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.

Worry About

- Unrecognized inborn errors of metabolism that produce Reye-like syndromes, such as fatty-acid oxidation defects, carnitine deficiency, and amino and organic acidopathies
- Recurrent liver dysfunction
- Permanent neurologic sequelae

Overview

- An acute, noninflammatory encephalopathy with hepatic dysfunction predominantly in children; typically starts several days after a viral illness, usually influenza or varicella.
- Encephalopathy heralded by protracted severe vomiting, with abnormal behavior and combativeness that may progress to coma.
- Dx is made by unexplained encephalopathy with one or more of following: Serum transaminases elevated to at least 3 times normal, blood ammonia levels at least 3 times normal, or hepatic microvesicular fatty infiltration on liver biopsy. There should be no other reasonable explanation for the cerebral or hepatic abnormalities.
- The CDC use a classification for progressive disease severity of stages 1–6. Mortality has decreased from 50% to 20% as a result of recognition of early phases and aggressive treatment.
- Prognosis depends on severity and duration of cerebral dysfunction. Severe disease can lead to subtle neuropsychological defects.

Etiology

- Abnormal reaction to viral illness modified by exogenous toxin in a susceptible host.
- Most frequently linked to influenza A and B and varicella. The exogenous toxin is aspirin in most cases. Salicylates were detectable in >80% of cases.

Usual Treatment

- Early recognition of mild cases and maintenance of fluid, lyte, acid-base, urine output and glucose balance.
- ET intubation may be required to ensure airway protection and control of ventilation to reduce ICP.
- Fluids restricted in pts with cerebral edema; ICP monitoring helps to improve cerebral perfusion pressure and manage ICP.
- Mannitol to induce cerebral dehydration and barbiturates to decrease cerebral metabolic demand.
- Coagulopathies treated with vitamin K and/or FFP.

Assessment Points

System	Effect	Assessment by Hx	PE	Test
GI	Hepatic dysfunction	Severe vomiting	Hepatomegaly	Hepatic transaminases, ammonia levels Liver biopsy PT, PTT
CNS	Delirium Combative behavior Seizures Lethargy Coma	Alteration in mental status	No focal signs	CT scan ICP monitor

Key References: Goetz CG: Aminoacidopathies and organic acidopathies, mitochondrial enzyme defects, and other metabolic errors. In Goetz C, editor: *Textbook of clinical neurology*, ed 3, Philadelphia, PA, 2007, Saunders; Tasker RC: Update on pediatric neurocritical care, *Paediatr Anaesth* 24(7):717–723, 2014.

Perioperative Implications

- Surgery should not be undertaken except in life-or-death emergencies.

Adjuvants

- Ondansetron to decrease vomiting.
- Treat seizures with phenytoin.
- Correct hyperammonemia with sodium phenyl-acetate/sodium benzoate.

Anticipated Problems/Concerns

- Hepatic dysfunction
- Neurologic sequelae
- Underlying inborn errors of metabolism, particularly in children under 5 y

Rheumatic Fever (Acute) and Rheumatic Heart Disease

Mark S. Weiss | Victoria M. Bedell | Roger A. Moore

Risk

- Common illness among children and young adults.
- Primary chronic sequelae is RHD.
- Worldwide estimate is over 15 million cases of RHD, with 282,000 new cases and 233,000 deaths annually.
- Up to 1% of all school-age children in Africa, Asia, Latin America, and the eastern Mediterranean show signs of the disease.

Perioperative Risks

- In ARF with acute cardiac manifestations (including first-degree heart block and pericarditis), medications and equipment for maintaining heart rhythm and function during anesthesia should be available.
- An actively febrile pt's surgery should be delayed unless it is urgent or emergent.
- The approach to anesthesia in RHD must be tailored to the pt's specific physiologic parameters. Control of the pt's hemodynamic profile to optimize cardiovascular stability will depend on which valves are damaged and the extent of the myocardial compromise, either from RHD or from secondary cardiovascular effects due to chronic valvular disease.
- In the presence of valvular disease, prophylactic antibiotics should be given to prevent bacterial endocarditis.
- Periop assessment of cardiac status, including direct and indirect effects of chronic valvular disease on cardiac function, must be performed prior to surgery. Pts may be clinically asymptomatic for 20 y after developing RHD owing to compensatory alterations in cardiac structure and function. Knowledge of the cardiac compensatory changes in heart function is essential.

Worry About

- If valvular damage is present, maintain tight control of cardiac rate and rhythm, pulmonary and systemic vascular resistance, and intravascular fluid volume.
- If pulm hypertensive crisis occurs, hyperventilate and increase inspired O_2 to 100%.

Overview

- ARF is primarily due to a pharyngeal infection with *Streptococcus pyogenes* or group A beta hemolytic streptococcus, which is a common cause of throat infections in children. If left untreated, the child can develop ARF, which is an inflammatory response occurring 2–3 wk after the initial infection.
- RHD is an autoimmune reaction with cardiac tissue, resulting in permanent deformities of heart valves or chordae tendineae.
- Scarring leads to valvular stenosis, classically in the mitral valve followed by the aortic valve. However, all cardiac valvular defects can occur.
- Joint pain and carditis with valve damage are the major clinical manifestations of ARF. Carditis can occur in up to 80% of people with ARF, leading to mitral or aortic valvular disease. About half of those affected will develop chronic RHD. Death from ARF is not common, but chronic rheumatic heart disease can lead to morbidity from arrhythmias, endocarditis, and stroke.

Usual Treatment

- ARF is easily treated with penicillin if initiated within 9 d of onset of pharyngitis; this will prevent the development of chronic manifestations, such as carditis. Corticosteroids can be used in the acute inflammatory stage. Bed rest for several wk is required, followed by gentle ambulation. Diuretics and vasodilators are given for severe carditis as well as furosemide for mild to moderate CHF. ACE inhibitors are given for severe aortic regurgitation. Serial ECHO will determine cardiac dimensions and function. Valve repair surgery should be delayed until after the acute inflammatory stage.
- Chorea: Treatment is supportive, including psychosocial support.
- Arthritis and arthralgias: NSAIDs may mask signs of acute rheumatic fever. By clinical consensus, paracetamol is preferred. After Dx has been made, naproxen twice daily is the drug of choice.