Rett Syndrome

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Risk

- Occurs almost exclusively in females.
- Incidence is 0.4–0.7:10,000.

Perioperative Risks

- Abnormal control of ventilation, with periods of apnea and hyperventilation
- May have GE reflux
- Multiple orthopedic and motor movement disorders

Worry About

- Risk of periop apnea not known
- Risk of succinylcholine-induced hyperkalemia not known
- Aspiration due to GE reflux and swallowing disorder
- Cardiac: Prolonged QTc, abnormal autonomic regulation, increased incidence of sudden death
- Difficult intraop positioning because of spasticity and contractures

Overview

- Characterized by normal early growth and development followed by a slowing of development and then regression characterized by loss of purposeful use of the hands, distinctive hand movements, slowed brain and head growth, problems with walking, seizures, and intellectual disability
- Dx based on clinical characteristics with inclusion and exclusion criteria, mutations in *MECP2* gene
- Abnormal EEG; nonspecific changes
- Pathognomonic stereotyped hand movements, tortuous hand-wringing or other hand automatisms
- Seizures very common
- Respirations abnormal when awake; hyperventilation alternating with hypoventilation or apnea and hypoxemia
- Orthopedic and movement disorders such as scoliosis, spasticity, ataxia, loss of locomotion
- ANS dysfunction with increased sympathetic tone
- Cachexia

Etiology

- Mutations in the *MECP2* gene' mechanism not yet determined.
- MECP2 is needed for brain development and acts as one of the many biochemical switches in gene expression.
- Although genetic, most cases occur spontaneously.
- Dx made by Hx and clinical features (inclusion and exclusion criteria established).

Usual Treatment

- Supportive only; no specific therapy
- Aimed at improving quality of life, seizure control, nutrition, PT, possible surgery for orthopedic problems

Assessment Points

System	Effect	Assessment by Hx	PE	Test
HEENT	Nonspecific Spasticity may make airway difficult		Neck ROM Airway exam	Neck x-rays if indicated
CV	Possible prolonged QTc Peripheral vasomotor disturbances		Extremities cool Trophic changes	ECG
RESP	Abn control of ventilation when awake, with hyperventilation, apnea, cyanosis Lung changes due to scoliosis or aspiration	Apnea, cyanosis Scoliosis, aspiration	Observation Chest exam	O_2 saturation CXR
GI	GE reflux possible, swallowing difficulties, constipation Growth failure	GE reflux, feeding difficulties	Thin, small for age	Studies for GE reflux
CNS	Severe developmental delay Seizures Ataxia, loss of locomotion	Developmental level, seizure activity	Assessment of cognitive and movement disorders	EEG
MS	Hypotonia (early), spasticity (late), ataxia Secondary orthopedic manifestations: Scoliosis, joint contractures	Progress and extent of MS abnormality	Chest exam for scoliosis Limb and joint positions	X-rays

Key References: Acampa M, Guideri F: Cardiac disease and Rett syndrome, *Arch Dis Child* 91(5):440–443, 2006; Coleman P: Rett syndrome: anaesthesia management, *Paediatr Anaesth* 13(2):180, 2003.

Perioperative Implications

Preoperative Preparation

- As for any pt with developmental delay.
- Optimize respiratory status.
- Assess respiratory control.
- Minimize aspiration risk.

Monitoring

- Routine.
- More invasive depending on procedure.
- Consider brain function monitoring because of anesthetic sensitivity.

Airway

- Normal face.
- Spasticity may make positioning difficult.

Preinduction/Induction

- Risk of hyperkalemia following succinylcholine unknown
- Possible aspiration risk due to GE reflux

Maintenance

- Respiratory control abnormal; unknown if spontaneous ventilation under anesthesia associated with significant apnea
- Attention to body temp because of thin body habitus and peripheral vasomotor disturbances
- Can be excessively sensitive to both sedative drugs and volatile anesthetics

Extubation

- Possible aspiration risk.
- Assess respiratory control.

Postoperative Period

- Respiratory control abnormal.
  - Effect of anesthetic agents.
  - Duration of respiratory monitoring.
  - Effect of narcotics versus local anesthetics for pain control.
- Intense monitoring in postop period is essential as frequent desaturations in these pts may cause progressive cerebral damage.

Adjuvants

- None

Anticipated Problems/Concerns

- Respiration control abnormality is not well understood. Therefore effect of anesthetic agents intraop and postop on respiration is not known. Need for postop monitoring for apnea is unknown.

Reye Syndrome

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Risk

- Incidence prior to 1990: 0.3–0.6:100,000.
- From 1987 to 1993: 0.03-0.06:100,000; 2 cases/y have been reported since 1994.
- During early 1980s, an association between aspirin and Reye syndrome was recognized; thereafter, incidence declined dramatically. In 1986, a warning label on all aspirin-containing products was mandated in USA.

Perioperative Risks

- Surgery (all but life-or-death emergencies) contraindicated during Reye syndrome.
- Following recovery, LFTs must be repeated.