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Hereditary Multiple Intestinal Atresia

Intestinal atresia is the most common cause of congenital bowel obstruction in newborns with an incidence of one in 1500 live births. Approximately one third of all cases of neonatal bowel obstruction are due to intestinal atresia which can be sporadic most commonly or hereditary with a possible autosomal recessive mode of inheritance. Intestinal atresia are caused by an intrauterine mesenteric vascular accident occurring after the embryonic stage where blood fails to irrigate some segment of bowel causing a membrane (Type I), fibrous cord (Type II) or with complete disappearance of a substantial length of the intestine. The proportion of children with multiple jejunoileal atresia varies from 6-32%, with hereditary multiple intestinal atresia (HMIA) first reported in 1956 as the rarest form of multiple bowel atresia. HMIA is an unusual form of intestinal atresia with possible autosomal recessive mode of inheritance. Immune defects have been described in several patients with various types of familial bowel atresia. The combination of HMIA and immunodeficiency is invariably fatal. The immune deficiency affects T- and B-cell functions with lymphopenia, agammaglobulinemia and impaired mitogen responses. HMIA maintains a 100% lethality rate from continued postoperative intestinal failure and an associated severe immunodeficiency that has been increasingly recognized with this disorder. The association of HMIA with immunodeficiency affect multiple organs such as intestine, thymus, lungs, spleen, and liver. Newborns born with HMIA show symptoms of intestinal obstruction at birth and radiopaque shadows on abdominal plain films. Presenting symptoms include bilious vomiting, abdominal distension, and failure to pass meconium. The abdominal films show signs of gastric or duodenal atresia such as single or double bubble combined with typical large rounded or oval homogenous calcifications in the abdominal cavity. The excessive dilatation of the stomach, the presence of intraluminal calcifications and the conformation of rectal atresia by contrast enema is considered pathognomonic of HMIA. The intraoperative findings demonstrate widespread multiple atresia exclusively type I and II extending from the stomach to the rectum. Multiple webs, both occlusive and non-occlusive, and atretic cords are found throughout the small intestine. No mesenteric defect as in Type IIIa or IIIb is identified. There is necrotic nonbilious calcified material within the lumen of the bowel and the intestinal mucosa appeared atrophic. The entire or part of the colon except for the ileocecal valve is a continuation of string-like solid fibrous cord extending to the distal rectum. There could be cystic dilatation of the bile ducts in some cases with both complete pyloric and duodenal or proximal jejunal atresia. The pathogenesis is still speculative though a combined immunodeficiency should be excluded. A fatal outcome occurs in most cases. Antenatally polyhydramnios is the presenting feature of HMIA in 20-35% of cases and is more frequently associated with proximal bowel

obstruction. Prenatal ultrasound can raise suspicion of HMIA in the presence of polyhydramnios, gastric dilatation, intraluminal calcifications and thickened echogenic wall. Fetal MRI findings particularly those of simultaneous pyloric and intestinal obstructions with numerous dilated bowel loops suggest the diagnosis. The aim of surgical intervention is to restore continuity of the gastrointestinal tract and maintain maximum length of viable bowel. An autosomal recessive transmission has been proposed as a probable explanation of this disease. Through whole exome sequencing of patients with HMIA, two mutations in a single gene, the tetratricopeptide repeat domain 7A gene TTC7A could explain the disease in the affected cases. Mutation in TTC7A is often associated with severe intestinal defects and severe combined immunodeficiency and inflammatory bowel disease. HMIA associated with a TTC7A mutational defect is characterized by multiorgan impairments. After operative repair babies with HMIA continue to have poor gastrointestinal function even if enough intestinal length is preserved to avoid the classic short gut syndrome. The outcome of HMIA depends upon the length and regions of bowel involved. The degree of postoperative short bowel syndrome is a major determinant of survival. The most common cause of death is infection related to pneumonia, peritonitis, and sepsis. The family should be aware of the dimmed prognosis.

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Commotio Cordis

Commotio Cordis (CC) is a life-threatening dysrhythmia produce by a direct nonpenetrating low impact blow to the chest. It is defined as sudden cardiac arrest in the absence of apparent structural heart disease after nonpenetrating chest injury. Most cases of commotio cordis occurs in young patients, specifically boys who are younger that 16 years, with almost three-fourth of cases occurring after some type of sport participation such as baseball, hockey, lacrosse, or softball. The other third occurs out of sport activities including intentional chest blows due to fighting, child abuse, or impact from snowballs or hollow plastic toys. In infants commotio cordis occurs in the setting of child abuse. CC is one of the most common causes of sudden cardiac death in athletes, with hypertrophic cardiomyopathy

being the most common. CC is characterized as a sudden disturbance of cardiac rhythm in the absence of demonstrable signs of significant mechanical injury to the heart induce by a direct blow to the chest. A structural injury is not a contributing aspect to the pathogenesis of the often-lethal rhythm disturbance seen in CC. In terms of the rhythm disturbance created, CC most commonly is limited to the sudden onset of ventricular fibrillation, though it may also manifest as other cardiac rhythm disturbances such as heart block, ventricular tachycardia, bundle branch block, ST-T wave abnormalities or asystole. It has been demonstrated experimentally that blow over the center of the heart are more likely to cause a ventricular fibrillation than at any other location and is associated to peak elevations of the left ventricular pressure. Coronary vasospasm may also play a role in some cases of CC. The occurrence of CC is thought to require the rare confluence of a blow over the heart and precise timing during the vulnerable phase of repolarization (10-30 milliseconds prior to the peak of the T-wave). The sudden blow causes a dramatic increase in left ventricular intracavitary pressure resulting in increased stretching of cell membranes and activation of an increase potassium ion concentration current across the cell membranes of the myocytes. Higher energy impacts are more likely to cause ventricular fibrillation compared to low velocity impacts. Collapse is instantaneous or within a few seconds. Early resuscitation seems to be the most important predictor for successful recovery including beginning therapy with standard cardiopulmonary resuscitation protocols. The rapid use of an automated defibrillator is the optimal approach and should be used wherever possible before transport to a health care facility. Early initiation of CPR, followed by early defibrillation in less than 4 to 8 minutes from time of collapse has the greatest impact on successful resuscitation. Survival rates has increased steadily over the years and recent survival is now greater than 65%. Any child who has experienced syncope during sport activities should not be allowed to return to the game. If an automated external defibrillator was utilized for resuscitation of a CC event, the computer disc storage system should be secured and studied to document the rhythm disturbance at the time of the shock. Once the child is transported to the hospital several therapeutic maneuvers are critical and related to the length of the cardiac arrest. Mortality and risk of hypoxic permanent brain injury is high during commotio cordis events. Cardiac reassessment and evaluation is recommended before the patients resumes sports activities. To assess the degree of injury continuous cardiac monitoring, ventilatory support, chest films and electrocardiogram with echocardiogram should be performed. Blood chemistry including levels of calcium, magnesium, and cardiac enzymes (serial troponin and creatine kinase) should be obtained. ICU monitoring with cardiac telemetry should be performed in the first 24 hours and subsequently as needed. Syncope can mimic CC but is usually not associated with impact to the chest wall. With CC the possibility of recurrence is so small that some physicians recommend return to participation after exercise stress test has normalized. CC is a survivable phenomenon. As a measure of prophylaxis, schools, universities, and municipal parks have implemented automated external defibrillators program where staff is trained in their use and one or more devices are placed in areas accessible to any athletic event. CC prognosis depends on the rapid identification of the event and availability of cardiac defibrillators in site.

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Priapism

Priapism refers to a prolonged full or partial penile erection which last more than four hours not related to sexual activity or stimulus. Priapism is a urological emergency, and the treatment of priapism aims to avoid penile disfigurement or shortening, erectile dysfunction and psychological sequelae. Priapism of the clitoris can occur and is called clitorism. Though rare, priapism does occur in the pediatric age. There are three widely accepted types of priapism: 1) ischemic priapism which is a low-flow veno-occlusive disease caused by blocked venous outflow that prevents oxygenated arterial blood from perfusing the corpora cavernosa; 2) stuttering priapism which refers to intermittent and recurrent ischemic, and 3) non-ischemic due to high flow arterial etiology. A fourth type is that which occurs in neonates. Ischemic priapism is the most common type seen in children, typically painful. Sexual activity and nocturnal erections are precipitators. The corpora cavernosa is markedly rigid, while the glans and corpora spongiosa is flaccid. A compartment syndrome within the tunica albuginea occurs due to elevated interstitial pressure causing microvascular compromise and ischemia. The lack of arterial inflow creates a hypoxic environment that damages the smooth muscle tissue leading to irreversible necrosis and fibrosis. Stuttering priapism is a recurrent unwanted painful erection, often self-limited, but may precede an ischemic priapism. It is trigger by nocturnal erections and is associated with sickle cell disease. Stuttering priapism causes recurrent visits to the emergency department, sleep deprivation, embarrassment, and sexual performance anxiety. Each episode carries a risk of fibrotic damage to the corpora cavernosa if untreated. The non-ischemic priapism variety is a partial erection due to unregulated cavernous arterial inflow which is usually painless. Piesis sign consisting of perineal compression that results in penile detumescence but recurs after removal of the pressure strongly suggest non-ischemic priapism in children. A history of blunt trauma to the penis or an iatrogenic needle injury is most commonly described etiology. The result is a disruption of the cavernous arterial anatomy creating an arteriolar-sinusoidal fistula. This type might not require emergency urological treatment. Neonatal priapism is a prolonged erection lasting more than four hours during the first month of life, it usually occurs in the first few days of life and might persist for 2-12 days with an average of 5 days. The incidence of priapism is almost one

for every 100,000 males per year, most frequently during the fifth decade of life. Sickle cell disease (SCD) is the most common cause of priapism in children with most cases of the stuttering variety. Mean episode is at age 15. The tumescence of priapism is initiated by relaxation of the cavernous arteries and sinusoidal smooth muscle. This increases the arterial inflow and capacitance. Sinusoids trap blood, tumescence occurs, and the tunica albuginea is stretched occluding emissary veins. Contraction of the ischiocavernous muscle increases the cavernosal pressure exceeding systolic blood pressure. Nitric oxide synthetase is produced increasing nitric oxide and acting on both cavernosa artery and sinusoidal smooth muscle elevating c GMP promoting smooth relaxation. In cases of SCD deoxygenated hemoglobin S causes sickling and microvascular obstruction stimulating hemolysis and increasing free hemoglobin levels which deactivate nitric oxide causing ischemic priapism. Nocturnal erections, sexual activity, dehydration, fever, and exposure to cold are the most common precipitants of priapism in children with SCD. Childhood leukemias can also cause priapism. Penile, perineal, or pelvic trauma such as straddle or coital injury are the commonest cause of non-ischemic priapism. In neonatal priapism the cause is unknown but subclinical birth trauma is hypothesized to cause most cases. Priapism can also be caused by drugs such as PDE-5 inhibitors, testosterone, and anti-psychotics. First evaluation to differentiate ischemic priapism from non-ischemic priapism is by using penile Doppler US. Management depends on the type of priapism the child develops. Opiates analgesia is usually required in ischemic priapism. Cold packs are analgesic and may cause vasoconstriction decreasing penile blood flow. Hemoglobinopathy and leukemia must be rule out. Successful aspiration and irrigation are reported under conscious sedation with local anesthesia in 4-18 years old. Ketamine is an established detumescence agent and may resolve priapism. During aspiration the presence of dark deoxygenated blood confirms the diagnosis of ischemic priapism, and the corpora should be immediate decompressed until bright blood appears. During aspiration and irrigation sympathomimetics (phenylephrine) might be used. If repeated sympathomimetic is unsuccessful a surgical fistula should be formed. In SCD induced priapism hyperhydration, oxygen therapy, analgesia and exchange transfusions might be needed. For leukemia induced priapism antileukemic therapy (chemotherapy/leukophoresis) and anticoagulation has been advocated. In refractory cases superselective embolization of the internal pudendal artery might be needed. Most hematologic induced priapism in children can be managed with conservative therapy including oxygenation, intravenous hydration, and minimally invasive procedure such as corporal aspiration irrigation and injections.

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