Trisomy 18:
Edward’s Syndrome:

The Decision

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Trisomy 18: The Overview

- First recognized as a specific entity by the discovery of the extra 18 chromosome in babies with a particular pattern of malformation.
- Second most common multiple malformation syndrome
- Incidence of 0.3 per 100 newborn babies.
Trisomy 18: Overview

- Trisomy 18 Karyotype
Trisomy 18: Overview

- 3:1 preponderance of females to males
- More than 130 different abnormalities noted on patients with Trisomy 18 syndrome
Trisomy 18: Overview

- Mosaicism for an additional chromosome 18 leads to partial clinical expression with longer survival and any degree of variation between near normal and the full pattern.

- Partial trisomy 18:
  Trisomy of the short arm causes very nonspecific clinical picture and mild or no mental deficiency.
Trisomy 18: Overview

- Trisomy for the entire long arm cannot be distinguished from full trisomy 18.
- Trisomy for the distal one third to one half of the long arm leads to a partial picture of trisomy 18 with longer survival and less profound mental deficiency.
Trisomy 18: Diagnosis

- Prenatally
  - Ultrasound Findings Include:
    - IUGR
    - Polyhydramnios or oligohydramnios
    - Cardiac malformations
    - Choroid plexus cyst
    - Hydrocephalus
    - Melomeningocele
    - Abdominal wall defects
    - Single umbilical artery
    - Clenched fists or radial limb defects
Trisomy 18: Diagnosis

- **Prenatal History**
  - Decreased fetal movement
  - Association with abnormal maternal screening: triple screen: decrease in maternal serum alpha-fetoprotein, HCG and unconjugated estriol
  - HCG appears to be most sensitive for suggestion of risk

- **Amniocentesis and Chromosome analysis**
Trisomy 18: Abnormalities

Abnormalities found in 50% or more

- Small for gestational age, IUGR
- Feeding difficulties, feeble activity, or weak cry
- Central nervous system manifestations: hypertonia, seizures, apnea, mental deficiency
Trisomy 18: Abnormalities

- Craniofacial anomalies: prominent occiput, narrow bifrontal diameter, microphthalmia, short palpebral fissures, low set malformed auricles, small oral opening with narrow palatal arch and micrognathia
Trisomy 18: Abnormalities

- Cardiac malformations: ~90% include VSD, ASD, PDA, and double outlet right ventricle and polyvalvular disease
- Extremity malformations: clenched hands, overlapping fingers, abnormal creases, hypoplastic nails, rocker-bottom feet
- Thorax: short sternum, hypoplastic nipples
Trisomy 18: Characteristics
Trisomy 18: Inheritance

**Trisomy 18:** All cells affected

- Usually not inherited, but occur as random events that occur during the formation of eggs and sperm. Chance increases with maternal age.
- Nondisjunction occurs during reproductive cell division resulting in three copies of chromosome 18.
Trisomy 18: Inheritance

- An error in maternal meiosis II is most common cause of nondisjunction of chromosome 18, unlike other human trisomies studied, that show a higher frequency in maternal meiosis I
- Increased incidence with advanced maternal age
Trisomy 18: Inheritance

- **Mosaic Trisomy 18**: only some cells affected
- Usually not inherited
- Severity depends on type and number of cells affected – may range from normal to severely affected
Trisomy 18: Inheritance

- **Translocation Trisomy 18**: long (q) arm of chromosome 18 attached to another chromosome
- May be inherited
- Unaffected person may carry a rearrangement of genetic material, but no extra material (balanced translocation). Even though they have no symptoms, they can be carriers and may pass the translocation to their children.
Trisomy 18: Molecular Basis

- Chromosome 18 spans about 76 million base pairs (the building blocks of DNA) and represents approximately 2.7 percent of the total DNA in cells.

- Extra copies of some genes on chromosome 18 disrupt the course of normal development, causing the characteristic features of trisomy 18 and the increased risk of medical problems associated with this disorder.
Trisomy 18: Prognosis

- Most 18 trisomic individuals die in embryonic or fetal life
- 50% die within the first week
- 5-10% survive the first year as severely mentally defective individuals
- At least 10 reports of affected children older than 10 years of age
- Prognosis for mosaic/translocation may vary from near normal to severely affected
Most children with trisomy 18 that do survive are happy kids and are capable of developing loving relationships with family.
Trisomy 18: Therapies

- Therapy currently includes supportive care
- If not a DNR, treatments and surgeries as needed
- There are current studies looking at non-invasive methods for determining genetic abnormalities to include Trisomy 18 from non-cell maternal serum which will be useful in diagnosis but not treatment
Ethical Issue

Abortion

Versus Continuation of Pregnancy

For Genetic Defects
Ethical Issue: Abortion for Genetic Abnormalities

Despite increased legal and social acceptance of abortion, it remains an ethically contentious subject (Jones, 2007).
Ethical Issue: Abortion for Genetic Abnormalities

Considerations in the Ethical Debate Concerning Abortion:

- Autonomy (of the woman)
- Rights (of the woman and unborn child)
- Maternal-fetal relationship and assessing best interests of potential children
Ethical Issue: Abortion

Support for Aborting Fetus:

- Legitimate reason for abortion
- “Logical”
- Autonomy: Right of the mother to choose
- “Moral status” of the fetus
- Disability
- Burden to family, society
Ethical Issue: Continuation of Pregnancy

Support for Continuation of Pregnancy:

- Morally wrong to actively end life of an unborn human being?
- Right to life for fetus – society’s duty to protect?
- “Moral status” of the fetus
- Asset to family
Ethical Issue: Continuation of Pregnancy

- Potential negative psychological consequences to mother -10-20% = 130,000 women annually (Adler, 1990).
- Moral distress to staff
Psychosocial Issues

- Mother and father united in decision against abortion. Supported by extended family
- Providers tendency to present the negative, withholding current information
- Reluctance to access of medical care with limited resources
Policies and Professional Codes

- Hospital policy of no abortions performed in institution
- Consciencious objection: Avoid participation in procedure. Does not extend to caring for the patient before, during or after.
Ethical Dilemma

Ethical Principles:

- Nonmaleficence: Do no harm to the fetus
- Autonomy: Allow the mother her right to choose
- Justice: Fair access to medical services
Personal Responses

- Personal belief: against abortion
  Had family decided to have an abortion, it would be necessary to support their decision despite disagreement.

- Would not have participated in the actual procedure, but support mother surrounding it
Personal Responses

- If decision to have an abortion, provide information to include list of places to obtain a legal abortion
- Since continuation of pregnancy was decided upon, provide parents with resources (support groups, web sites, literature, information on respite care)
Personal Responses

- Involve parents in infant care. Teaching.
- Focus on positive aspects of situation
Possible Legal Issues

- Potential for legal action if all options are not discussed
- Potential for legal action if medical services are withheld due to discrimination related to the disability
"Every child . . . no matter how fragile their life or brief their days, forever changes our world."

~Victoria Miller, Founder, Trisomy 18 Foundation
References


References


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