Pre-Operative Services Teaching Rounds 14 April 2011

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Down's Syndrome

- History
- Genetics
- Incidence
- Clinical features
- Anesthesia implications

Dental procedures

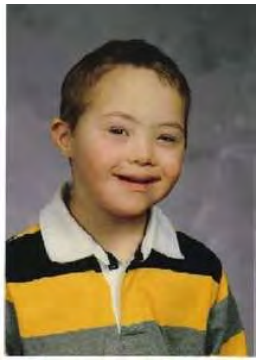
- Positioning
- Anesthesia
- Airway management

CASE

- 29 yr old Down's syndrome for dental rehab under GA
- Typical Down's features
- VSD
- No previous anesthesia

DOWN'S SYNDROME - HISTORY

- John Langdon Down, a British physician, first described the syndrome in an article in 1866.
- Mongol
  - synonym used by Dr Down, referring to work of Johann Friedrich Blumenbach on ethnic classes because of facial similarity to Mongoloid class
- A letter to the Lancet in 1961 by 18 geneticists saying that *Mongolian idiocy* had "misleading connotations," and had become "an embarrassing term," and should be changed. *The Lancet* supported *Down's Syndrome*.
- WHO accepted this in 1965, as requested by Mongolian delegate
- Down's syndrome is the accepted medical term today



- The cause was not identified till 1959 when Jerome Lejeune in France discovered trisomy 21.
- Found in all ethnicities

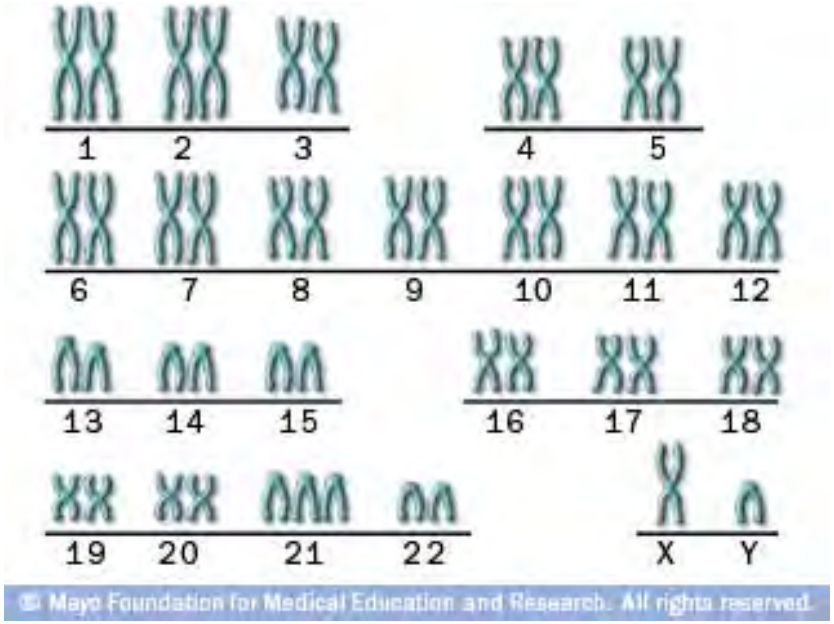
Down's equivalent found in animals – well described in mice and chimps

GENETICS

Usual karyotype:

- 23 pairs of chromosomes
  - 22 paired
  - XX or XY sex chromosomes

Down's: additional chromosome



GENETICS

- **Trisomy 21.**
  - > 90%
  - three copies of chromosome 21 in all cells
  - abnormal cell division during the development of the sperm cell or the egg cell.
- **Mosaic Down syndrome.**
  - Rare
  - some cells with an extra copy of chromosome 21.
  - abnormal cell division after fertilization.
- **Translocation Down syndrome.**
  - uncommon
  - part of chromosome 21 becomes attached (translocated) onto another chromosome, before or at conception.
  - two copies of chromosome 21, with additional material from chromosome 21 stuck to the translocated chromosome.

No known environmental trigger for abnormality

INCIDENCE

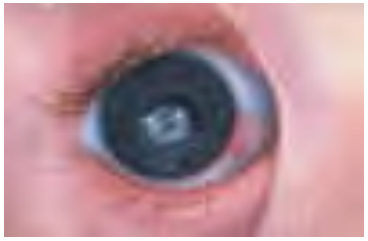
- Most common chromosomal disorder
- 1 of every 600–800 births
- 5500 born /yr in the US
- Advanced maternal age is implicated
  - 1-4% after age 40
  - although 80% born to younger women as majority of babies are born to younger women
- Paternal age recently implicated
- Life expectancy – 49 years (used to be 10 years)

CLINICAL FEATURES

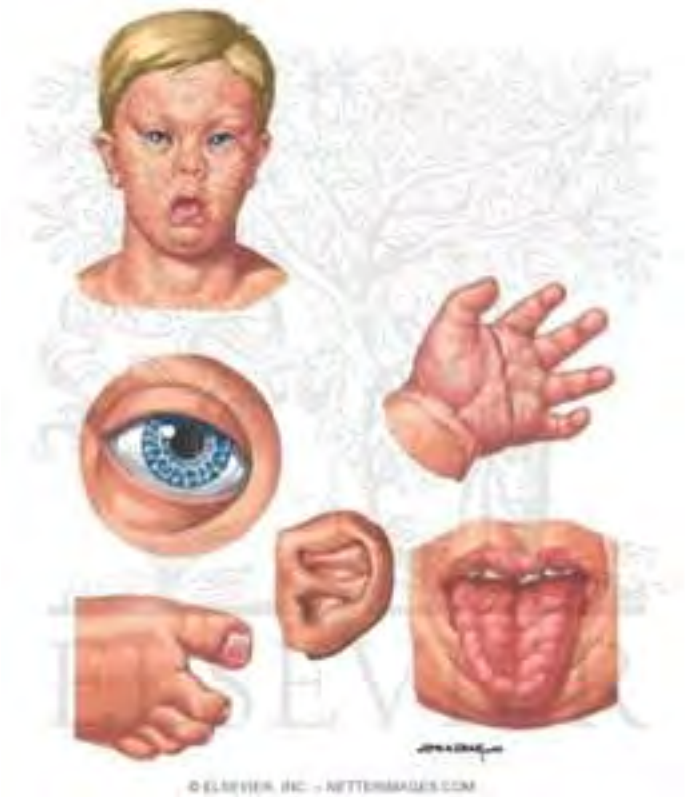
- Mental retardation
  - IQ is below 65 (average 50)
  - Delayed fine motor skills
  - Delayed gross motor skills
  - Language delay
- Hypotonia
- Excessive flexibility
  - upper cervical spine instability produced by ligamentous laxity, skeletal anomalies, or both
  - can result in neurologic impairment, including quadriplegia

CLINICAL FEATURES

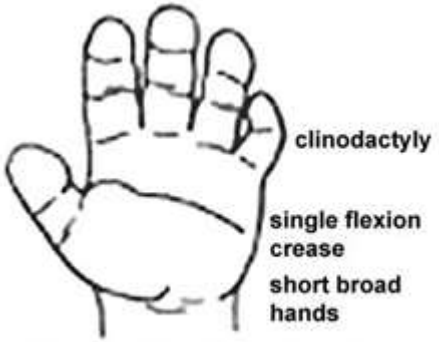
- Craniofacial abnormalities
  - Brachycephaly
  - Flattened facial features /Flat nose
  - Large Protruding tongue
  - small chin
  - upslanting palpebral fissures
  - Prominent epicanthic folds
  - Unusually shaped ears



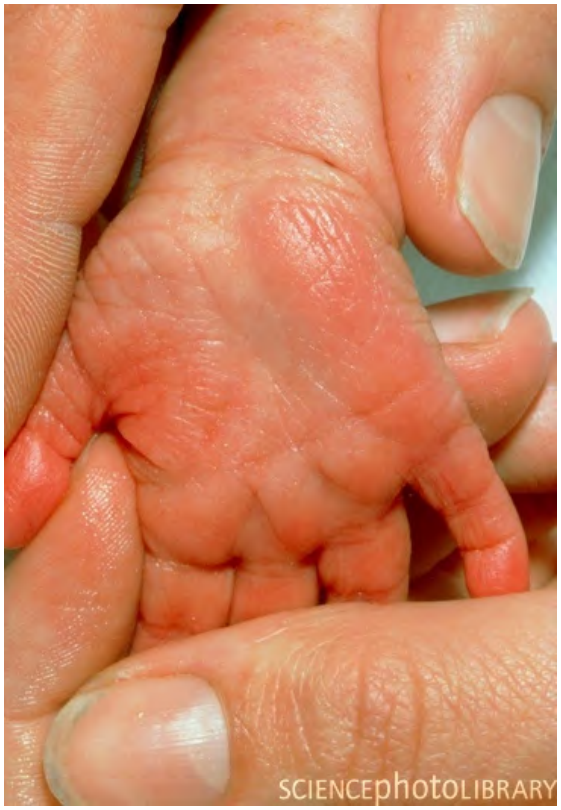
CLINICAL FEATURES

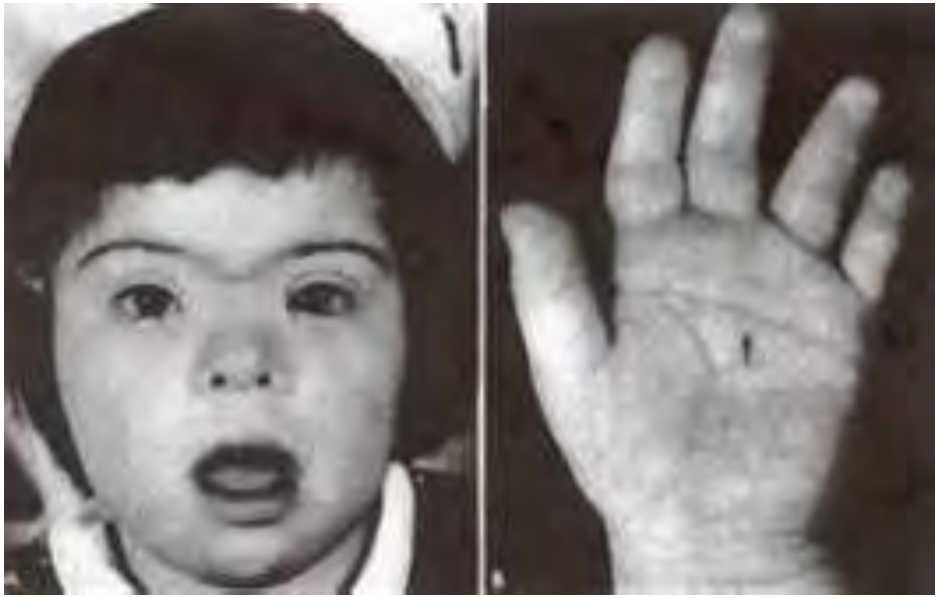


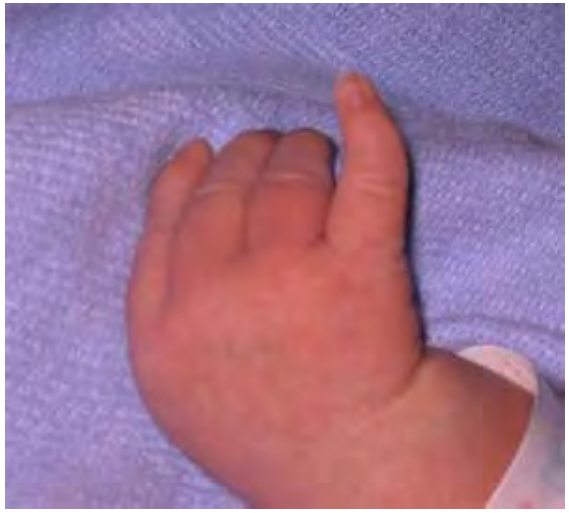
- Upper airway
  - Macroglossia
  - Tonsillar hypertrophy
  - High arched palate
  - Narrow trachea for age
  - Small teeth
- Broad, short hands with a single palmar crease
- Clinodactyly of 5th finger, hypoplasia middle phalanx
- Relatively short fingers
- Short limbs
- Infants born with Down syndrome may be of average size, but typically they grow slowly and remain shorter



Trisomy 21 - Hand Features







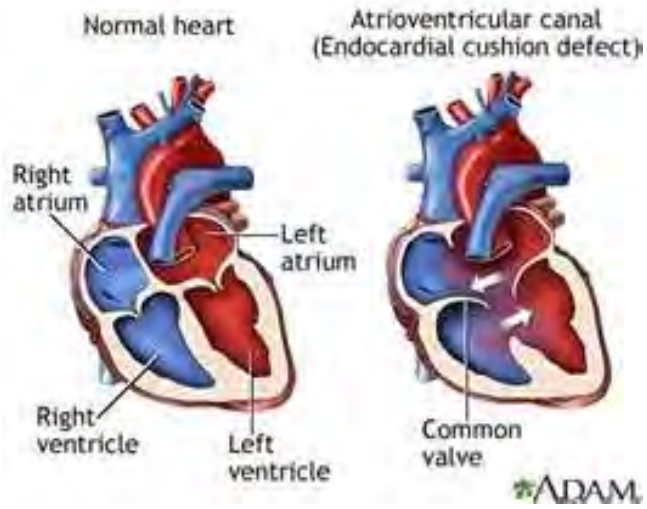
CLINICAL FEATURES

Cardiovascular

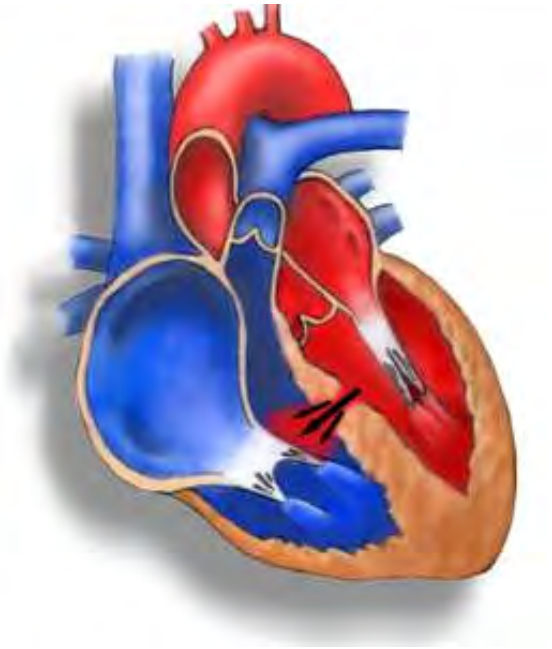
- congenital heart disease (50%),
  - Endocardial cushion defects (AV canal) (32-40%)
  - Ventricular septal defect (30-40%)
  - Tetralogy of Fallot /ASD / PDA
  - Cyanosis/Eisenmenger's syndrome
  - Pulmonary hypertension
- Lower risk of atherosclerosis

COMMON CARDIAC LESIONS

Endocardial cushion defects



Ventricular septal defect



CLINICAL FEATURES

Gastrointestinal findings:

Congenital:

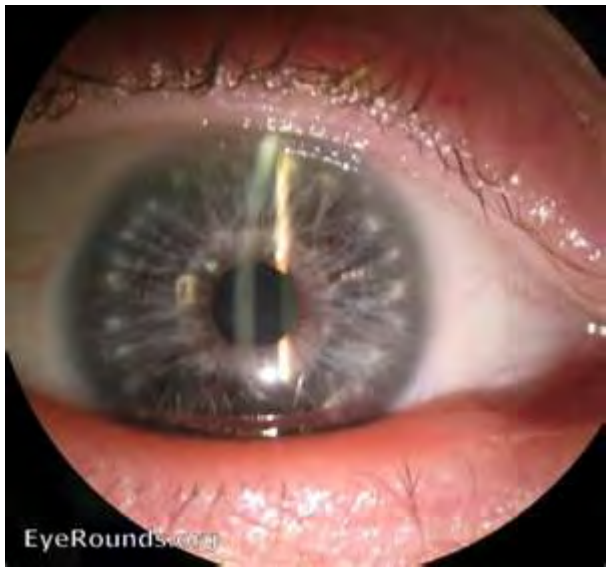
- Imperforate anus
- Trache-oesophageal fistula
- Duodenal atresia
- Hirschprung's

Acquired:

- GERD
- Celiac disease
- Constipation

CLINICAL FEATURES

- Seizure disorder (2-9%)
- Psychiatric
  - Early dementia (age 40)
  - Depression
  - OCD
  - ADD/autism
- Eye lesions
  - Strabismus
  - Cataracts
  - Glaucoma
  - Refractive errors
  - Brushfield spots (small white/grey/brown spots found on iris periphery)

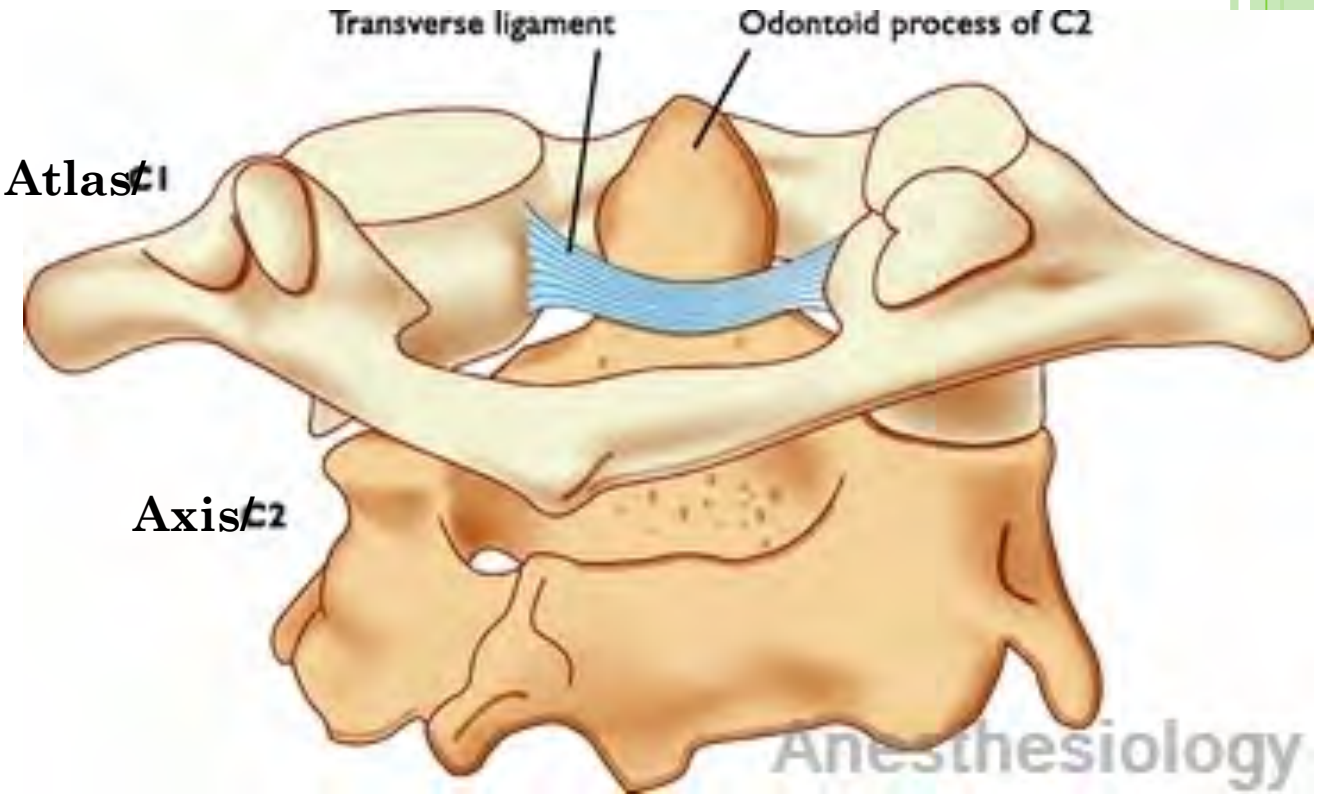


CLINICAL FEATURES

- Sleep apnea
- Obesity
- Thyroid
- Infertility males>females
- Tumors:
  - Leukemia
  - testicular
  - solid tumours are less common – increased tumor suppressor cells
- Immunocompromise
- Respiratory and ear infections

ANESTHESIA IMPLICATIONS

- Airway:
  - Difficult – anatomy:
    - Tongue
    - Small chin
    - C-spine



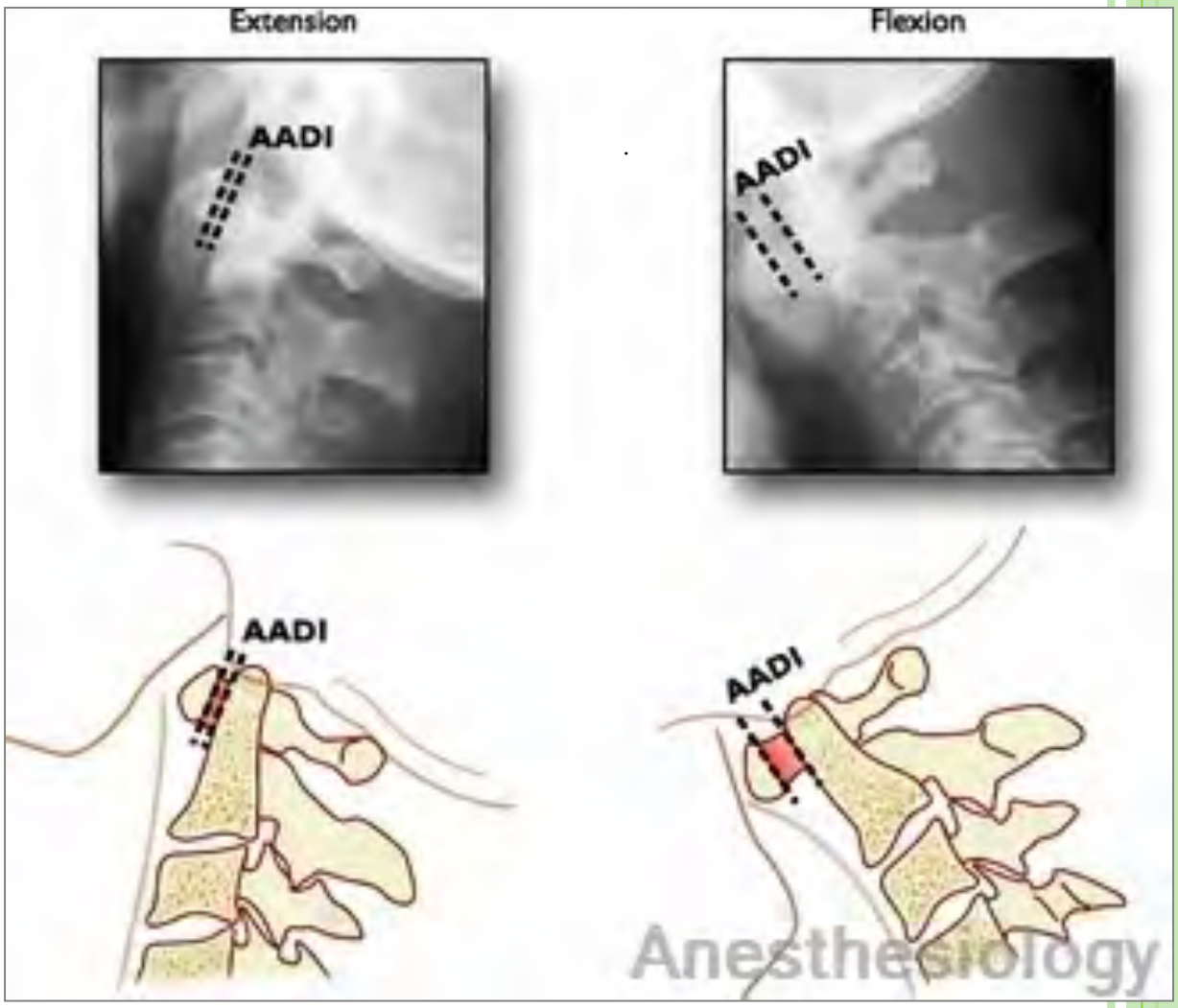
C-spine

Atlanto-axial instability (15%)

Laxity of transverse ligament

AADI greater than 4–5 mm in any lateral view is abnormal.

Natural progression
- can become unstable (3-24%)
- can stabilize (less common)



Atlantoaxial instability. Note the increased anterior atlantodental interval (AADI) with flexion and the corresponding decrease in the space between the posterior aspect of odontoid and the posterior arch of C1 (through which the spinal cord passes).

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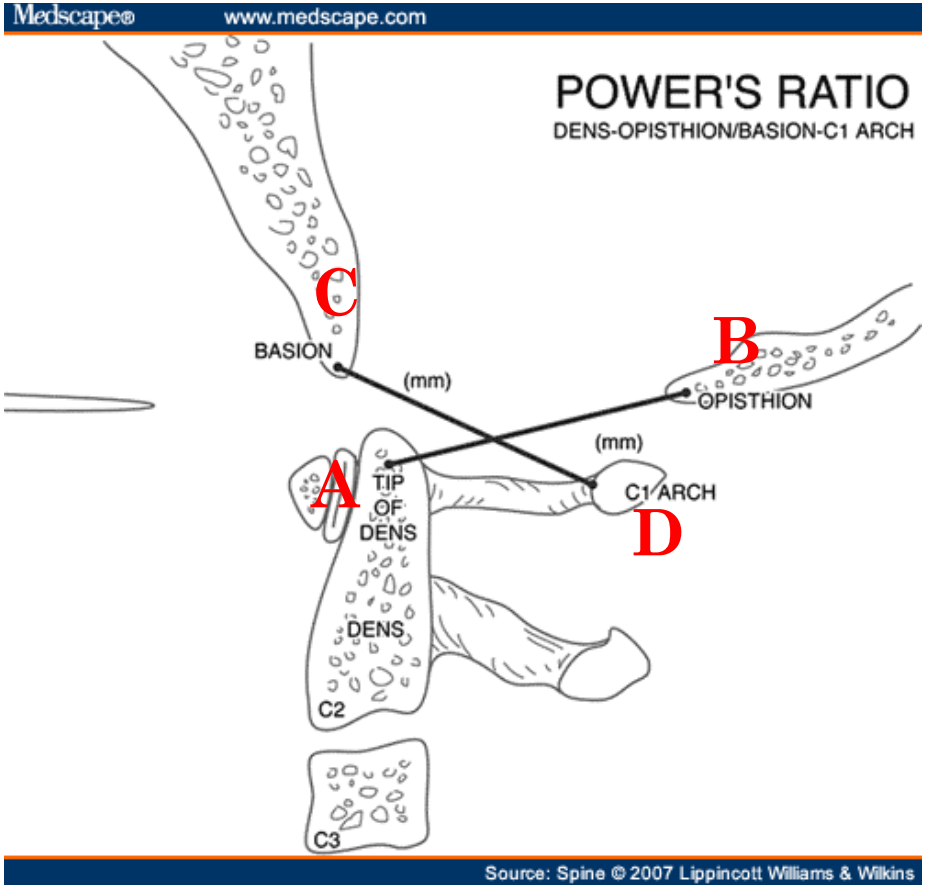
Atlanto-axial instability:

Greater subluxation when the neck is **flexed**, with normalization during extension.

But **extension**, particularly with "lifting" of the skull and C1 that occurs during laryngoscopy, may result in movement

Rotation of the head may also result in C1–C2 subluxation



Atlanto-occipital instability (9-20%)

Powers ratio used to diagnosis occipitocervical dislocation

Powers ratio = C-D/A-B

C-D:
distance from basion to posterior arch

A-B:
distance from anterior arch to opisthion

Ratio ~ 1 is normal
if > 1.0 concern for anterior dislocation ratio
< 1.0 raises concern for posterior atlanto-occipital dislocation (odontoid fractures
ring of atlas fractures)

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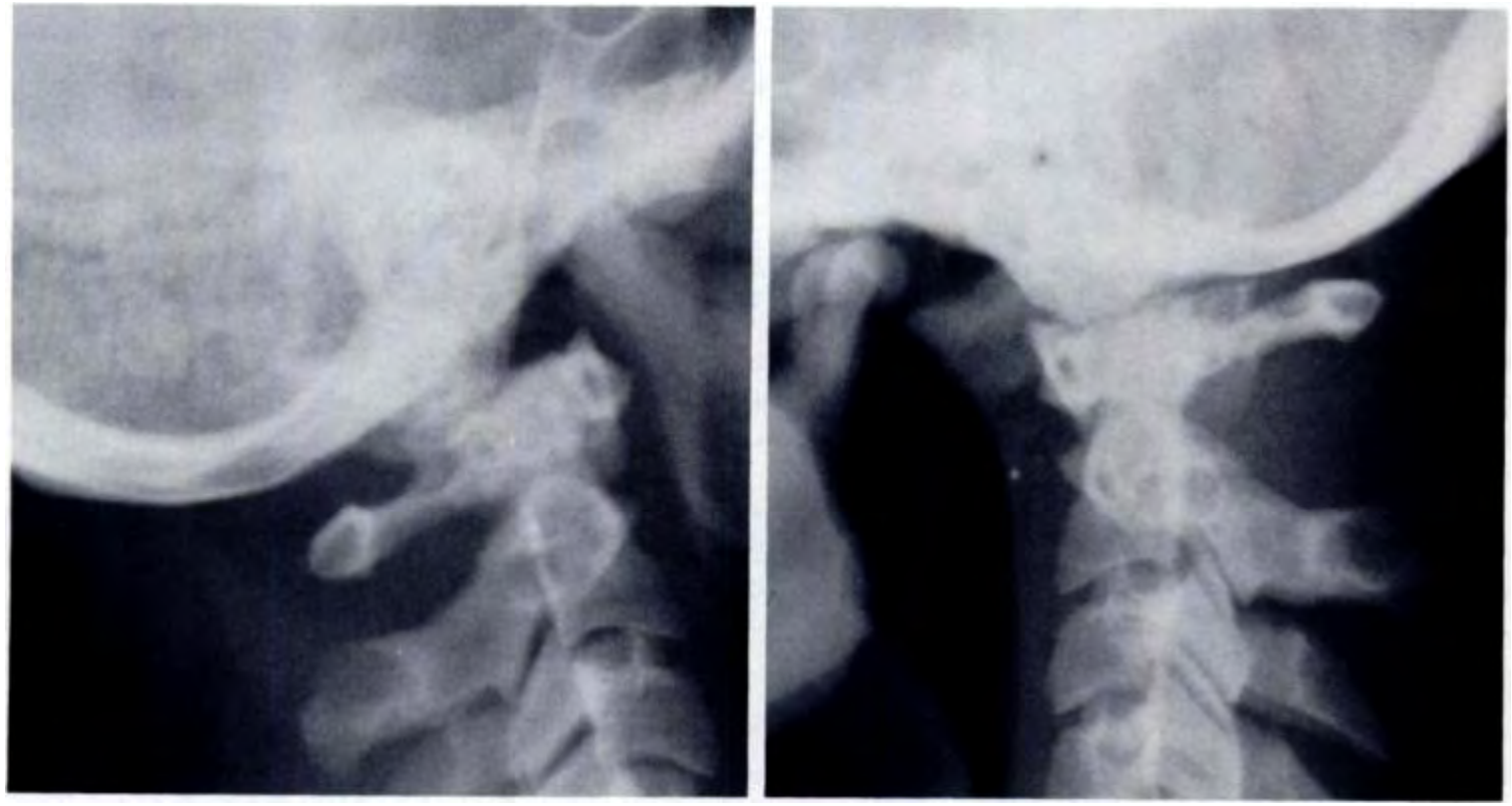
A  B

Fig. **1**

A: Lateral radiograph in extension shows normal position of occiput on C-1 with basion directly over odontoid.

B: Flexion results in 1.2 cm forward movement of occiput along relatively flat articular surfaces of C-1 .

C-SPINE

- cervical spinal stenosis
- only 1-2% will become of these 3 neck c-spine abnormalities are symptomatic.

  symptoms include
  - neck pain
  - torticollis
  - weakness / gait abnormality
  - quadriplegia
  - clonus

HINTS AS TO NEW SPINAL CORD ABNORMALITY

History:

Has the patient's behavior changed (refusal to participate in usual activities)?

Ability to ambulate worsened?

Fine motor function decreased?

Any change in bladder or bowel function?

Any pain in the head or neck area?

Does the patient refuse or seem unable to turn his or her head?

Have there been any episodes of dizziness or syncope?

Physical examination:

Abnormal range of motion

Neck tenderness

Gait disturbances

Weakness

Spasticity

Abnormal reflexes or clonus

American Academy of Pediatrics recommends:

- screening once between ages 3-5
  especially if they are involved in contact sports.
- lateral flexion/extension c-spine Xray
- Controversial - not endorsed by Special Olympics
- Flexion recommended first while patient is cooperative (more important view)
- Specifically review:
  - C1/C2
  - Atlanto-occipital
  - Size of neural canal
- CT/MRI more sensitive but cost is high, may need anesthesia
  - Used for abnormal Xray or symptoms

Literature review :

- no evidence-based practical guidelines
- risk of spinal cord injury during anesthesia –unknown
- very few anesthesia complications in Down's

Intraoperatively – stabilize head for procedures

e.g. For BMTs – strap head and patient to table and tilt whole table laterally.

Ethical discussion regarding c-spine fixation for minor procedure under GA

Risks/benefits and burdens

ANESTHESIA IMPLICATIONS (CONT)

Easy desaturations intra- and post-

- Airway
  - macroglossia,
  - hypotonia of pharyngeal muscles
  - enlarged T's and A's
- Respiratory infections
- Cardiac
- OSA
- Obesity
- ETT that is 1-2 sizes smaller (subglottic stenosis and for nasal ETT – smaller nares with midface hypoplasia)

- Postoperative stridor more common

Cardiac

- Uncorrected congenital defects beyond the scope of this talk
- History of effort tolerance
  - Cyanotic spells
  - Squatting
  - Ability to drink bottle without cyanosis/stopping/sweating
- Exam
  - Clubbing
  - Cyanosis
  - Murmurs
  - Loud P2
  - Edema – sacral/calves
- Measure oxygen saturation

- Air embolus
- SBE prophylaxis

SBE PROPHYLAXIS (AHA 4/2007) *NEED TO HAVE BOTH LESION AND SURGERY*

Heart lesion

- -Prosthetic heart valves,
- -A prior history of IE.
- -Unrepaired cyanotic congenital heart disease, including palliative shunts and conduits.
- -Completely repaired congenital heart defects with prosthetic material or device, during the first six months after the procedure.
- -Repaired congenital heart disease with residual defects
- -Cardiac valvulopathy in a transplanted heart.

Surgery type

- all dental procedures that involve manipulation of either gingival tissue or the periapical region of teeth
- perforation of the oral mucosa. (T's A's etc

- **IV access – obesity and xerodermia**
- **?** exaggerated response to atropine, with tachycardia and cycloplegia.
- ?altered concentrations of neurotransmitters, such as serotonin, dopamine, norepinephrine and acetylcholine. Altered sensitivity to sedatives and narcotics.
- **Labs:**
  - **C-spine lat flex/extend Xray**
  - **Recent ECG and oxygen saturation if heart disease**
  - **CBC if pale**
  - **as per our guidelines**

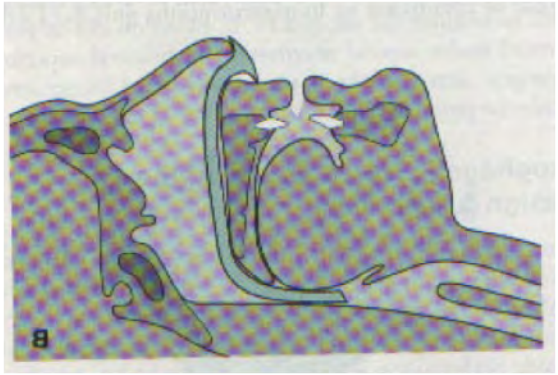
DENTAL PROCEDURES

- Positioning – supine
- Shared airway
  - C- spine
  - Aspiration risk

ANESTHESIA

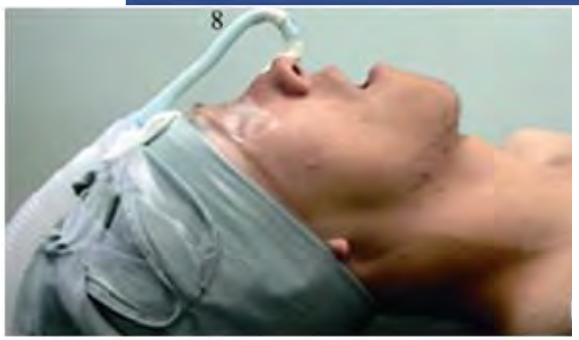
- Endotracheal tube
  - Oral
  - Nasal
- Throat pack – wet (saline)
  - Sore throat
- Arrhythmias common
- ASC candidate – cooperation/airway

Nasal trumpet



Oral Rae

Nasal Rae



Magill's Forceps

RECOMMENDATIONS

C-spine Xray

- Any age
- Flexion first
- Especially if surgeon involved in head and neck

Repeat Xray

- New signs and symptoms
- If first done before age 3
- Prior abnormality
- Extreme positioning in surgery