



Understanding Down Syndrome

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Down Syndrome - Impact on the Family

- **Parents**

- grieve
- guilt and blame
- anger
- depression
- worry
- marital strife
- financial stress

- **Sibs**

- rivalry
- anger
- failure to understand
- embarrassment

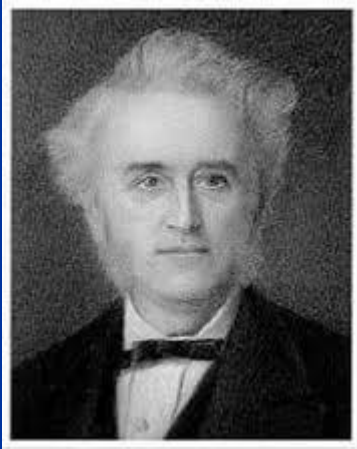
To Parents :

- It is **not** your fault
- It is **not** your child's fault
- **Accept** and love your child
- Have closure, move forward
- Understand what you have to deal with
- Become the **best advocate** for your child
- Nurture and love your family as a whole

Coping Strategies

- **Acceptance**, not avoidance
- Knowledge, not denial
- Self-reliance
- Spouse support and bonding
- Family support and bonding
- Extended family support
- Social support

First Reports



John Langdon Down

London Hospital Reports, 3:259-262, 1866

- described the clinical syndrome
- Down syndrome (Down's syndrome)

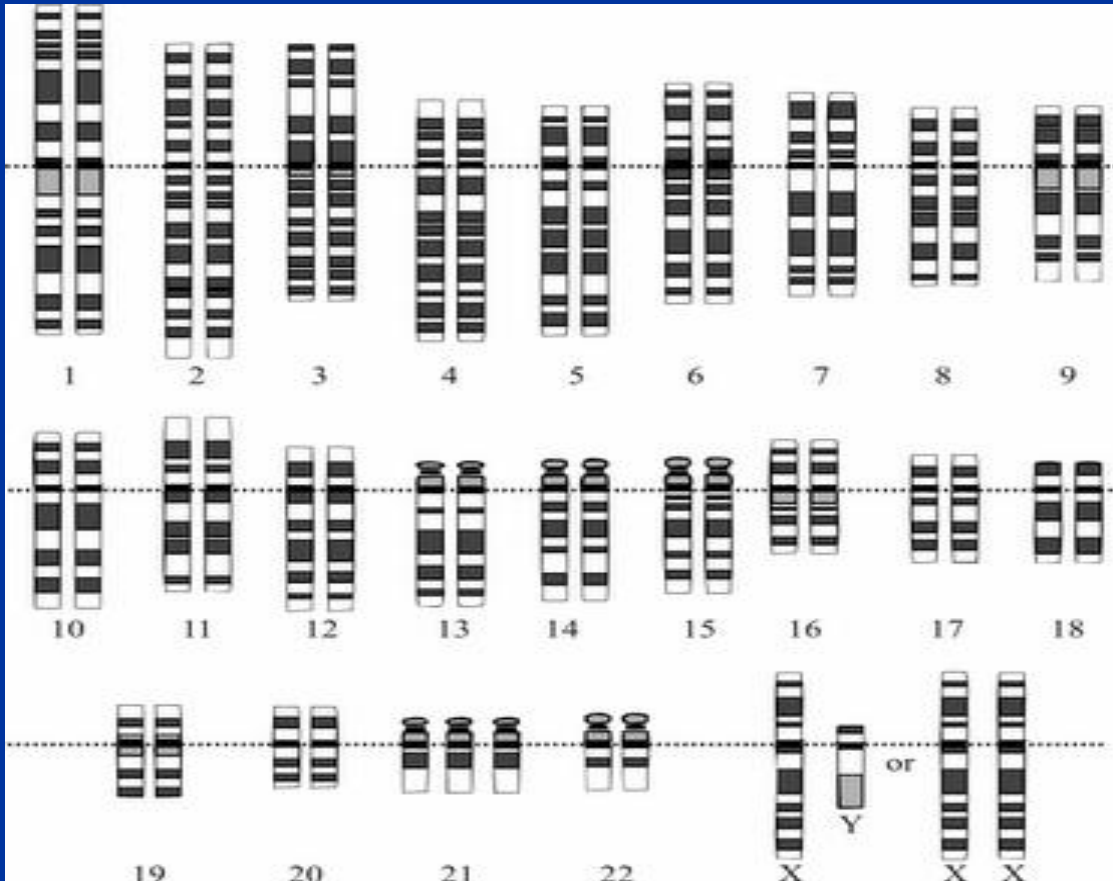


Jerome Lejeune

March 1959

- demonstrated presence of the extra chromosome
- Trisomy 21

Down Syndrome - Basic Genetics



Karyotyping : Trisomy 21

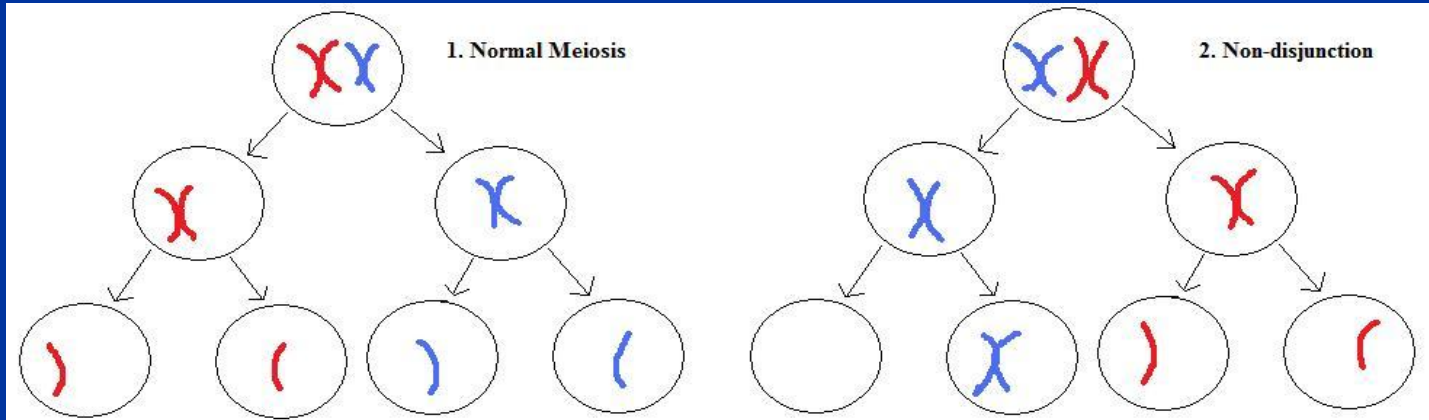
	No Down Syndrome	Down Syndrome
Boy	46 XY	47 XY + 21
Girl	46 XX	47 XX + 21

Genetic Types

- Regular Trisomy 21 (94.6%)
- Translocation Down syndrome (0.7%)
- Mosaic Down syndrome (4.7%)

Ref: Azman et al : Cytogenetic and clinical profile of Down Syndrome in Northeast Malaysia.
Singapore Med J. 2007 Jun;48(6):550-4.

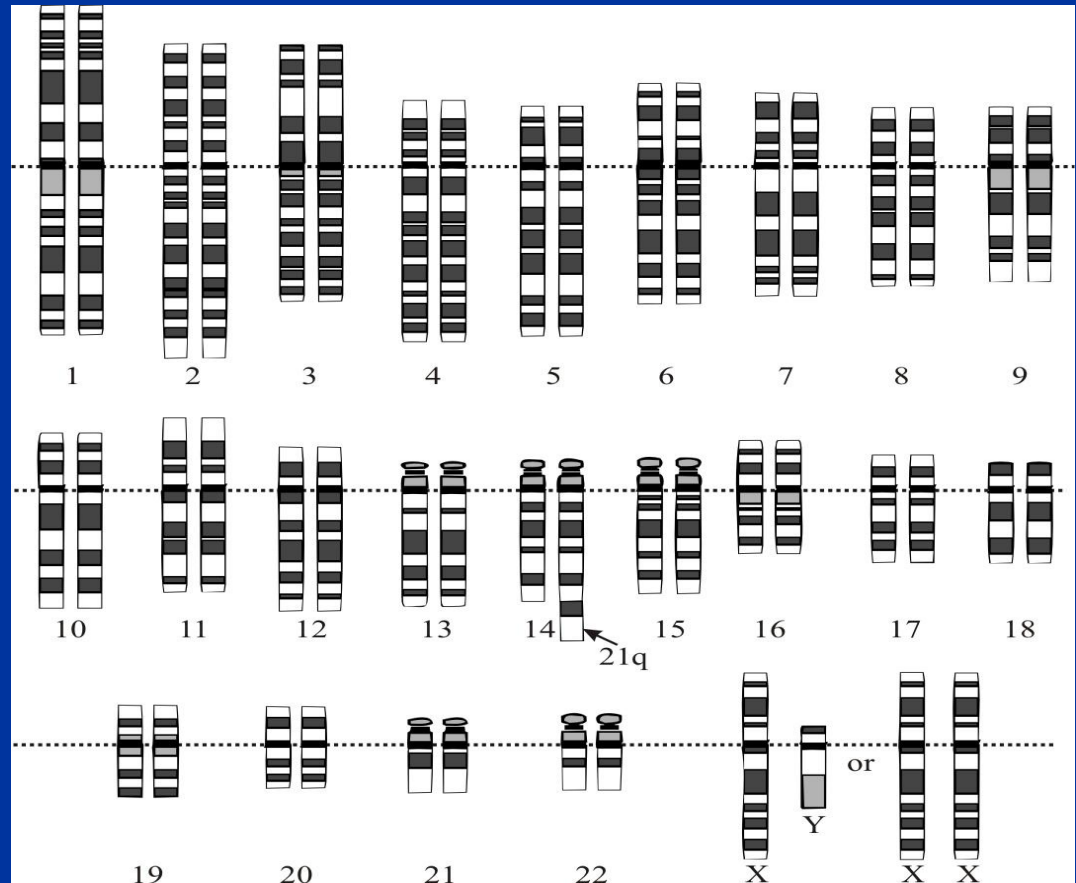
Regular Trisomy 21



- Non-disjunction during meiosis
- Advanced maternal age
- Less often advanced paternal age
- With maternal age of 35 years, risk is 1:385
- 40 years, risk is 1:106
- 45 years, risk is 1:30

Translocation Down Syndrome

- Genetic material of chromosome 21
- Attached to another chromosome
- Can be new occurrence
- Can be inherited



Mosaic Down Syndrome

- Post-zygotic event
- Form 2 cell lines in the individual
- One cell line = trisomy 21
- Other cell line = normal karyotype

Clinical Problems in Down Syndrome

- Congenital malformations
- Medical conditions
- Cognitive impairment
- Behavioural problems

Health Surveillance

Common Medical Problems in Down Syndrome	%
Hearing problems	75
Vision problems	60
Cataracts	15
Refractive errors	50
Obstructive sleep apnea	50-75
Otitis media	50-75
Congenital heart disease	40-50
Delayed dental eruption	23
Seizures	1-13
Hypothyroidism	5
Anemia	3
Iron deficiency	10
Transient myeloproliferative disorder	10
Leukemia	1
Atlantoaxial instability	1-2
Autism	1
GIT disorders (Hirschsprung's disease, duodenal atresia)	5

AAP 2011



Down Syndrome

- Intellectual impairment - variable
 - Mild (IQ 50 to 70)
 - Moderate (IQ 35 to 50)
 - Severe (IQ 20-35)

Developmental Profile in Down Syndrome

- **Slower progress** in all areas of development
- **Uneven profile**
- **Strengths** : social understanding
self-help skills
behaviour
visual processing/visual memory
- **Weaknesses** : motor skills
speech/language skills
learning disability

Down Syndrome - 5 years old

- **Social** : social interaction, can sit and listen to stories, follow teacher's instructions, understand emotions of others
- **Self-help** : feeding, toilet-trained, help in dressing
- **Behaviour** : age appropriate behaviour, not anti-social
- **Motor** : delay in motor skills achievement, may affect self-help skills and fine motor skills, walking and participate in activities

Down Syndrome - 5 years old

- **Speech/language** : understand more than they can say, 2-3 word phrases, difficult to understand their speech, **SIGNING** important bridge to speaking
- **Counting** : start counting, know simple math concepts
- **Reading** : know letters, recognise sight words
- **Memory** : weak verbal short term memory, stronger visual-spatial processing and visual memory – **visual learners**.
- **Thinking** : weak thinking, problem-solving and reasoning

Down Syndrome - 12 years old

- **Social**
 - personal identity
 - self esteem, self confidence
 - friends
- **Self-care**
 - independent self care in home
 - independent in school
 - independent in the community
- **Cognitive/Academic**
 - improved speech,
 - language, literacy and numeracy skills
 - expanding knowledge

Children with Down Syndrome

- Aims and Goals

- As *independent* as possible
- Develop *positive identity*, self confidence, self esteem
- Develop *network* of friends, personal relationships, leisure interests
- *Progress* in cognitive, speech and language, academic skills
- Prepare for *work/employment*

Down Syndrome

- Effective Strategies

- **Early intervention**
- Inclusive school education *
- Independence
- Employment

Remember :

“Use strengths to reduce challenges”

Children with Down Syndrome Meeting Needs – The First Years

- **Health needs**
 - Medical problems
 - Hearing difficulties
 - Visual impairments
 - Hypotonia
- **Developmental needs**
 - Delay with uneven profile
 - Gross motor
 - Fine motor
 - Personal-social
- **Health visits**
 - Physiotherapy
 - Occupational therapy
 - Speech therapy

The Child with Down Syndrome

- Speech and Language

- Reduce effects of **hearing problems**
- Encourage **interaction**
- **Communicate** through all aspects of daily life
- Interactive **play and games**
- Encourage **attention, listening and looking**
- Allow **imitating**
- Advantages of **signing** (bridge to spoken language)
 - Hold baby's attention
 - Sign gives added cue to meaning of words
 - Stress the words, speak slowly
 - Not a substitute for spoken words

The Child with Down Syndrome

- Reading and Writing

- Introduce reading early
- *“Teach reading to teach talking”*
- **Sight words** first
- Phonics later

The Child with Down Syndrome

- Numbers and Math

- Make opportunities for child to learn numbers and counting in the home
- Songs
- Games
- Fun
- **Visual learning** strengths

The Child with Down Syndrome

- Motor Development

- Encourage **gross motor activities**
 - rolling, sitting, standing, walking
- Encourage **fine motor activities**
 - reaching, grasping
- **Basic everyday skills**
- **Recreational skills**

The Child with Down Syndrome

- Social Development

- Social understanding, empathy
- Encourage **social interaction**
- *“Bring child along”*

The Child with Down Syndrome

- Memory Development

- Long term > Short term memory
- Visual-spatial > Verbal short term memory
- **Visual memory training**
- *“Reading for memory”*

Children with Down Syndrome Meeting Needs – The Older Years

- Health needs
 - medical problems
- Developmental needs
 - delay
 - disabilities
 - language
 - personal-social
 - behavioural
- **Independence**
- **Life skills**
- **Planning support**

To Parents...

- See child's strengths, abilities
- Understand child's special needs
- Help child develop and learn
- Develop support systems
- Know child's rights
- Advocate effectively for your child
- Gain access to desired services/activities in the community for your child

Reference: Bailey DB et al Journal of Early Intervention. 2006;28:227.251

What children with Down syndrome tell us...

- We are individuals
- Do not stereotype us
- Sure, I may have some areas of special needs, but I have strengths as well
- I need to be loved as you love your other children
- I am not defined by Down syndrome, I am ME
- I wish to live a full life

Thank You