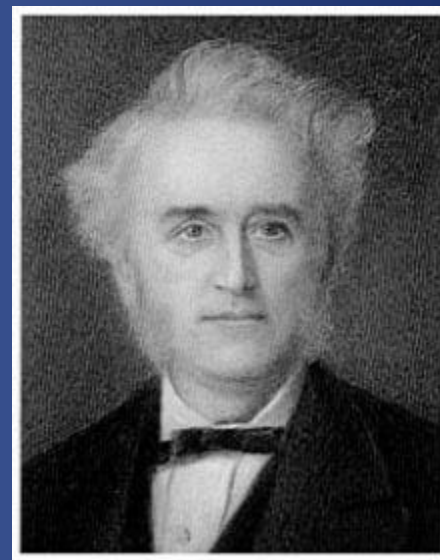


# Down Syndrome

- Neurodevelopmental syndrome named for John Langdon Haydon Down who first described the physical and behavioral characteristics in 1862



# Down Syndrome

- 1958 Jerome Lejeune and his team identified trisomy 21 (47 chromosomes-one extra chromosome 21 in cells) as cause of Down Syndrome
  - Only 2 years after it was discovered that the typical number of human chromosomes was 46



## Down syndrome

- > A genetic disorder that causes delays in physical and intellectual development.



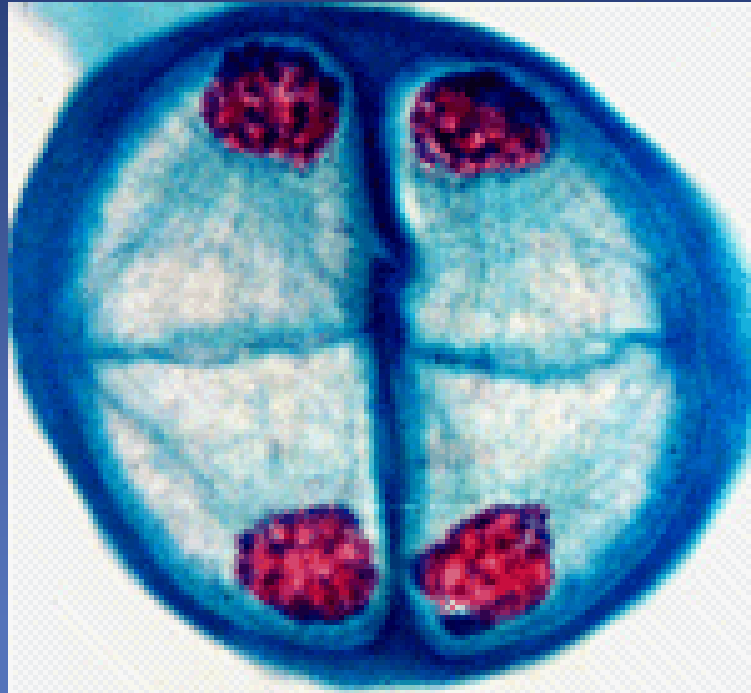
# Human Karyotype (picture of chromosomes that hold genes)



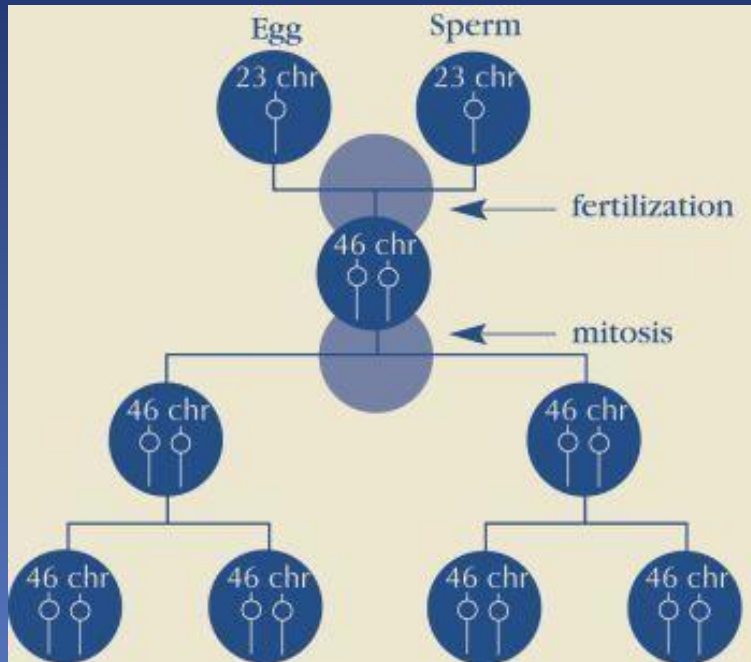
# Meiosis

Watch Video [Here](#)

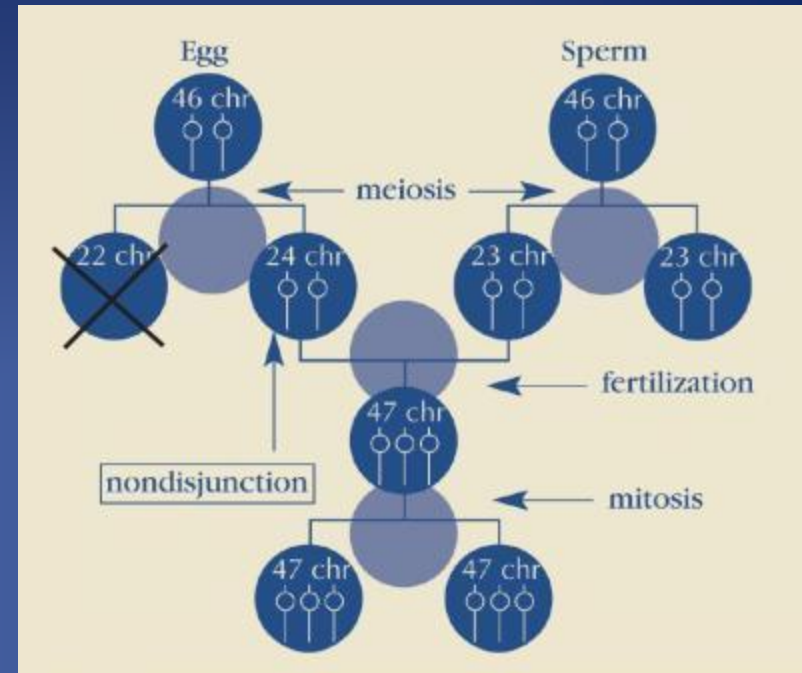
<https://www.youtube.com/watch?v=nMEyeKQClqI>



# Down Syndrome



Typical cell division- 46 chromosomes



Non-dysjunction cell division- 47 chromosomes

# Health in Down Syndrome

- Heart:
  - 40-60% born with abnormal hearts



# Health in Down Syndrome

- Eyes: cataracts and glaucoma
  - 5% Cataracts
  - 1% Glaucoma
  - Near-sightedness
- Ears: hearing loss
  - 30-75% by preschool





# Health in Down Syndrome

- Thyroid: low thyroid
  - 30% by 25 years
- Gut: celiac disease (wheat allergy)
  - 4-7%
- Immune dysfunction:
  - autoimmune/leukemia



# Down Syndrome

- Brain:
  - Seizures 8%-50% by 50 years
  - Sleep apnea
  - Developmental delays/learning differences

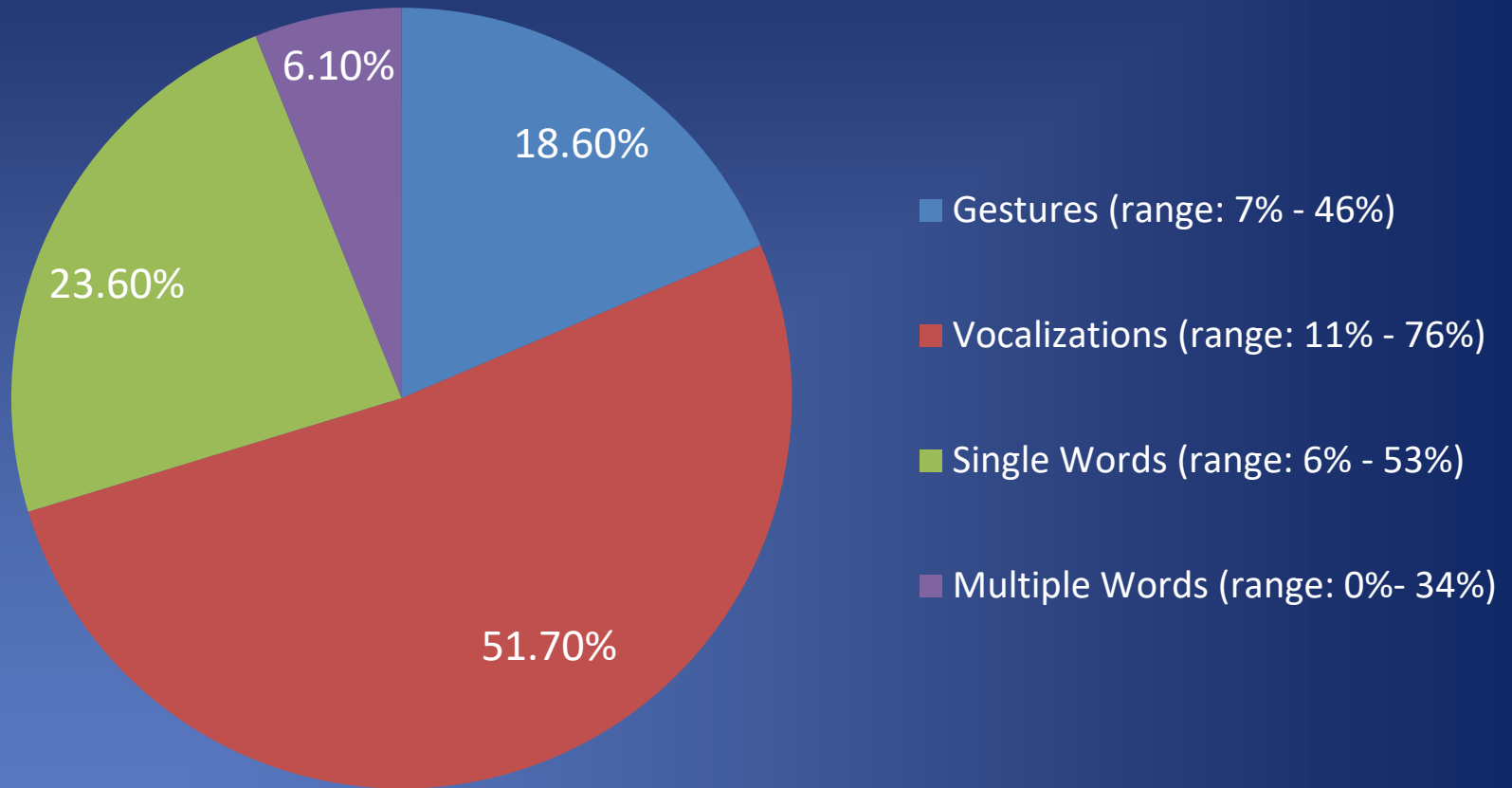


# Development in Down Syndrome

- Social and self help skills usually on track early and often remain strengths
- Motor skills (sitting, walking, running) delayed due to low muscle tone, joints
- Language skills often start on time, but usually slow in second year and affect talking more than understanding



# Early Communication Skills in DS



# Down Syndrome

- Academic/cognitive skills usually lower than adaptive/every day skills, which show improvement through mid-adulthood
- Onset Alzheimer dementia (losing memory and skills) 10 years earlier than general population
- Diabetes/obesity
- Less cardiovascular disease
- Lower blood pressure



# Development in Down Syndrome



- Social supports, cognitive skills, emotional and physical health predict adaptive function and employment

# Down Syndrome

- Kumin et al (2015) J of Applied Research in Intellectual Disability
  - 57% employed (3% full time)
  - 20% volunteered
  - 3% self-employed
  - 30% unemployed



# Down Syndrome and other diagnoses

Autism spectrum disorders thought to occur in 10% of people with Down Syndrome

ADHD thought to occur in 6-8% of people with Down Syndrome

Other behavioral diagnoses in 10-15%





# Prenatal Testing

## > Amniocentesis:

- procedure in which a small sample of amniotic fluid is drawn out of the uterus through a needle inserted in the abdomen



# Prenatal Testing

- > Chorionic Villus Sampling (CVS):
  - prenatal test to detect birth defects that is performed at an early stage of pregnancy



# Prenatal Testing

- > Percutaneous Umbilical Blood Sampling (PUBS):
  - a genetic test that examines blood from the umbilical cord to detect fetal abnormalities



# Prenatal Testing

- > Alpha-fetoprotein Test (AFP)
  - Alpha fetoprotein (AFP) is a protein normally produced by the liver and yolk sac of a developing baby during pregnancy in adults.
- > Sonogram
  - It is a picture taken of the baby during an ultrasound test.



# Age Matters

- > The chance of having a child with Down syndrome increases with the mother's age

