eXtraordinary Kids
Research in Colorado

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Developmental-Behavioral Pediatrics
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XXY/Klinefelter syndrome
Trisomy X
XYY
XXXXY
Tetrasomy X
Pentasomy X
Turner syndrome
Common Themes

Families felt that medical professionals, therapists, and educators usually knew very little about their child’s genetic condition (except some endocrinologists)

Most had experienced a “diagnostic odyssey” prior to the sex chromosome variation diagnosis

Many problems and needs crossed the expertise of multiple disciplines (medical/health, psychology, speech therapy, social work, education, etc.)

There was a marked spectrum of involvement

There was a significant need for clinical research and updated publications to establish care guidelines and support families in getting evaluations and services
### eXtraordinary Kids Clinic: Interdisciplinary Team

**Goal:** To provide comprehensive interdisciplinary evaluations and treatment recommendations for children and adolescents with X and Y chromosome variations

#### Developmental-behavioral pediatrics
- **Clinic director**
- **Team leader**
- **Evaluation of:**
  - Physical features
  - Medical problems associated with SCA
  - Psychopharmacologic medication recommendations and management

#### Child psychology
- **Evaluation of:**
  - Behavioral and emotional functioning
  - Cognitive functioning
  - Social skills/autism spectrum disorders
  - Adaptive functioning
  - Home and school behavioral strategies

#### Pediatric neuropsychology
- **Evaluation of:**
  - Cognitive functioning
  - Learning disabilities
  - ADHD
  - Executive functioning
  - Visual-spatial skills
  - Memory functioning
  - Educational recommendations

#### Speech-language therapy
- **Evaluation of:**
  - Understanding of language
  - Verbal expression
  - Speech production including apraxia
  - Recommendations for therapy and school/home strategies if needed

#### Occupational therapy
- **Evaluation of:**
  - Fine and gross motor skill
  - Adaptive/daily living skills
  - Sensory processing differences
  - Recommendations for therapy and school/home strategies if needed

#### Pediatric endocrinology
- **Evaluation/treatment of:**
  - Hypogonadism (Testosterone deficiency) in XXX, XXXY, XXXY, and XXXXY
  - Endocrine risks (including type II diabetes, thyroid disorders)
  - Overweight/obesity

#### Nursing
- **Communication and coordination with families**
- **Support for medication management and refills**
- **Social work**
- **Identification of community and government resources**
- **Assistance with transition planning**
- **Involvement in complex cases**

#### Genetic counseling program:
- **Clinic coordination (intake and scheduling)**
- **Genetic counseling for cases ascertained through:**
  - Non-invasive prenatal testing (NPT)
  - Prenatal CVS and amniocentesis
  - Postnatal diagnosis
  - Family support and strategies for thoughtful disclosure of diagnosis to affected child/adolescent
  - Family planning for affected adults

#### Infant developmental evaluations:
- **Developmental monitoring and evaluation for infants diagnosed in the prenatal or newborn period.**
- **Evaluation includes:**
  - Developmental-behavioral pediatrics
  - Occupational therapy
  - Speech therapy
  - Physical therapy

#### Psychopharmacological medication management:
- **By DBPeds for treatment of:**
  - ADHD, anxiety, other behavioral and mood disorders
  - Nursing support

#### Testosterone therapy management:
- **By endocrinology**
- **Nursing support**
- **Joint appointment with DBPeds if needed**

#### Feedback session with family including discussion of concerns, team impressions, and recommendations, comprehensive written report

#### Research
- Neurodevelopmental phenotypes
- DNA repository
- Genotype-phenotype studies
- Clinical trails

#### Education
- Continuing education presentations for professionals
- Trainees (residents, graduate students and fellows)
- Development of professional and training materials

#### Support and advocacy
- Family-centered events and conferences
- Development of family support materials
- Collaboration with national advocacy organizations

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*Children's Hospital Colorado*
WHAT WILL HAPPEN TO MY BABY?
Study Aims

To describe the profiles of neurodevelopmental, medical, and hormonal features during the first 3-4 years of life for infants with XXY/Klinefelter syndrome, XYY, Triple X, XXYY, and others.

To identify risk factors and early predictors of poor neurodevelopmental and health outcomes in X&Y chromosome disorders.

To identify the best set of tools for primary care doctors to use to screen neurodevelopment in infants prenatally diagnosed with X&Y chromosome disorders.

Build a biobank of DNA, RNA, and other biological samples for future studies.
Participants/Visits:

- Prenatal Diagnosis of XXY, XYY, XXX, XXYY
- Enroll at 12 months or younger
- Visits at 2m, 6m, 12m, 18m, 2y, 3y, 4y, 5y

2 sites:

- Univ of Colorado / Denver
- Nemours-Dupont in Delaware (Judy Ross, MD; Karen Kowal)
**Study Procedures**

Data collected:
- Demographics
- Medical / Health history
- Family history
- Previous / Current interventions

Developmental Assessments:
- Cognitive, Language
- Social
- Motor
- Adaptive functioning

Parent Questionnaires (Quality of Life, Attachment)

Parent Cognitive / Language / Executive functioning

Physical examination

PEDPod Body Composition

Blood, Urine, and Stool samples:
- Hormone levels
- DNA / RNA
- Other metabolic measures

Biobank
BIOBANK FOR FUTURE STUDIES

BIOBANK = “Bank” of biological samples to be used for future research

When study complete – detailed database of medical and developmental assessments

Speeds research and decreases costs of future research questions
# eXtraordinary Babies Study Enrollment

(2 sites)

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<th></th>
<th>Enrolled</th>
<th>Goal</th>
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<tbody>
<tr>
<td>XXY</td>
<td>77</td>
<td>120 (64%)</td>
</tr>
<tr>
<td>XYY</td>
<td>7</td>
<td>40 (18%)</td>
</tr>
<tr>
<td>XXX</td>
<td>15</td>
<td>40 (38%)</td>
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<tr>
<td>XYY</td>
<td>2</td>
<td>--</td>
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<tr>
<td><strong>TOTAL</strong></td>
<td><strong>101</strong></td>
<td><strong>200 (51%)</strong></td>
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Come to our poster for more details and preliminary results!
Participation Includes:
Travel to the closest site (Denver or Philadelphia/Delaware) including hotel and airfare

Feedback report to share with providers at home
Acknowledgements

eXtraordinarY Kids Clinic & Research Team
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AXYS
Facebook

THANK YOU ALL FAMILIES AND EXTRAORDINARY BABIES!!