# eXtraordinarY Kids
# Research in Colorado

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Children's Hospital Colorado

Medicine

University of Colorado 1876

XXY/Klinefelter syndrome
Trisomy X
XYY
XXXY
XXXXY
Tetrasomy X
Pentasomy X
Turner syndrome

# Common Themes

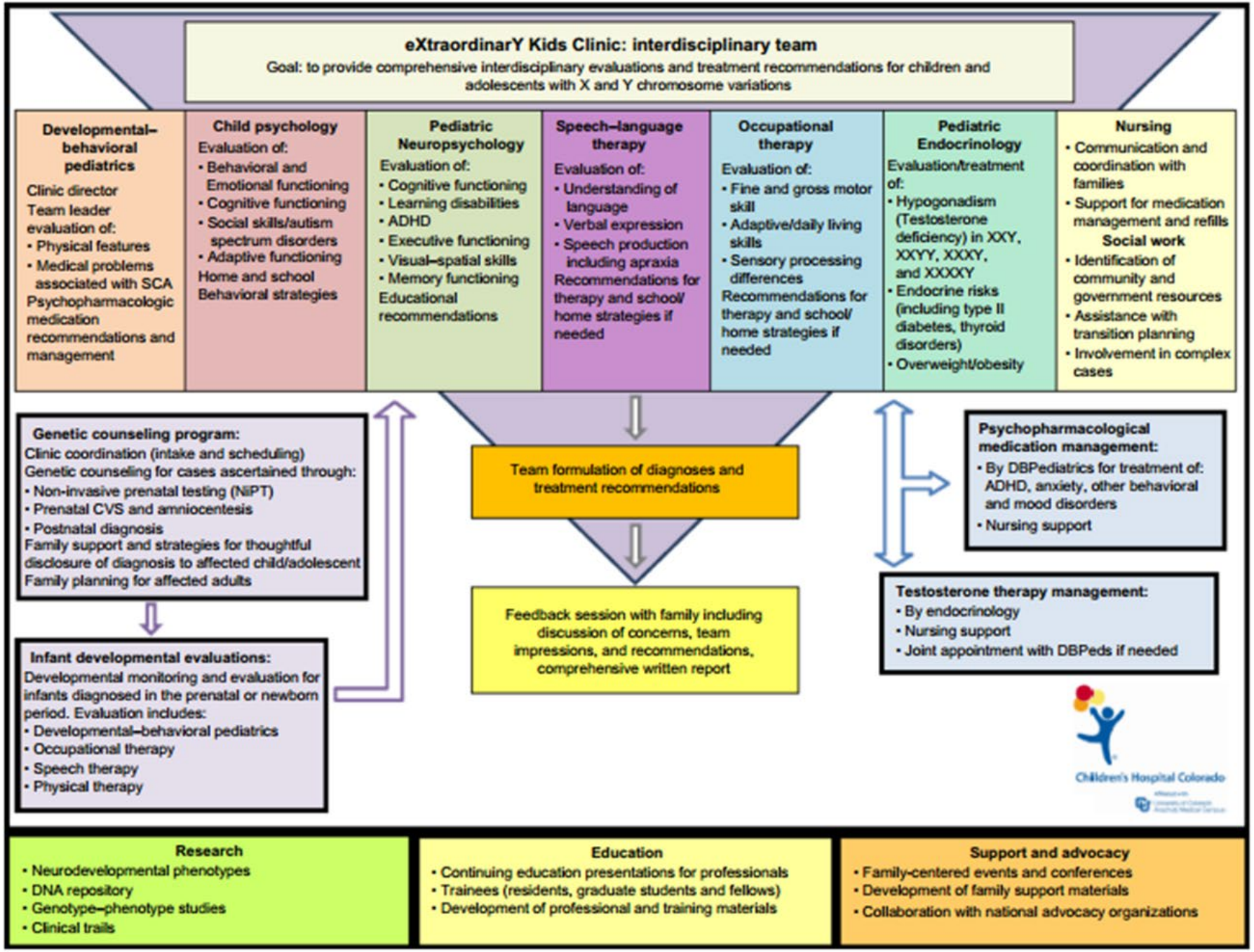
Families felt that medical professionals, therapists, and educators usually knew very little about their child's genetic condition (except some endocrinologists)

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 XXY, XXYY, 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 (NiPT)
• 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
Team formulation of diagnoses and treatment recommendations
Feedback session with family including discussion of concerns, team impressions, and recommendations, comprehensive written report
Psychopharmacological medication management:
• By DBPediatrics 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
Children's Hospital Colorado
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

A New Look at XXYY Syndrome:
Medical and Psychological Features

Original Article

REVIEW ARTICLE

RESEARCH ARTICLE

AMERICAN JOURNAL OF medical genetics PART A

Increased Number of Sex Chromosomes Affects Height in a Nonlinear Fashion: A Study of 305 Patients With

J Genet Counsel (2015) 24:88–103
DOI 10.1007/s10897-014-9741-4

ORIGINAL RESEARCH

"How Should I Tell my
of Sex Chromosome A

Anna Dennis · Susan Howell · Lisa Cor
Nicole Tartaglia

ORIGINAL ARTICLE

AMERICAN JOURNAL OF medical genetics PART A

Expanding the Phenotype of Triple X Syndrome:
A Comparison of Prenatal Versus Postnatal Diagnosis

Kristen Wigby,[1,2] Cheryl D'Epagnier,[1] Susan Howell,[1,3] Amy Reicks,[4] Rebecca Wilson,[3] Lisa Cordeiro,[1] and Nicole Tartaglia[1,3]*

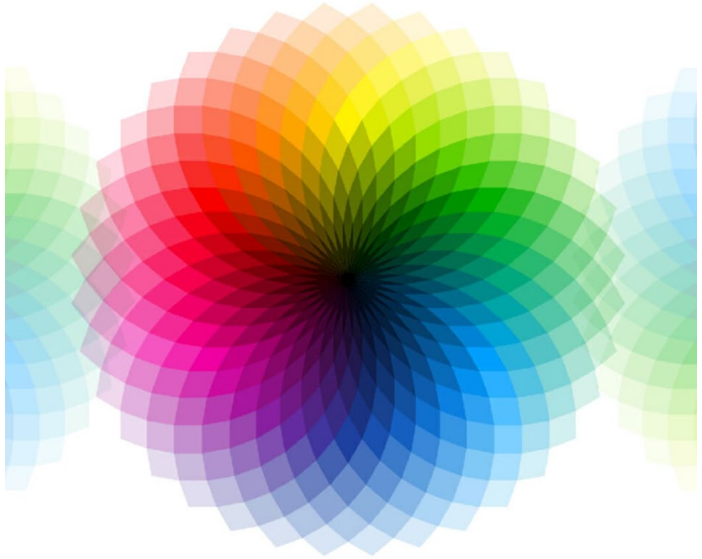
XXY).

Rogol AD[1], Tartaglia N.

role of increased M
features

M. Dalva** and A. R. Zinn††

Nicole R Tartaglia*[1,2], Susan Howell[1,2], Ashley Sutherland[1], Rebecca Wilson[2] and Lennie Wilson[3]



# WHAT WILL HAPPEN TO MY BABY?

?

# eXtraordinarY Babies Study

NBSTRN
Newborn Screening Translational Research Network

NIH *Eunice Kennedy Shriver* National Institute of Child Health and Human Development
Health research throughout the lifespan

LPDR
Longitudinal Pediatric Data Resource

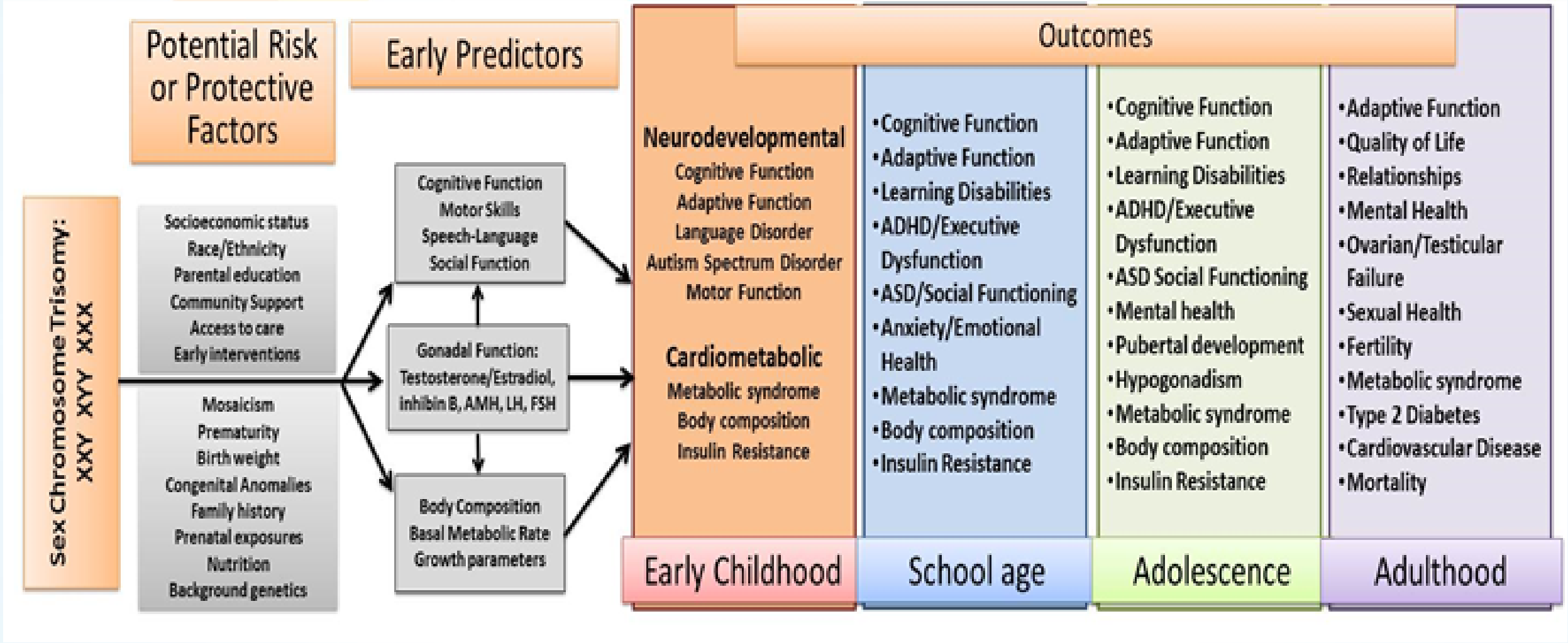
# Study Aims

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




eXtraordinarY
Babies Study

eXtraordinarY Babies Study

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)

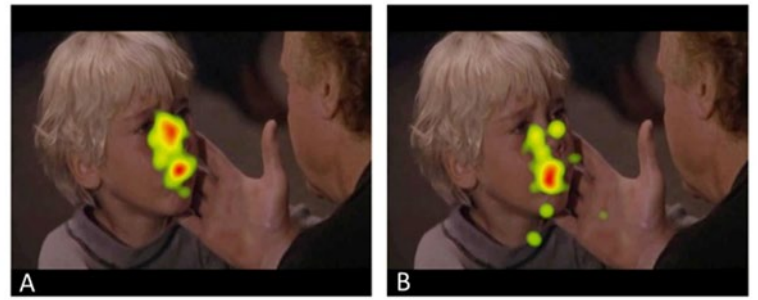
Parent Cognitive / Language / Executive functioning

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)

| | Enrolled | Goal |
|---|---|---|
| XXY | 77 | 120 (64%) |
| XYY | 7 | 40 (18%) |
| XXX | 15 | 40 (38%) |
| XXYY | 2 | -- |
| **TOTAL** | **101** | **200 (51%)** |

**Come to our poster for more details and preliminary results!**

# Enrolling!

extraordinarykidsclinic@childrenscolorado.org

<u>Participation Includes:</u>

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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Shanlee Davis, MD, MS
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Laura Pyle, PhD
Caroline Harrison, MPH

**Collaborators:**

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Judith Ross, MD (Thomas Jefferson Univ/Nemours)
Karen Kowal, PA-C
Najiba Lahlou, MD, PhD

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AXYS
Facebook


**THANK YOU ALL FAMILIES AND EXTRAORDINARY BABIES!!**