



**The eXtraordinary Babies Study:  
Researching the Natural History of Health and Neurodevelopment in Infants and  
Young Children with Sex Chromosome Trisomy**

**Purpose of the study:**

This study is designed to research the natural history of neurodevelopment, health and early hormonal function in infants with XXY/Klinefelter syndrome, XYY, XXX and other sex chromosome variations in an effort to identify early predictors of developmental and health outcomes. We will also evaluate different developmental screening tools in infants with sex chromosome variations so we can develop recommendations for pediatrician caring for infants and young children with XXY/Klinefelter syndrome, XYY, XXX, and other sex chromosome variations.

**What's Involved:**

Children age 6 weeks to 12 months old, who have a prenatal diagnosis of XXY, XYY, XXX, XXYY, and other sex chromosome variations are eligible.

- 4-8 study visits over 2-4 years, depending on age and timing of enrollment

Assessments will include:

- Measures of skills in different areas of development including problem solving, language, social, motor, and life skills.
- Physical exam
- Blood, urine, and stool testing for hormone levels, metabolic functioning and DNA banking.
- Scans for body composition (muscle and fat mass, bone density)
- Quality of life outcomes
- Questionnaires & surveys to be completed by parents or caregivers

You will be paid for each visit in this study, dependent upon how many visits are completed. You will help us gain valuable information about sex chromosome variations. The results of this study will help plan for future studies in sex chromosome aneuploidies

**To learn more about this study, contact Karen Kowal at 215-955-9008 or email:**

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Study sites include Colorado, Delaware or Philadelphia. Travel budget included if needed.

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For questions regarding the rights of research patients call the Nemours Office of Human Subjects Protection:  
302-298-7613 or 800-767-5437.

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