

Use of Telemedicine Approach to Conduct Dysmorphology Exams: The MotherToBaby/ OTIS Pregnancy Studies Experience

Kao KK, Chambers CD, Johnson DL, Jones KL.
Department of Pediatrics
Division of Dysmorphology and Teratology
University of California San Diego



UC San Diego
SCHOOL OF MEDICINE



Disclosure

- No conflict of interest
- Dr. Chambers receives research funding from the following industry sponsors and a foundation:
 - AstraZeneca; Celgene; GlaxoSmithKline; Janssen Pharmaceuticals; Pfizer, Inc.; Regeneron; Hoffman La-Roche-Genentech; Genzyme Sanofi-Aventis; Takeda Pharmaceutical Company Limited; Sanofi; UCB Pharma, USA; Sun Pharma Global FZE; Gilead; Novartis Pharmaceuticals and the Gerber Foundation.

INTRODUCTION



What are MotherToBaby Pregnancy Studies?

MotherToBaby Pregnancy Studies is a research program seeking to understand how certain medications, vaccines, or health conditions may affect pregnancy. Our research will help moms and health providers understand what may be safe and what should be avoided in pregnancy. All of our studies are observational: we will never ask a mom to take or change any aspect of her current health routine.



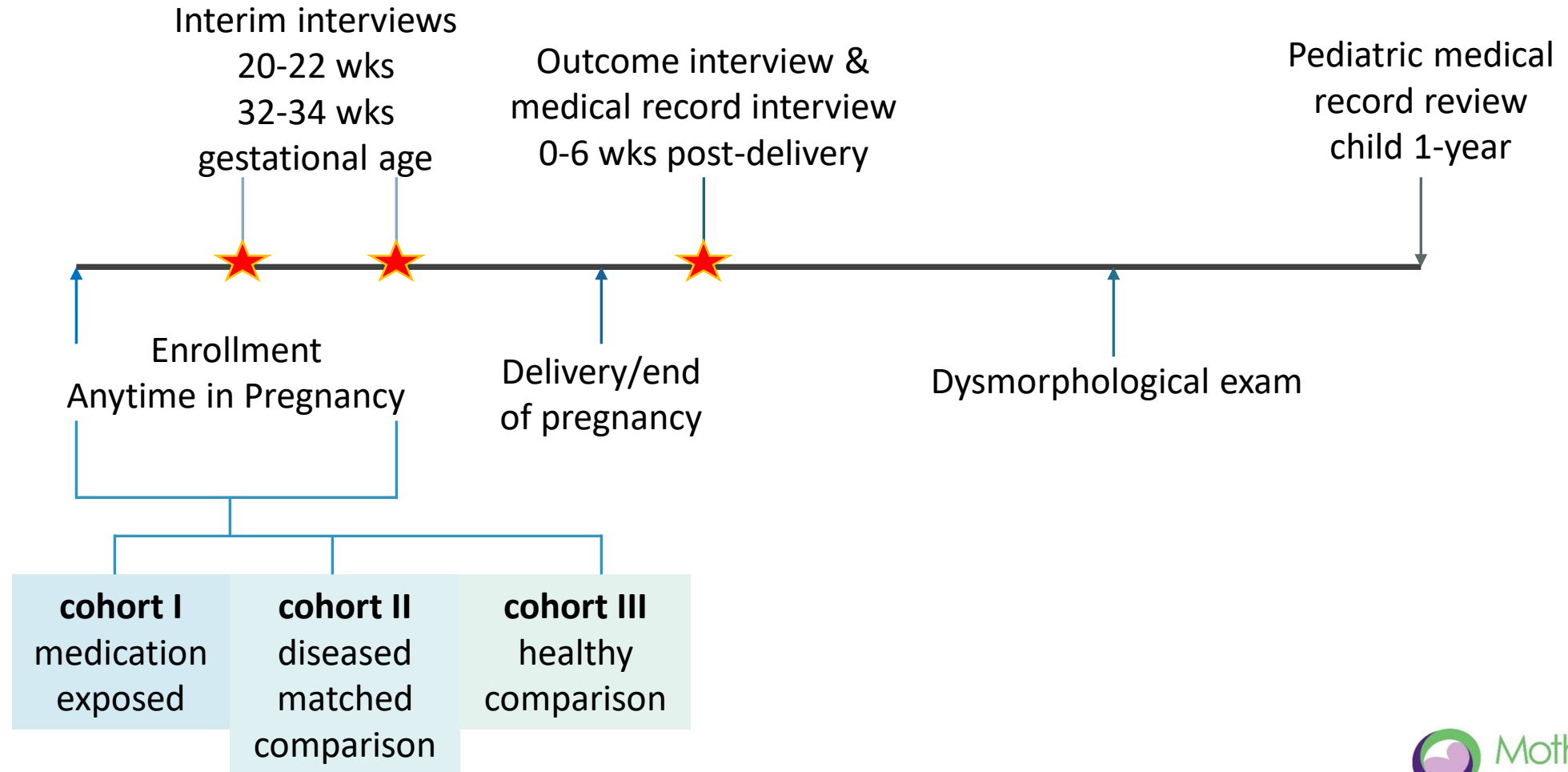
GET HELP



Cohort Study Design

- Prospective cohort studies of pregnancies exposed to target medication/vaccine with comparison groups
- Women residing in US and Canada
- Information collected:
 - demographics, SES, pregnancy and medical history
 - Rx and OTC exposures
 - alcohol, tobacco, vitamin use
 - prenatal testing, medical procedures
 - disease severity
 - pregnancy, delivery, and neonatal complications

Cohort Study Design Timeline



MotherToBaby Pregnancy Studies Objectives

Is there an increased risk for:

- Major structural birth defects, specifically a pattern up to 1 year of age
- **Pattern of minor structural birth defects**
- Spontaneous abortion, stillbirth, preterm delivery
- Birth and postnatal growth, serious infections, hospitalizations up to one year of age

Importance of Dysmorphology Exam

- Most known human teratogens are associated with a pattern of congenital birth defects, which may include minor structural anomalies that occur more frequently than associated major structural defects
- Given the typically small sample sizes in pregnancy registries, dysmorphology exams may provide added sensitivity to identifying an effect of prenatal exposure on embryo/fetus structural development

Examples of Minor Malformations



A single maxillary central incisor



Syndactyly (fusion) of the toes, can also be seen in the fingers



Fifth finger clinodactyly (incurving of the 5th finger)

Solomon BD, Muenke M. When to suspect a genetic syndrome. Am Fam Physician. 2012;86(9):826-33.

Also see Stanford Newborn Nursery Photo Gallery

METHODS

In-Person to Telemedicine Exams

- With the limited ability to perform in-person home visit examinations during the COVID-19 pandemic, a telemedicine approach to performing the physical exams was developed and tested to continue to assess children for minor anomalies
- The standard checklist of 132 minor anomalies used for the in-person exam was modified to exclude features that would not be feasible to identify remotely
- An instructional manual was developed to provide information on what to expect during the telehealth exam and how to prepare for it
- A check list was developed to aid with exam scheduling, as well as to assist the scribe while recording the examiner's findings
- With consent, photographs and video were captured as part of the exam

Instructional Manual

7. Hands



- a. Focus the camera on top of one of your child's hand so that all fingers and fingernails can be seen by the doctor. Hold for 5 seconds.
- b. Stretch your child's fingers out if they're curled in (uncurl a fist) so that your child's hand is open flat with palm down (towards the floor).
 - i. Focus the camera on your child's thumb. Hold for 5 seconds.
 - ii. Focus the camera on the pinky finger. Hold for 5 seconds.
- c. Turn your child's hand so that the palm is face up, and focus the camera on your child's palm.
 - I. Uncurl your child's fingers so the doctor can see the creases on the palm of the hand. Hold for 5 seconds.
 - II. Focus the camera on each of the uncurled fingers (still palm side up) so the doctor can see the creases on the fingers.
 1. Start with one finger and move onto the next.
 2. Hold the camera for 3 seconds for each individual finger.
- d. Repeat with the other hand

Telemedicine Exam Form

Patient's Name: _____
 Mother's Name: _____
 DOB: _____ Preg ID: _____
 Date Seen: _____ Budget: _____

Calvarium	+	Eval	No Exam
Bifrontal Diameter Narrow			
Frontal Bossing			
Metopic Ridge			
Metopic Suture Open			
Third Fontanel			
Large Anterior Fontanel			
Occiput Prominent			
Occiput Flat			
Hair Whorl Double			
Hair Whorl Triple			
Hair Whorl Absent			
Hair Whorl Midline			
Hair Pattern Unruly			
Frontal Upsweep			
Widow's Peak			
Depigmentary Hair Changes			
Scalp Defect			
Plagiocephaly			
Sutural Synostosis			
Other			
Face			
Supraorbital Ridges			
Prominent			
Hypoplastic			
Eyebrows			
Synophrys			
Medial Flare			
Nasal Bridge			
Flat			
Prominent			
Nostrils			
Anteverted			

Mouth	+	Eval	No Exam
CLEFT LIP			
Cleft Alveolar Ridge			
CLEFT PALATE			
Micrognathia			
Prognathia			
Macroglossia			
Microglossia			
Prominent Lingual Frenuli			
Other			
Ears			
Preauricular Pit			
Left			
Right			
Preauricular Tag			
Left			
Right			
Altered Shape / Position			
Other			
Neck			
Webbed			
Short			
Broad			
Low Post Hair Line			
Hair Upsweep			
Branchial Sinus			
Other			
Chest and Abdomen			
Supernumerary Nipples			
Left			
Right			
Poland Sequence			
Pectus Excavatum			
Pectus Carinatum			
Diastasis Recti			
Umbilical Hernia			
Other			



After the Telemedicine Appointment

- Post exam letter and summary letter of the exam findings
- Telehealth participation survey

RESULTS

Telemedicine Exams Completed

Between January 20, 2021 and June 20, 2022:

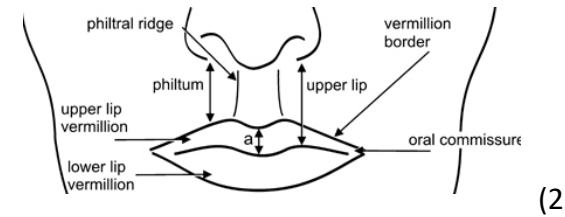
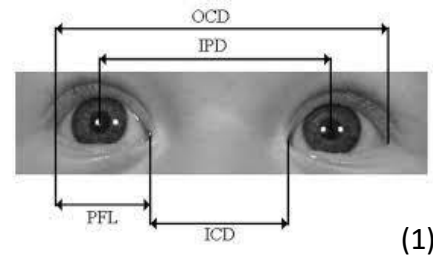
- 202 children (4 twin pairs) received the telehealth exam
- Age range 11 months to 7 years

Benefits of Telemedicine

- Feasible to examine majority (84%) of the standard physical features
- Cost efficient
- Time saving
- Convenience in scheduling for participants and dysmorphologists
- Recording of exam

Limitations of Telemedicine

- Some features, such as measurements of the philtrum, palpebral fissure (PF), and inner canthal distance (ICD) were not possible to assess



- Technology problems (e.g. poor internet connection, problems with lighting and camera angle, video quality)
- Cooperation of the child (e.g. holding still)
- More frequent postponement or cancellations

(1) https://dentistry-ipce.sites.olt.ubc.ca/files/2019/03/E2c_Gomez.pdf; (2) <https://elementsofmorphology.nih.gov/anatomy-oral.shtml>

Conclusions

- It was feasible to complete a majority of the standard physical features using a telemedicine approach
- Telehealth exams offer a cost-efficient and sustainable approach to performing physical examinations to identify minor structural malformations
- Future work will:
 - Compare findings on telemedicine to in-person examinations for a sample of the same children
 - Loss of sensitivity for features that cannot be evaluated by telemedicine will be considered
 - Exploring methods to capture palpebral fissure, inner canthal distance, and philtrum measurements

Telemedicine Exam Feedback

- Mostly positive feedback, especially regarding the convenience of scheduling and multiple re-scheduling. Some comments by participants:
 - ✓ They are flexible with the time so it worked when my daughter was most amenable to the appointment
 - ✓ It was fast and easy, but also unpleasant/not fun for my baby
 - ✓ I was very impressed with the whole exam experience
 - ✓ I have a very wiggly three year old and they were super accommodating
 - ✓ I am happy to have participated

DISCUSSION QUESTIONS?