Ethical Issues in Treatment Decisions for Children with Trisomy 13 and 18

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Ethical Issues in Trisomy 13 and 18
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Goals and Objectives
Understanding the Issues

- Review the nature of trisomies in general and Trisomy 13 and 18 in particular
- Discuss the medical problems commonly associated with Trisomy 13 and 18 and the treatments available
- Review the ethical issues raised by the conditions, their prognosis and the positive and negative effects of treatments
- Learn from the parent of a child with Trisomy 13 the challenges and rewards of the experience
- Discuss a process for approaching cases of Trisomy 13 and 18, identifying and supporting the decision-maker/s
What Is a Trisomy?
They’re More Common Than You Suspect

A Trisomy is a condition in which the cells of a person have three copies of a chromosome instead of the normal two copies.

- Trisomy 21 (Down Syndrome)
- Trisomy 18 (Edwards Syndrome)
- Trisomy 13 (Patau Syndrome)
- Trisomy 9
- Trisomy 8 (Warkany Syndrome 2)
- Trisomy 22

Trisomy 16 is the most commonly occurring (1%) trisomy in pregnancies, but virtually all miscarry.
How Do Trisomies Happen?
A Brief Review of Meiosis (Egg and Sperm Creation)

Types of Trisomies

- **Nondisjunction** – Extra whole chromosome in every body cell
- **Translocation** – Partial duplication
- **Mosaicism** – Only some of the body’s cells have the extra chromosome
Trisomy 18
What the Karyotype (Chromosome Analysis) Looks Like
Trisomy 18 (Edwards Syndrome) Statistics

- Incidence in live births 1/6,000 – 1/8,000
- Second most common autosomal trisomy after Trisomy 21 (Down Syndrome)
- Previously said to be “incompatible with life,” but 50% live longer than 1 week, and 5 – 10% live beyond the first year
- Causes of death
  - Central apnea (forgetting to breath) very common
  - Upper airway obstruction with hypoventilation
  - Aspiration
  - Heart failure from congenital heart abnormalities
  - Decisions to withhold aggressive care

www.ncbi.nlm.nih.gov/pmc/articles/PMC3520824/
Trisomy 18
Common Features

- Small head (microcephaly)
- Small jaw (micrognathia)
- Low-set ears
- Clenched hands (“Fisting”)
- rocker bottom feet
- Umbilical or inguinal hernia
- Delayed growth
- Kidney problems
- Part of the intestinal tract is outside the stomach (omphalocele)
- The esophagus doesn’t connect to the stomach (esophageal artesia)
- Heart defects (80 – 100%):
  - VSD (Ventricular Septal Defect): a hole between the lower chambers
  - ASD (Atrial Septal Defect): a hole between the upper chambers
  - Coarctation of the aorta: a narrowing of the exit vessel from the heart
- Brain Cysts (Choroid Plexus)
- Severe developmental delays

www.trisomy18.org/what-is-trisomy-18/
Developmental Delay in Trisomy 18

The Range

- All older surviving children have significant delays ranging from marked to profound
  - Most have no expressive language
  - Most never learn to walk
  - Typical developmental age in older children is 6 – 8 months

- However
  - Some learn to feed themselves, use sign boards and follow simple commands
  - “All children acquire abilities such as recognizing their family and smiling appropriately.”

- Thus many have more abilities than the stereotype suggests

www.ncbi.nlm.nih.gov/pmc/articles/PMC3520824/
Prenatal Diagnosis of Trisomy 18
Often an Incidental Finding on Ultrasound
Prenatal Diagnosis of Trisomy 18
Often an Incidental Finding on Ultrasound

Omphalocele – Intestines outside the abdomen
Trisomy 13 (Patau Syndrome)
Incidence and Characteristics

- **Statistics**
  - Incidence: 1/16,000
  - Frequency increases with maternal age

- **Characteristics**
  - Cleft lip plus or minus cleft palate
  - Small or poorly developed eyes (microphthalmia)
  - Extra fingers or toes (polydactyly)
  - Heart defects
  - Brain and spinal cord abnormalities
  - Weak muscle tone (hypotonia)
  - Severe intellectual disability

- Due to the presence of several life-threatening medical problems, many infants with trisomy 13 die within their first days or weeks of life. Only 5 – 10% of children with this condition live past their first year.

Prenatal Diagnosis of Trisomy 13
Often an Incidental Finding on Ultrasound
Prenatal Diagnosis of Trisomy 13 and 18
An Early Decision Point

- Elective termination of the pregnancy (86% in one study)
- Spontaneous miscarriage
- Fetal death during the delivery
- Absence of spontaneous respirations after birth
- Identification of serious congenital abnormalities after birth
Common Medical Issues in Trisomy 13 and 18

Need for a Birth Plan and On-Going Decision Making

- Respiratory insufficiency
- Difficulty or inability to feed orally
- Cardiac malformations
- Other organ malformations
- Profound developmental delay
Medical Treatment Options
Some Straightforward, Some Not So Much

- Resuscitation and respiratory support
  - At birth
  - Subsequent
- Feeding
  - By mouth
  - By gastrostomy tube
- Surgical repair of cardiac and other abnormalities
- Special education facilities and medical daycare
- Hospice and palliative care
Parent Support Networks
Sources of Information and Encouragement

Trisomy 13
A Handbook for Families

SOFT
Support Organization for Trisomy 18, 13 and Related Disorders

CHROMOSOMES
SHOULD NOT DETERMINE THE VALUE OF A LIFE
Basic Ethical Principles
Doing the Right Thing

- Autonomy
- Beneficence
- Non-Maleficence
- Justice
- Truth-telling
Informed Consent and the Pediatric Patient

- Ordinarily, parents have the right to give (or refuse) permission for treatment for their children.
- The “best interests” standard should serve as the basis for decision making.
- Care providers play an important role in providing accurate, unbiased information to inform parental decision making.
- Physicians should only attempt to override parents’ decisions if it appears clear that they are not in the best interests of the child.

Ethical Issues
What Are the Arguments Against Intervention?

- **Futility**
  - Merritt et. al. repeatedly use the term “lethal” in discussion
  - “There is no cure for trisomy 18 or 13.”
  - “. . . outcomes are often not improved.”

- **Non-Maleficence**
  - “Some physicians believe that aggressive intervention violates the Hippocratic injunction of *primum non nocere*.”

- **Resource Allocation**
  - “Although economic considerations should not be deliberated at the bedside, we believe they cannot be ignored.”

- **Quality of Life**
  - Some authors such as Merritt et. al. believe that the quality of life experienced by trisomy patients does not justify intervention.

Appreciation of the Rights of the Handicapped: “Quality of Life”

- The experience of life as viewed by the patient
- Evaluation by others is inherently subjective
- Assessments involve weighing the benefits and burdens of continued life and the therapies needed to maintain it
- Should not include considerations of potential “social worth”

The authors favor a more paternalistic approach to case management

- Information to be provided to parents stresses the “lethal” nature of trisomies, the limited benefit of treatment and its burdens
  - “When families insist on medical interventions that are not appropriate . . . we must (1) identify for families the bodily injury and suffering we were (are) inflicting on the family member, (2) provide intense emotional support, and (3) maintain a good relationship . . .”
  - “There are good reasons to make sure that infants with trisomy 18 do not suffer unnecessarily and that they are not subjected to major surgery or prolonged intensive care where there is little chance of benefit.”

- The authors appropriately encourage a Palliative Care referral at the time of fetal diagnosis

Basic Ethical Principles
Doing the Right Thing for Trisomy Infants

- **Autonomy** – Parents remain the most appropriate decision makers for their infants.

- **Truth-telling** – Care providers should provide accurate, factual, unbiased information to inform parental decisions.

- **Beneficence** – Many treatments can and do benefit infants and children with trisomies.

- **Non-Maleficence** – Appropriate management to alleviate pain and suffering are critically important, whether or not more aggressive interventions are undertaken.

- **Justice** – Otherwise beneficial treatments should not be withheld simply because infants are expected to be handicapped.
Community
PedsCare

OUR FAMILY BIRTHING PLAN

Our beloved baby has been diagnosed with _______________________. Your compassion and understanding during this bittersweet and difficult time are appreciated deeply. We believe that the memories of our actions during this time with our baby will later console us.

We understand that birth situations may arise that were not anticipated and decisions will need to be made. We simply ask you to keep us informed so we can participate in the decisions as to what is best for our baby, _______________________. We ask that no intervention be taken without our approval, other than what is outlined below. We trust you will respect our wishes.

Please call our baby _______________________. Ask us how we feel and what special stories we have from this pregnancy. This validates and honors our baby’s life.

Please check appropriate box for your request.

☐ We would like to remain in the same room for labor, delivery and recovery. If a C-section is necessary, we want our baby to remain in the operating room with both parents and we want to have recovery in a birthing room to allow for more time together as a family.

During labor and delivery we would like

(Special music, soothing practices, etc.)

☐ We want to hear our baby’s heartbeat before labor progresses.

We would like ☐ no ☐ external ☐ internal fetal monitoring.

If our baby’s heart stops prior to delivery we ☐ do ☐ do not want to be informed.

We ☐ want ☐ do not want to videotape the birth.

We ☐ want ☐ do not want to have photos during the birth.

We would like to have these people in attendance: _______________________

Any medications given during labor to the mother should be given in doses to provide maximum comfort while allowing mother to remain alert.

Please allow ______________________ to cut the umbilical cord.

We would like oral/nasal suctioning for ______________________’s comfort only and NO intubation without our permission.

After our baby is born, we ask that he/she be wiped, suctioned, wrapped in a blanket and:

If alive, handed to ______________________.

If stillborn, handed to ______________________.
☐ We wish to cuddle our baby immediately and ask that vital signs, weight, labs and medications be postponed if possible.

☐ If our baby has fewer or more problems than expected, please discuss all options with us.

Other than routine post-delivery care, we wish for private time with our baby. We will discuss exceptions that should be made.

If our baby can’t suck or nurse, we wish to provide comfort with ☐ breast milk or ☐ formula given by ☐ a dropper, or with ☐ placement of a nasogastric/orogastric tube.

☐ We wish to use the services of the hospital chaplain (either for pastoral support or to contact a representative from our faith).

☐ We have planned for our spiritual family to be here for support.

They are:

We are planning a ☐ baptism/christening or ____________________.

Memorial/funeral plans ☐ have been ☐ have not been made for our baby. 
Funeral Home is: ____________________.

☐ We wish to hold our baby as he/she is dying or has died and want to keep their precious body with us as long as possible.

☐ We would like to bathe and dress our baby. ☐ We have our own clothes.
☐ We need a burial garment.

We have arranged for photos by Now I Lay Me Down to Sleep. Our photographer is ____________________.

☐ We would like to have keepsake/memory making to include: cord clamp, lock of hair, ID bracelet, crib card, hand and footprints/molds, or ____________________.

Other considerations:

Signed by:

_____________________________   ______________________________
Parent                                          Parent

Date: ______________________________

Our obstetrician: ______________________________

Our Community PedsCare staff: ______________________________

Patient # ______________________________

Community PedsCare is a palliative and hospice program for children with life-limiting and life-threatening conditions and their families. Established in 2000, it is a program of Community Hospice of Northeast Florida in collaboration with Wolfson Children’s Hospital, Nemours Children’s Clinic and the University of Florida-Jacksonville, bringing comfort and enhanced quality of life to the youngest members of our community.

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