

# Prenatal, Birth, and Neonatal Outcomes in Sex Chromosome Aneuploidies: **Data from the INSIGHTS and GALAXY Registries**

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## Background

- Sex chromosome aneuploidies (SCAs) are a group of genetic conditions caused by additional or missing sex chromosome(s) and are associated with complex medical manifestations.
- Recent guidelines recommending prenatal cell-free DNA screening have increased the volume of early SCA diagnoses, thus increasing the need to understand perinatal health outcomes.
- A comprehensive evaluation of perinatal history in SCAs has not been conducted since birth cohort studies in the 1960s-70s.
- This study aims to investigate perinatal outcomes associated with SCAs during the prenatal, birth, and neonatal periods.

## **Methods**

- A retrospective cohort study was conducted using data from the Inspiring Science in Guiding Healthcare in Turner Syndrome (INSIGHTS) and Generating Advancements with Longitudinal Analysis in Xand Y variations (GALAXY) registries.
- Data included in this analysis were prenatal health (i.e., maternal and fetal complications), birth characteristics (i.e.mode of delivery, length and weight at birth), and neonatal health history, defined as outcomes in the first 30 days of life.
- The cohort included Turner syndrome (45,X), Klinefelter syndrome (47,XXY), Triple X syndrome(47,XXX), XYY syndrome (47,XYY), and XXYY syndrome (48,XXYY).
- Data were analyzed using descriptive statistics and one-way proportion tests to compare our sample with the US population.
- US population prevalences were determined via either Centers for Disease Control and Prevention (CDC) or National Institutions for Health (NIH) reports.



# pregnancies.

### Feeding difficulties (NG tube, etc.) Cardiac concerns (bradycardia arrhythmia structura Significant respiratory difficulties Hyperbilirubinemia (iaundice) requiring treatment Hypoglycemia (low blood sugar) requiring trea Sepsis or other systemic infection (need for antibiotics) ntubation and ventilation Meconium aspiration Renal abnormalities (diagnosed after birth) Respiratory distress syndrome (RDS/surfactant treatment) Anemia (requiring intervention) Intraventricular hemorrhage (IVH)

births.

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Figure 1. Maternal complications in SCA



Figure 3. Neonatal complications in SCA

Figure 2. Fetal complications in SCA pregnancies.

| Effect                                     | Population<br>Prevalence | Sample<br>Prevalence | 95% CI |       |
|--|--------------------------|----------------------|--------|-------|
|  |                          |                      | LL     | UL    |
| Cardiac<br>concerns                        | 2.4%                     | 26.8%                | 22.7%  | 30.9% |
| C-section birth                            | 32.1%                    | 38.3%                | 33.9%  | 42.7% |
| Hypoglycemia                               | 7.5%                     | 14.1%                | 10.8%  | 17.3% |
| NICU stay                                  | 15.0%                    | 36.8%                | 32.4%  | 41.3% |
| Sepsis and/or<br>systemic<br>infections    | 0.01%                    | 5.8%                 | 3.6%   | 8.0%  |
| Significant<br>respiratory<br>difficulties | 7.0%                     | 18.3%                | 14.7%  | 21.9% |
| Feeding<br>difficulties                    | 26.9%                    | 28.4%                | 24.2%  | 32.5% |

 
 Table 1.
 Prevalence of health concerns in SCA
neonates versus the general US population.





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# Conclusions

- The sample consisted of 667 participants, with similar racial (77% white) and insurance (61% private) statistics as the general US population.
- The most common maternal complications were preterm labor (10%) and hypertension/preeclampsia (9%) (Figure 1).
- The most common fetal findings were poor intrauterine growth (15%) and congenital heart malformation (13%) (Figure 2).
- Average gestational age at delivery was 38 weeks, with an average birthweight of  $3.0 \pm 0.6$  kg and average birth length of  $47.3 \pm 5.0$  cm.
- During the neonatal period, the most common health concerns were feeding difficulties (28%), cardiac concerns (27%), and significant respiratory difficulties (18%) (Figure 3).
- C-section, NICU admission, cardiac concerns, hypoglycemia, sepsis/systemic infection, and respiratory difficulties were more prevalent in our sample than the general US population (Table 1).

# Implications

- These findings represent the first descriptive update to perinatal health in SCAs in over 40 years.
- While many SCA pregnancies and neonates are healthy, these data suggest the presence of increased perinatal risks that may inform clinical recommendations and guide future research.

# Disclosures

The authors declare that they have no relevant or material financial interests that relate to the research described in this poster.