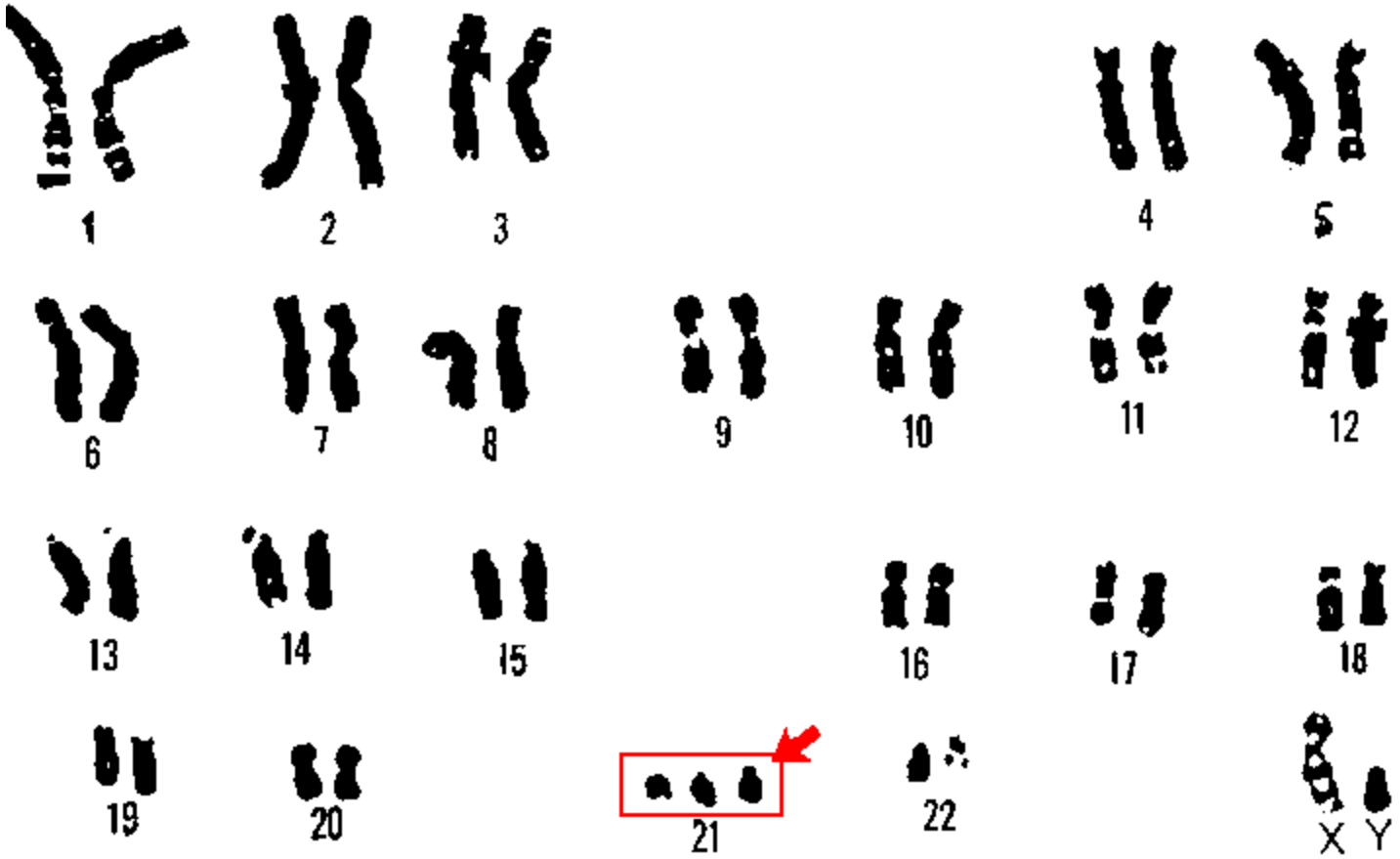
DOWN SYNDROME

DOWN SYNDROME (DS)

- Down Syndrome (DS) is caused in most cases by a **triplication of chromosome 21**, called ‘trisomy 21’ (extra chromosome on chromosome pair 21)
- Most commonly recognized genetic cause of mental retardation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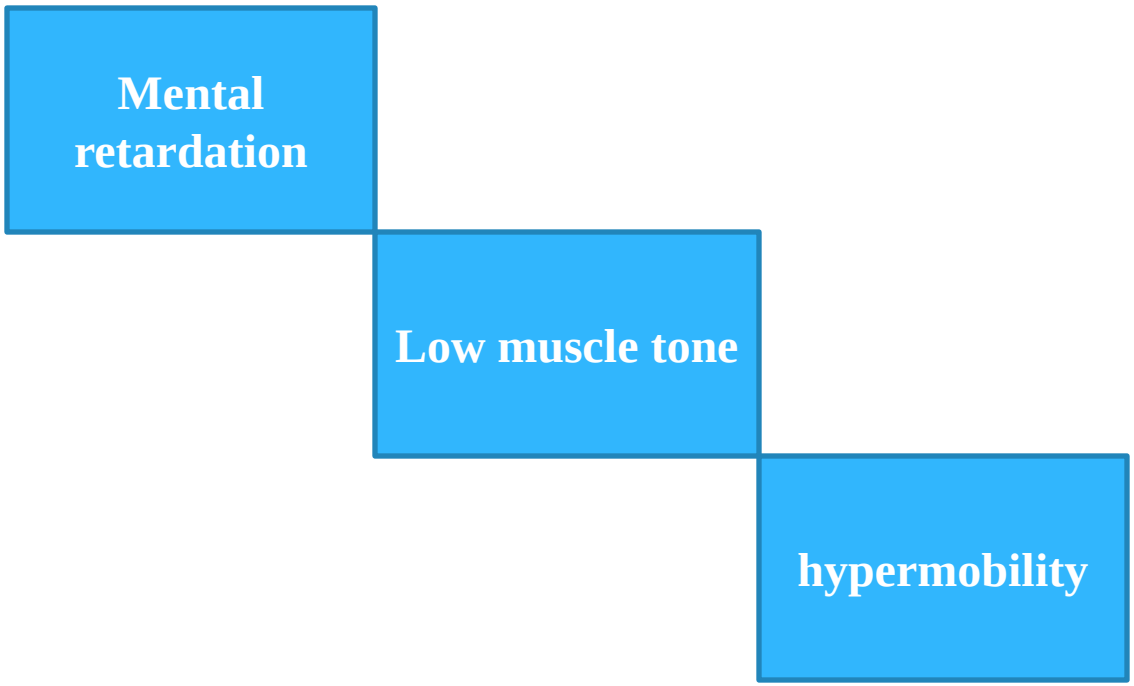
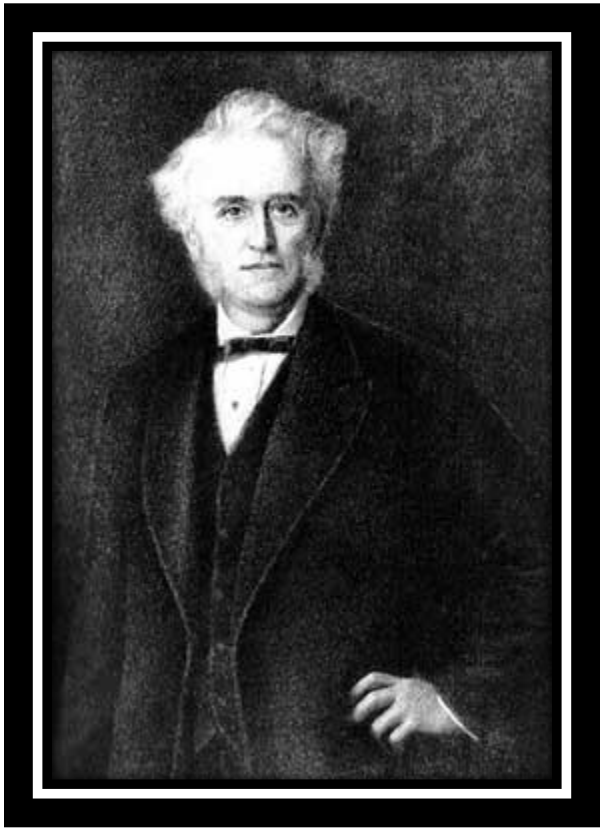
DOWN SYNDROME



Who discovered it?

- Down syndrome was first described by John Down in a published paper in 1838.
- John Down is credited for describing details and distinguishing children with Down Syndrome.



Etiology

- Most commonly recognized genetic cause of mental retardation: prevalence of 9.2 cases per 10,000 live births
- Diagnosed by Karyotype:
  - 95% - Trisomy 21
  - 2% - Mosaic (mix of diploid and trisomy 21)
  - 3 % - Robertsonian translocation (part or all of extra chromosome 21 fused with another chromosome

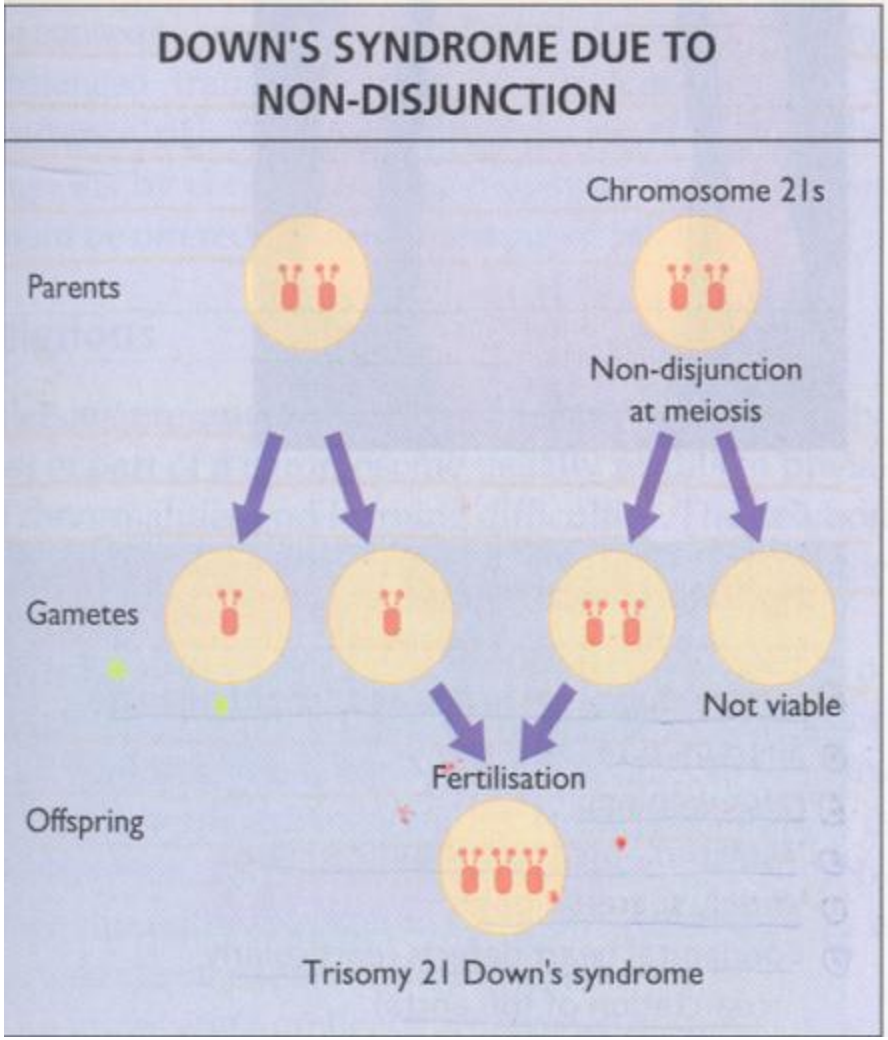
ETIOLOGY

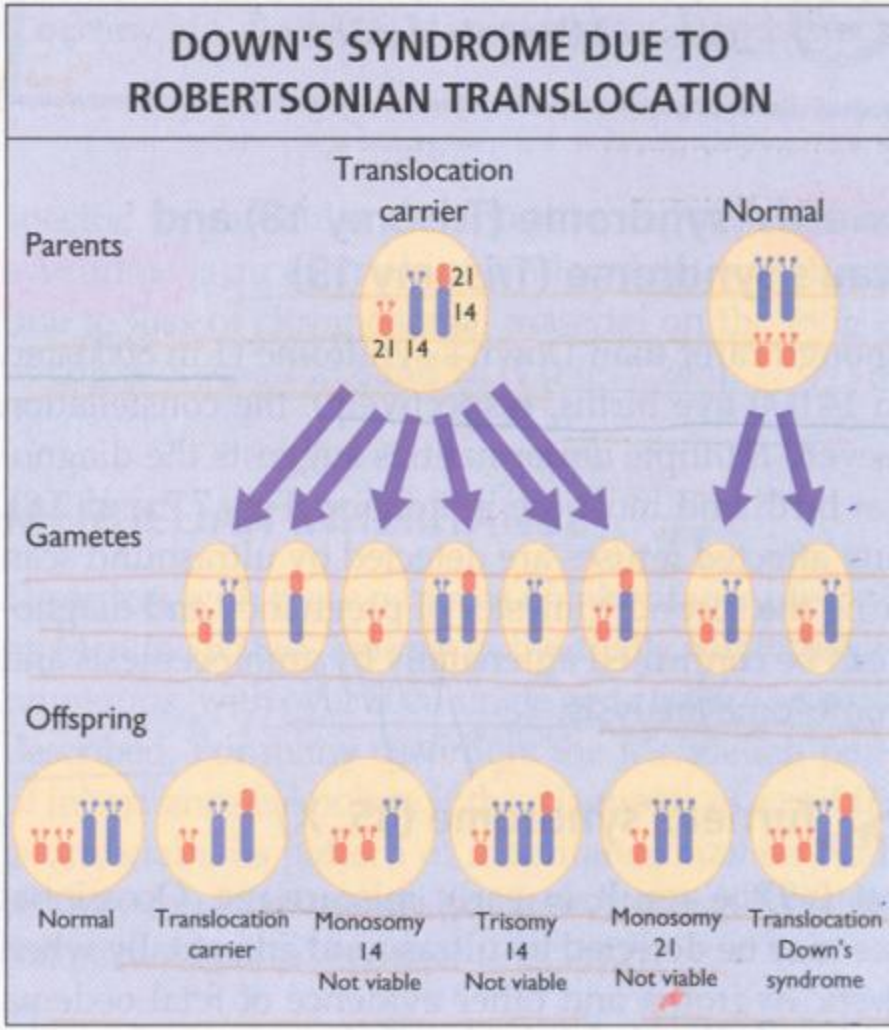
- 95 percent of occurrences of trisomy 21 result from nondisjunction during meiotic division of the primary oocyte
- Most trisomy 21 pregnancies prove to be nonviable
- 1/4 of fetuses with trisomy 21 survive to term

Why It Happens

The reason for Down Syndrome is that there is an extra chromosome. This results in 47 chromosomes instead of 46.

INHERITANCE OF DOWN'S SYNDROME





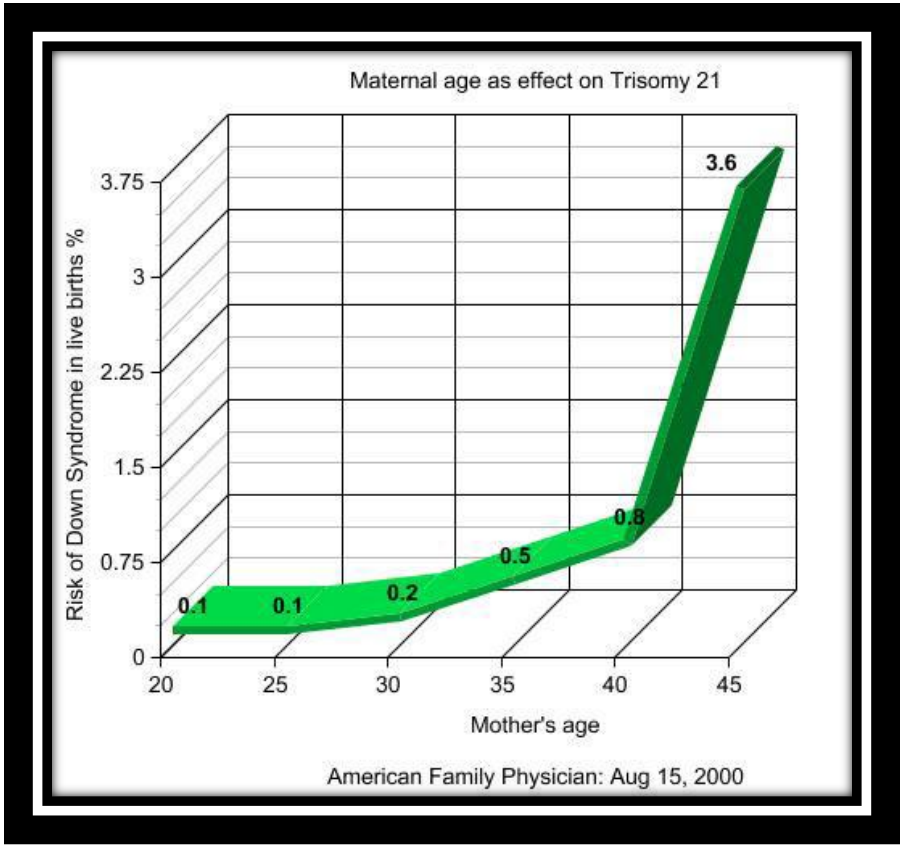
Risk factor

- Maternal Age
  - 1/1,300 for a 25-year-old woman
  - 1/365 for age 35
  - 1/30 for age 45

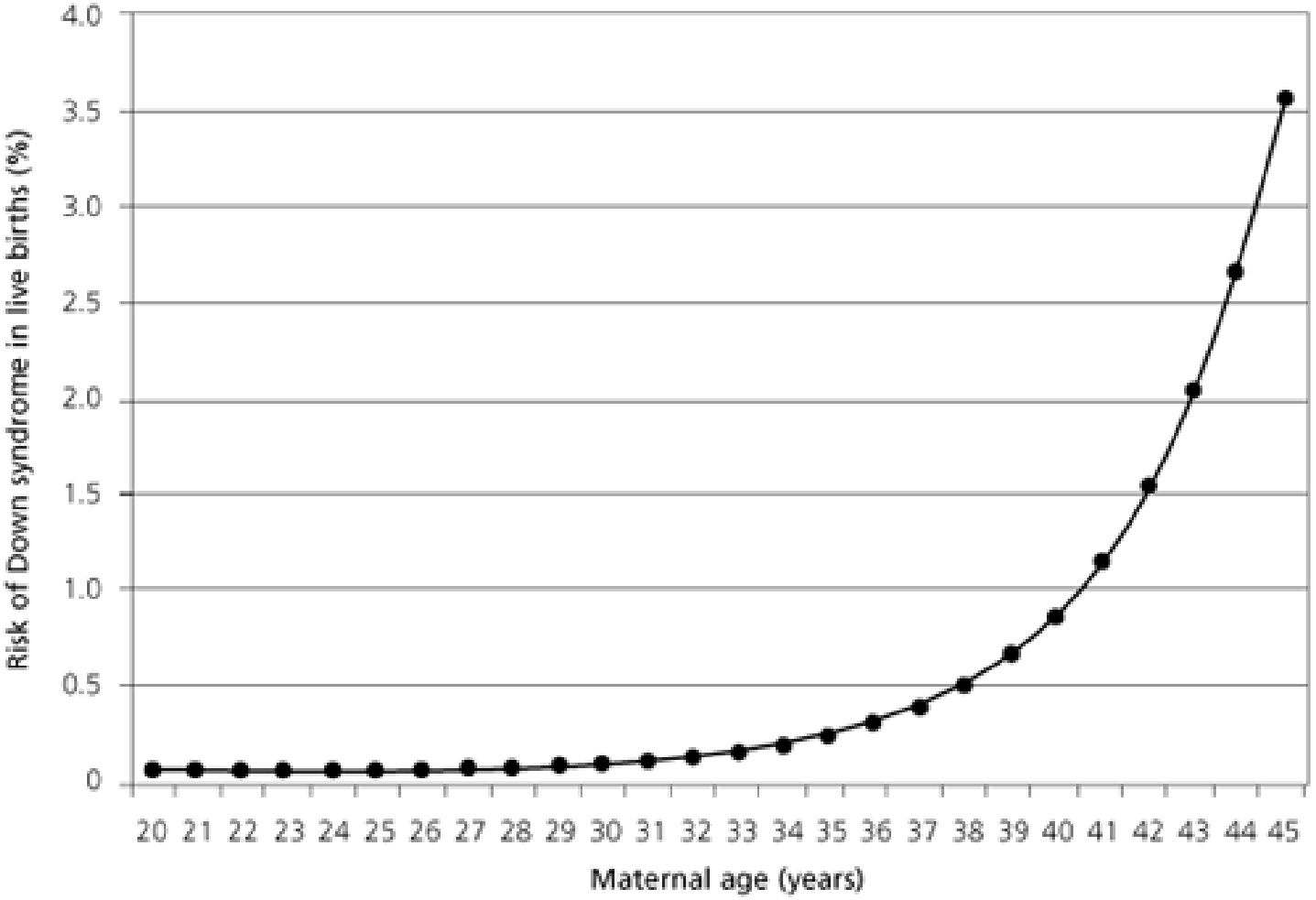
Risk Factors

Although Down Syndrome can't be completely avoided, the following things may increase the risk for Down Syndrome.

- Older mothers
- Mother exposed to X-Rays
- Parents carrying the Down Syndrome gene



Advanced Maternal Age as Risk Factor



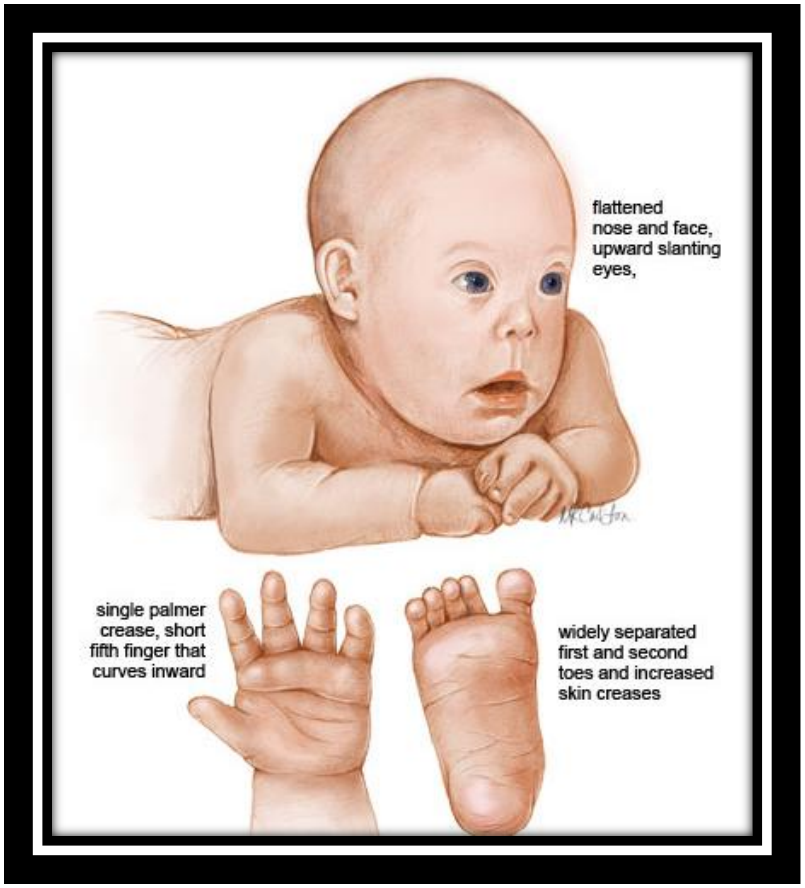
Life Expectancy

Down Syndrome can shorten life because it increases the person's risk for **health problems**. In the past two decades however, the life expectancy has almost doubled for people with Down Syndrome, from **25 to 49**. Down Syndrome itself is not fatal.

Physical signs

Physical signs of Down Syndrome include:

- Flat face
- Upward slanted eyes
- Deep palm creases
- White spots in the iris of eyes
- Loose ligaments
- Small hands
- Short neck
- Abnormally shaped ears



Health Problems

Down Syndrome puts one at a higher risk for:

- Congenital heart failure
- Intestinal problems
- Celiac disease
- Eye Problems
- Thyroid Dysfunction
- Skeletal Problems
- Dementia

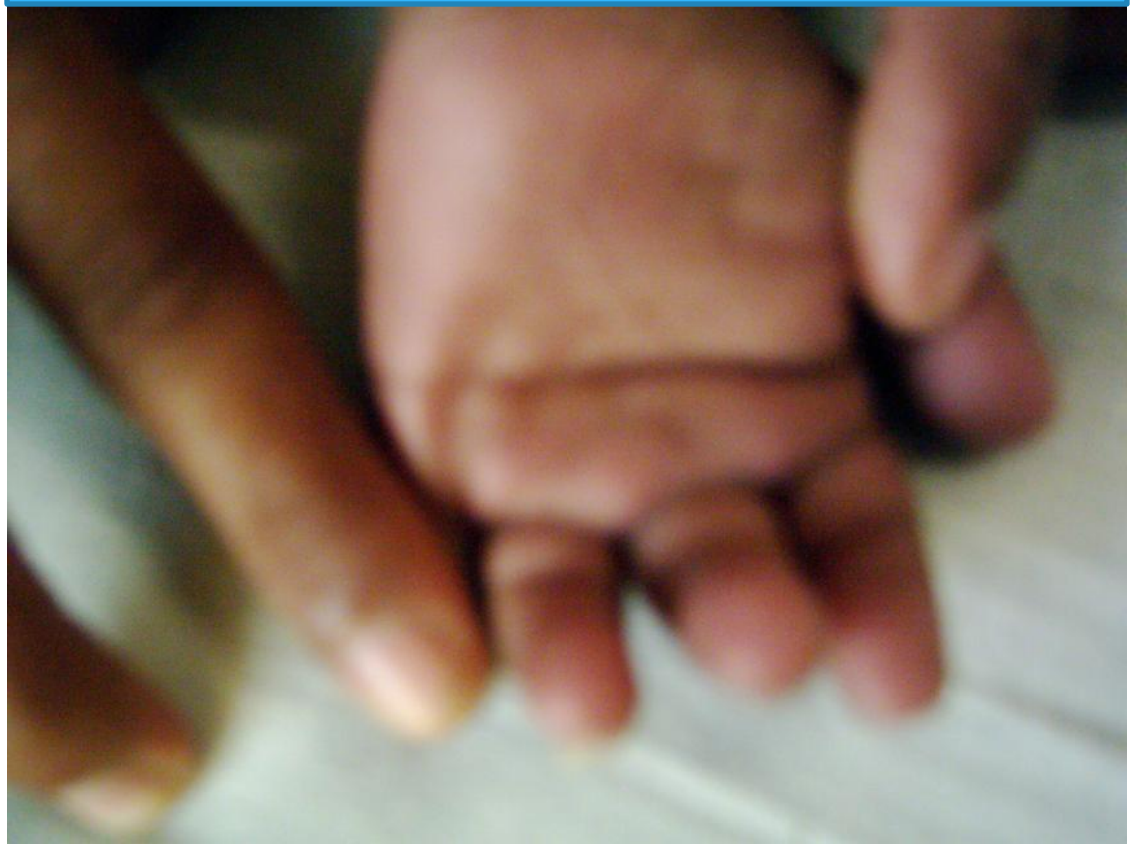
Other Symptoms

- Other symptoms can range from mild to severe.
- People with Down Syndrome have **intellectual and developmental disabilities (IDDs)** which limit intellectual and adaptive behaviors.
- They also may have **delayed language development** and slow motor development.

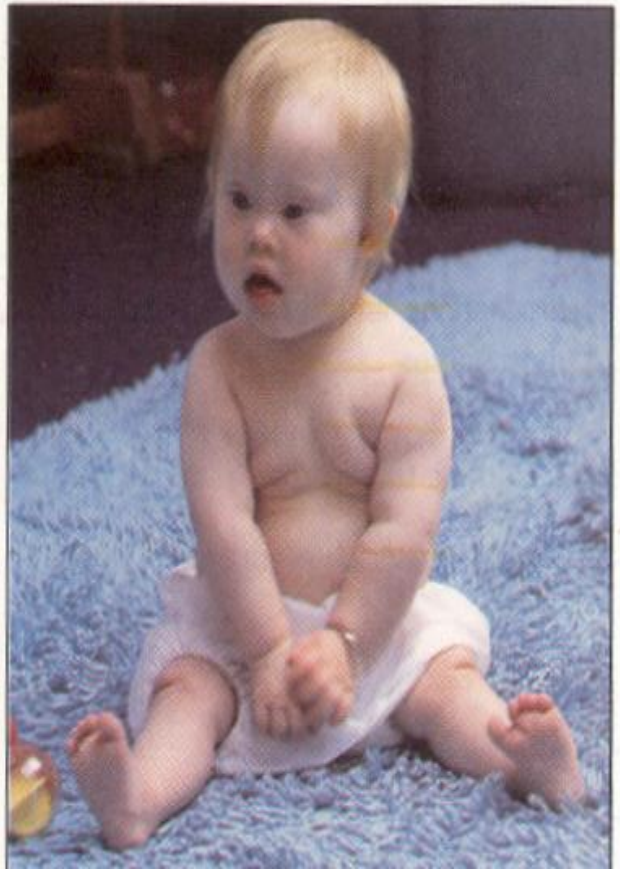
Clinical Findings

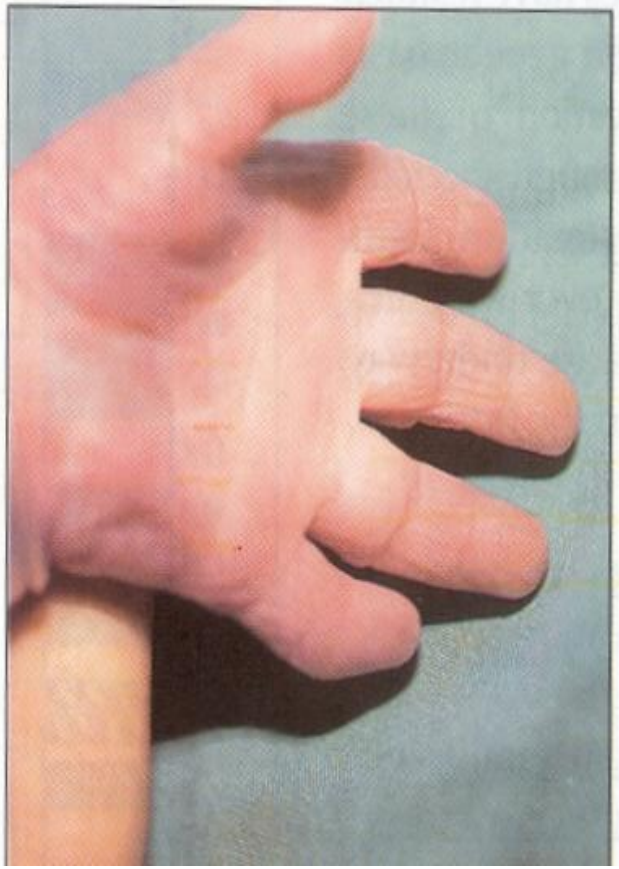
- Brachycephaly
- Upslanted eyes
- 1st & 2nd toe gap
- Loose skin on neck
- Hyperflexibility
- Ear abnormalities
  (Dysplastic ear, an abnormal shape of the ear, narrow canal)
- Protruding tongue and small, narrow palate
- Flat nasal bridge
- Muscular hypotonia
- Epicanthal folds
- Brushfield spots (ring of iris speckles)
- Short fifth finger
- In-curved fifth finger
- Short broad hands
- High arched palate
- Single palmar crease
- Cardiac defect
  (1/2 AV canal defects)
- Duodenal atresia

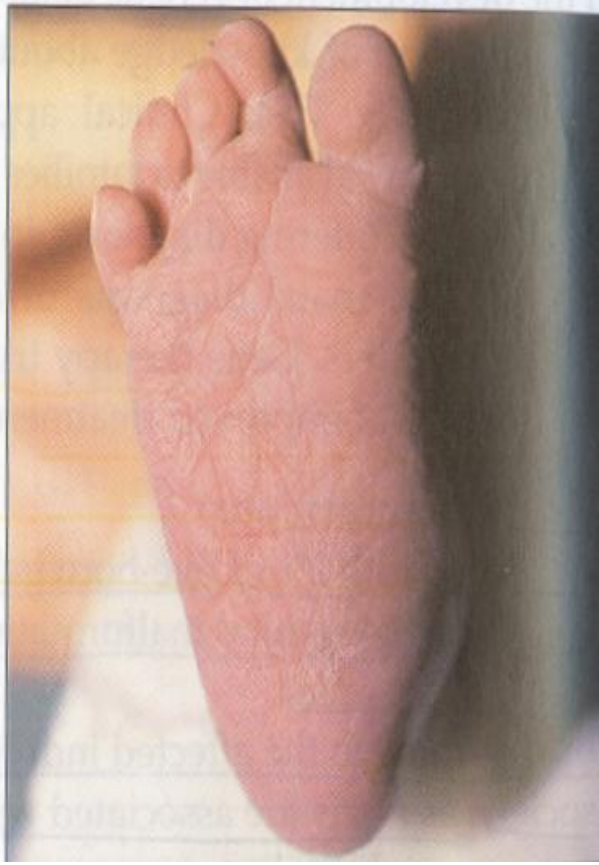
Deep palmar creases

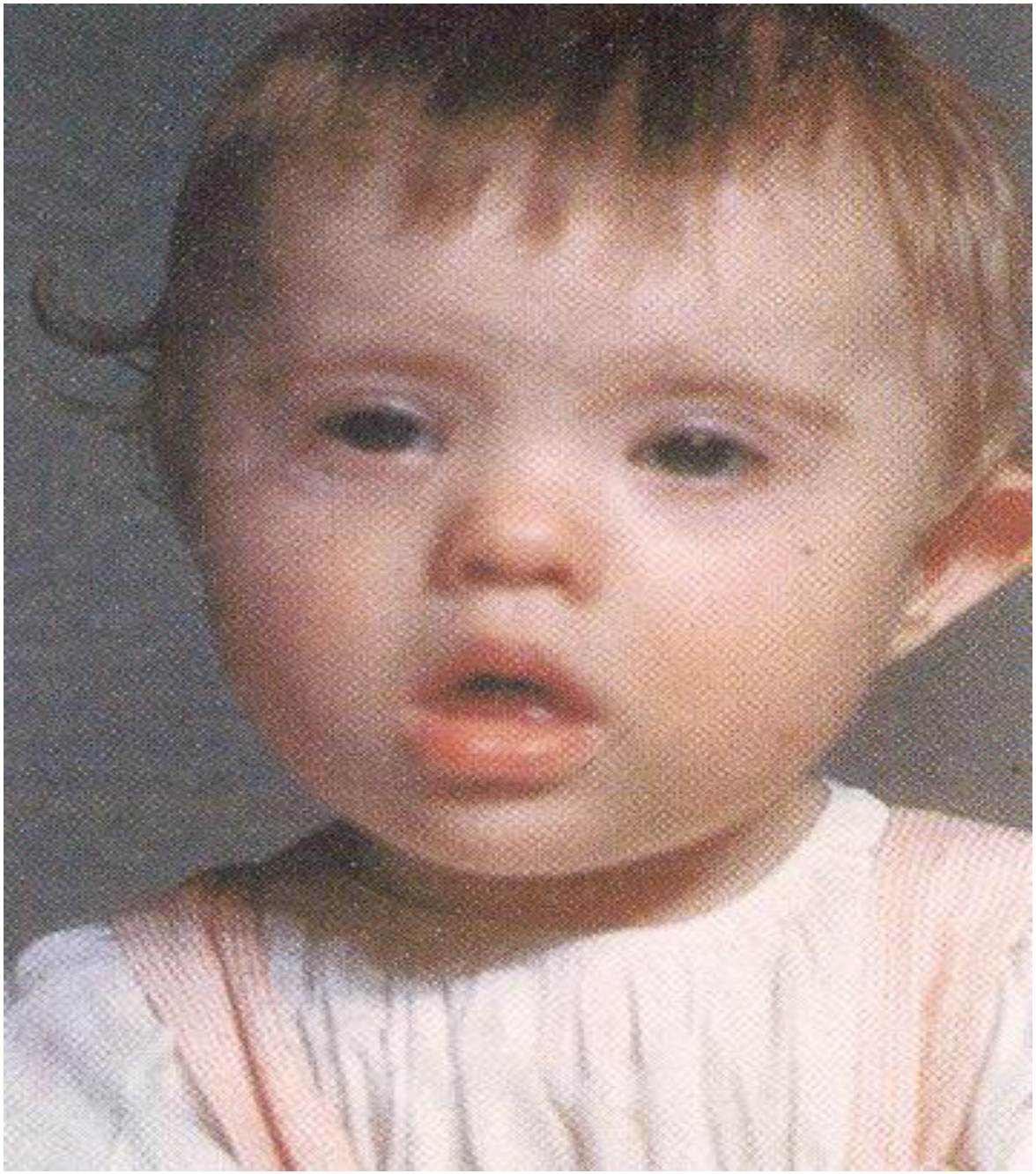


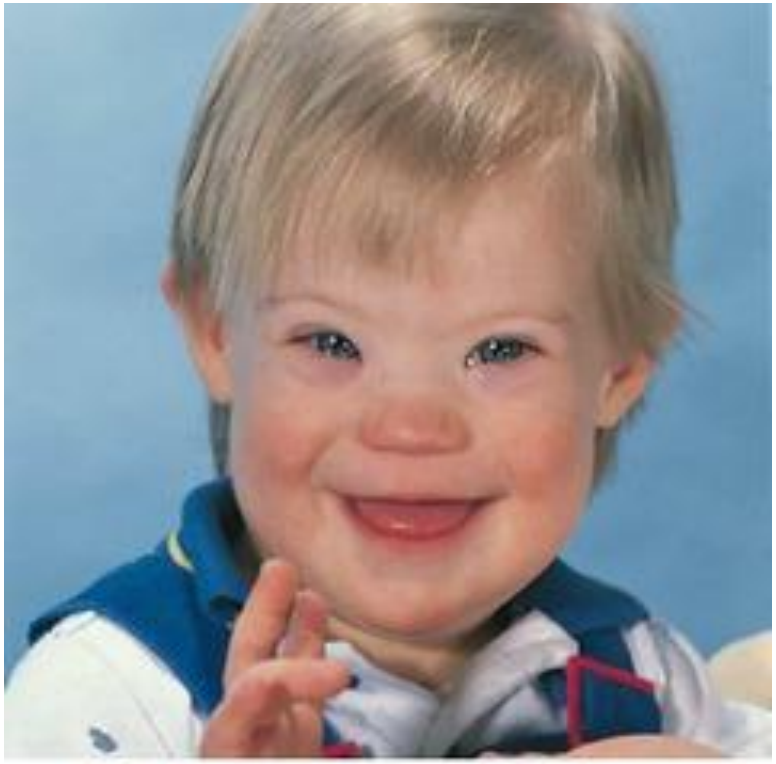
DOWN'S SYNDROME











Hypoplasia

- Head growth-especially AP diameter-results in midface hypoplasia
- Linear growth retardation limbs affected more than trunk esp leg length. Metacarpals and phalanges 10 to 30% shorter
- Tendency to obesity
- Lungs- Enlarged alveoli, reduced number of alveoli (reduction in area of vascular bed)

Musculoskeletal Anomalies

- Retardation of skeletal maturation
- Retardation in growth of cranium
- Narrow palate
- Rib anomalies
- Decreased acetabular and iliac angles (most)
- Instability patello-femoral joint
- Increased incidence mild scoliosis

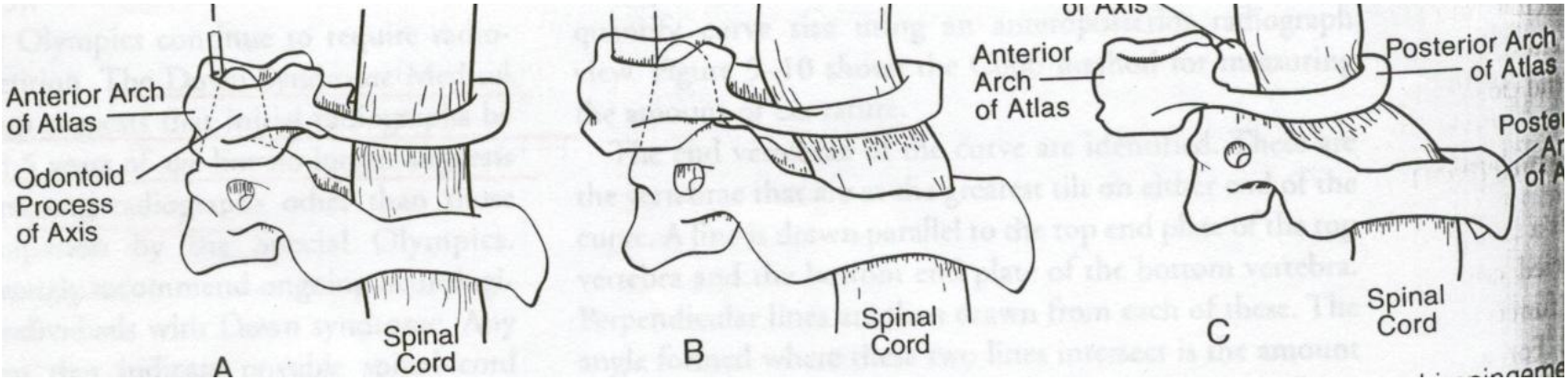
Musculoskeletal anomalies (cont)

- Foot anomalies, pes planus, hindfoot valgus
- Increased incidence of muscles missing (finger and wrist flexors,psoas minor)
- Lack of differentiation of midface musculature
- Hyperflexible joints due to ligamentous laxity (deficit in collagen)

Atlanto-axial instability

Abnormally large space and excessive movement between first and second cervical vertebrae

Cord may become compressed



Atlanto-axial instability -

- 12-20% of children with down syndrome
- estimated 1 to 2% may be symptomatic
  - Abnormal gait
  - Head tilt, limited neck motion, neck pain
  - Pyramidal tract signs (walking difficulties,spasticity, hyperreflexia, incoordination, clumsiness, loss of bladder and or bowel control)

Obesity

- 1/3 of children overweight by age 3
- Suggested that length of trunk relative to limb length and hypoactivity may be contributing factors

Nervous system

- Brain weight reduction
- Dendrites thin,
- Synaptic density in visual cortex 1 to28% lower than typical
- Reduced surface area of synaptic connections in brain
- Abnormal neurons in cerebrum
- delay in myelination in a few cases
- Premature aging 33% mineral deposits,

Development of Movement

- Delay but huge variability.
- Muscle tone, and health have impact on early motor development

Table 23-1. *Variability in motor milestones* [a]

	Average	Range
Sitting alone	10 months	6 to 28 months
Creeping	15 months	9 to 27 months
Standing	20 months	11 to 42 months
Walking	24 months	12 to 65 months

[a] From Pueschel. [38]

Influence of heart disease

- 40% of children with Down syndrome have congenital heart disease
- Surgical advances have improved survival and function
- Moderate to severe heart disease associated with greater motor delays

Table 23-2. *Median Age of Attainment of Gross Motor Skills by Cardiac Status* [a]

	Age (in months) of Attainment	
Gross Motor Skills	**With and Without Mild CHD**	**With Moderate or Severe CHD**
Sitting	9.6	15.0
Reciprocal creeping	19.4	22.3
Standing with support	14.0	21.5
Walking independently	25.0	32.2

[a] From Zausmer and Shea. [40]

Factors in Motor Delay

- Decreased muscle strength and ability to activate musculature
- Ligamentous laxity in some joints
- Latency of response (CNS) slow reaction time
- Deficits in processing /integrating information across modalities (visual, proprioceptive auditory)
- Slow emergence of righting and equilibrium responses
- Hypersensitivity of hands and knees

Early Motor Development

- Often in infancy, performance on mental scales is superior to performance on motor scales
- Infants often can maintain postures but have difficulty with weight- shifting and rotational components of movement that are essential for transitional movements
- Often spend more time supine with a more limited movement experience
- More likely than typical children (who spend time in supine) to have hypersensitivity in hands and feet

Gait

- Flat footed contact rather than heel-toe
- Reduced push off
- Smaller step length
- Increased flex at hip and knee during stance phase
- Calf weakness(delayed change to push off)
- There is greater variability in children with DS

Strength/ endurance

- Studies have found:
- Reduced strength in grip, elbow flex/ext,bicycling, plantarflexion
- regular training improved trunk strength and endurance

Postural control

- Delay in emergence of equilibrium reactions
- postural responses slower and less efficient than typical children in response to moving platform resulting in increased muscle sway

Sensory factors

- Disrupted spatial sense
- Decreased integration of information across modalities;visual/proprioceptive
- Auditory processing consistently more deficient than visual task performance
- Need visual cues for balance longer than typical infants (need for higher level of vestibular input?)
- Improvement with practice
- 78% may have hearing impairment

MANAGEMENT

Physiotherapy Management

- Assessment
  - History: health, progress, current status, caregivers,
  - Observation of spontaneous movement, quality of movement, positioning and handling by parent
  - Postural alignment, resistance to passive movement, strength, range of motion, postural reactions, response to sensory stimuli

Parent education

- Physical handling to promote desired movement or posture
- Reinforce activities parents do, adding visual, vestibular and tactile input
- Add to parent knowledge regarding down syndrome
- Periodic monitoring,

Treatment :improve patterns of posture and movement

- Improve muscle tone
- Improve stability
- Strengthening
- Transitional/rotational movements
- Static and dynamic balance

Prevent abnormal posture and movement

- Weight shifting in prone
- Sitting with narrow base
- Transition to sitting/crawling
- Head and trunk posture

- Facilitate new movement and sensory experiences
- Promote new independent motor and self help skills, safety
- Promote physical activity for play, peer relations and fitness

Studies of Physiotherapy in DS

Table 23-4. *Down Syndrome (DS) Intervention Studies that Included Physical Therapy*

Study	N / Age	Duration / Frequency	Disciplines Intervening / Type of Intervention	Outcome Measure / Outcome
1a. Connolly and Russell (1976) [69]	All DS 40 (variable treatment) 53 (controls) 0-3 y	½-d, 10 wk 3 times/y	Several disciplines Intensive sensori-motor stimulation	Developmental tests Significantly higher scores in treatment group
1b. Connolly et al Follow-up (1980) [72]	20 (treatment) 53 (controls) 3.2-6.3y	. . . . . .	. . . . . .	Developmental tests Intelligence tests Significantly higher scores in treatment group
2. Kantner et al (1976) [70]	3 (normal) 4 (with DS) Randomized treatment and control 6-24 mo	2 wk 10 sessions/d	PT Vestibular stimulation	Inhibition of post-rotary nystagmus 1 child with DS in treatment group approximated normal value
3. Piper and Bless (1980) [74]	All DS 21 (treatment) 16 (controls) $\bar{X}$ age 9.3 mo	6 mo 1 hr biweekly	Several disciplines Transdisciplinary sensori-motor	Griffiths scale No significant difference between groups
4. Harris	All DS	0 wk		

Physiotherapy in Down syndrome

					groups
4.	Harris (1981) [75]	All DS 10 (treatment) 10 (controls) 2.5-21.5 mo	9 wk 40 min 3 times/wk	PT Neurodevelopmental treatment	Bayley, Peabody, treatment objectives No significant difference in developmental tests, treatment group better performance on treatment objectives
5.	Lydic et al (1985) [78]	All DS 9 (treatment) 9 (controls) 4-10 mo	12 wk 3 times/wk	PT Vestibular stimulation	Peabody, MAI No differences between groups
6.	Purdy et al (1987) [80]	5 (with DS) 21-31 mo (single subject design)	28-54 d daily	PT-Parents 2 behavior modification 3 oral motor Rx for tongue protrusion	Tongue posture Some indications of improvement each method
7.	Sellers and Capt (1989)[81]	1 (with DS) 18 mo (single subject)	12 d Use of restraint for specific periods	PT Abduction restraint and NDT	Transitional movements and reciprocal crawling Carry over in prone position to sit and prone pivot

PHYSIOTHERAPY INTERVENTIONS

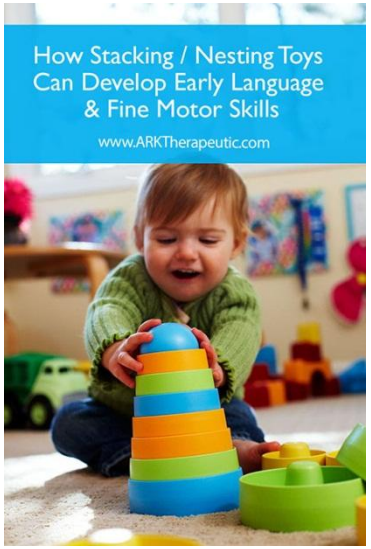
STRENGTH TRAINING

BALANCE TRAINING

PROPERIOCEPTION

GROSS MOTOR DEVELOPMENT

FINE MOTOR TRAINIG



SENSORY INTEGRATION

AEROBICS & DANCE

SUMMARY

- People with Down Syndrome may take certain therapies.
- One type of therapy people with Down Syndrome may take Physical Therapy to avoid poor muscle tone.
- They may also take Speech Therapy to help with speech, and
- Occupational Therapy to help the person learn to perform everyday tasks.
- Children with Down Syndrome may also take special education classes.

COULD THERE BE A CURE?

- Scientists think that they may be able to find a cure for Down Syndrome.
- To find a cure, it is important to pinpoint how Down Syndrome leads to retardation.
- One cure that is being tested is high-potency antioxidants, which seems hopeful.
- Scientists also believe that drugs used to treat Alzheimer's disease may be useful.

DS & EBP

PROGRESSIVE RESISTANCE TRAINING

- The effect of progressive resistance training on leg strength, aerobic capacity and functional tasks of daily living in persons with Down syndrome: RCT 2011

- progressive resistance training is an effective intervention for persons with DS to improve leg strength and stair-climbing ability.

- Read More: http://informahealthcare.com/doi/abs/10.3109/09638288.2011.563820

GMF & HIPPOTHERAPY

- Improving gross motor function and postural control with hippotherapy in children with Down syndrome: Case reports, 2010
- The results indicate that both children improved on many dimensions of the GMFM

- Read More: http://informahealthcare.com/doi/abs/10.3109/09593981003623659

AEROBIC EXERCISE TRAINING

- **Aerobic exercise training programmes for improving physical and psychosocial health in adults with Down syndrome: SYSTEMIC REVIEW (2010)**
- There is **insufficient evidence** to demonstrate that aerobic exercise in adults with Down syndrome improves physical or psychosocial outcomes . Although evidence exists to **support improvements in physiological and psychological aspects** from strategies using mixed physical activity programmes

- http://onlinelibrary.wiley.com/doi/10.1002/14651858.CD005176.pub4/abstract?systemMessage=Wiley+Online+Library+will+be+disrupted+on+the+18th+October+from+10%3A00+BST+%2805%3A00+EDT%29+for+essential+maintenance+for+approximately+two+hours+as+we+make+upgrades+to+improve+our+services+to+you

- **Acute Bouts of Assisted Cycling Improves Cognitive and Upper Extremity Movement Functions in Adolescents With Down Syndrome:** April 2014,
- http://www.aaiddjournals.org/doi/abs/10.1352/1934-9556-52.2.124

- **SPATIAL AND TEMPORAL VARIABILITY OF MOVEMENT PARAMETERS IN INDIVIDUALS WITH DOWN SYNDROME[1]**
- **M. Horvat[1], R. Croce[2], J. Zagrodnik[3], B. Brooks[3] and K. Carter[3]**

- it may be possible to use variable sensory feedback and specific training to modify and adjust movement responses and improve gait performance in Down syndrome.

Read More: http://www.amsciepub.com/doi/abs/10.2466/25.15.26.PMS.114.3.774-782

Thank you
&
Ask???