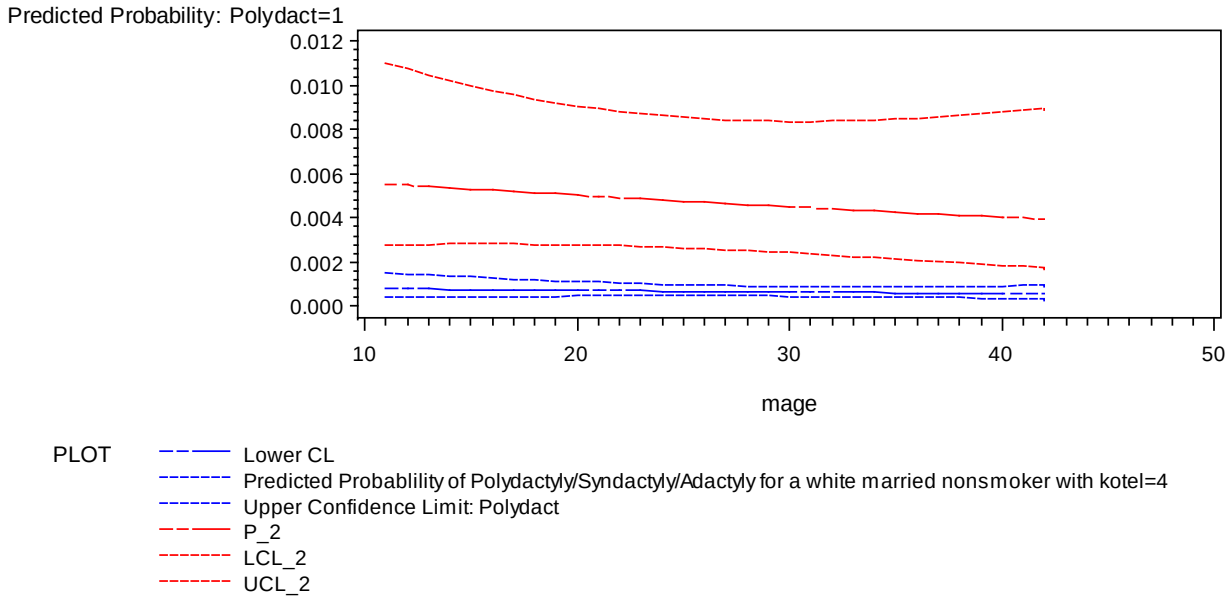


**Figure 4:** Predicted probabilities (with 95% confidence intervals) of Polydactyly/Syndactyly /Adactyly. Race is significant at the 0.05 level.

**Refrences**


1. Chen, Xi-Kuan, “Teenage pregnancy and congenital anomalies: which system is vulnerable?”, Oxford University, 2007.
2. Ledbetter, Daniel J., M.D., “Gastroschisis and Omphalocele”, Elsevier Inc., 2006.
3. Newberger, David S., M.D., “Down Syndrome: Prenatal Risk Assessment and Diagnosis”, State University of New York, 2000
4. North Carolina State Center for Health Statistics (NCSCS)
5. Howard W. Odum Institute for Research in Social Science

   Website: http://arc.irss.unc.edu/dvn/dv/NCVITAL
6. SAS, Version 9.2 SAS Institute Inc., Cary, NC, 1989-2007.