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Neonatal Hyperparathyroidism

Neonatal severe hyperparathyroidism (NSHPT) is a very rare autosomal recessive disease presenting in children during early infancy, usually within the first six months of life though the majority of cases occur in the first few weeks. Babies present with signs of hypercalcemia and hyperparathyroidism including poor feeding, polyuria, dehydration, lethargy, failure to thrive, hypotonia, gastrointestinal dysmotility, osteopenia and respiratory distress due to poorly developed chest cage. Early diagnosis and management of NSHPT is critical due to high morbidity, mortality and devastating effect to neurodevelopmental status. Calcium sensing receptors (CASR) are the major sensors of serum calcium level and have a critical role in maintaining calcium homeostasis. CASR are present in parathyroid chief cells and epithelial lining of renal tubules. The human CASR gene maps to 3q13.3. Through this receptor serum calcium higher than the set point inhibits the parathyroid hormone release from chief cells preventing renal tubular reabsorption of calcium. Mutation of the CASR causes loss or gain of function of the receptor. Heterozygous mutation of CASR results in familial hypocalciuric hypercalcemia and homozygous loss of function mutation results in NSHPT due to uncontrollable release of parathyroid hormone and severe hypercalcemia. Management of NSHPT hypercalcemia initiates with aggressive hydration and forced diuresis with furosemide and use of calcitonin which may provide some transient improvement in heterozygous cases. Homozygous cases do not respond well. Treatment could be escalated to the use of bisphosphonate therapy and calcimimetic such as Cinacalcet which is effective in heterozygous cases. If the child does not respond to medical therapy due to the homozygous state of the mutation then will require early parathyroidectomy. Total parathyroidectomy is performed without autotransplantation due to the high incidence of recurrence of the disease. Localization of parathyroid glands preoperative using Sestamibi nuclear scan and MRI influence the surgical approach.

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Incarcerated Inguinal Hernia Revisited

Inguinal hernia is the common surgical condition in children affecting almost 30% of premature infants. Inguinal hernia results from an incomplete obliteration of the processus vaginalis developed around the 6th month of fetal development. Most inguinal hernia in children are indirect. Incarcerated inguinal hernia is a common and serious emergent situation in pediatric patients. The risk of incarceration in children with inguinal hernia fluctuates between 3 and 16% with the highest incidence of 30% in premature. The rate of inguinal hernia strangulation is also higher in prematurely born infants. The median age of presentation of inguinal hernia is two years. Incarcerated inguinal hernia occurs in 12% of all cases at a mean age of 1.5 years, mostly in boys and mostly on the right side. Due to the significant incidence of incarceration of inguinal hernia, once diagnosed inguinal hernias in children should be repair within the next two weeks. The management of incarcerated inguinal hernia is manual reduction which is successful in most cases, followed by open repair of the hernia in the next 48-72 hours once the edema of the cord has subsided. The procedure can also be done laparoscopically. The advantage of laparoscopic incarcerated inguinal hernia repair include excellent visual exposure, reduction of the incarcerated viscera and inspection for gangrene, immediate repair of the defect, the ability to evaluate the contralateral side, minimal dissection and avoidance of access trauma to the vas deferens and the testicular vessels, iatrogenic ascent of the testis and decreased operative time specially in obese and recurrent cases. The cons are that a subcutaneous procedure is converted into a transabdominal procedure with the incidence of adhesions and later bowel obstruction development. In the case of female patients the possibility of having an incarcerated ovarian inguinal hernia is high. The use of US to determine if an ovary is incarcerated can provide evidence of flow and plan urgent surgery before torsion occurs. Both open herniotomy and laparoscopic repair offer safe surgery with comparable outcomes for incarcerated inguinal hernia in children.

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Eccrine Spiradenoma

Eccrine spiradenoma is a very rare benign tumor of the skin and subcutaneous tissue originating from the eccrine glands described by Kersting and Helwig in 1956. Eccrine spiradenoma (ES) usually appears as a single bluish/pinkish cystic nodule with size between 0.3 and 5 cm, but in rare occasions can be multiple distributed as a linear form or zosteriform. Generally occurs in young adults age 15 to 35 years and rarely can be found in infants. ES occur mainly in the head, neck and trunk associated with paroxysmal pain and tenderness. It tends to arise on the upper part of the body. It is difficult to differentiate eccrine spiradenoma from other more common lesions and biopsy is necessary to establish a diagnosis. The pathogenesis is thought to be related to differentiation mainly of the secretory coil of eccrine sweat glands. It is important to find epithelial cells, myoepithelial cells and lymphocytes along with void spaces between blood vessels under the microscope for diagnostic confirmation. The presence of myoepithelial cells and phosphorylase demonstrate the eccrine origin of the tumor. ES may be associated with similar tumors of apocrine origin such as trichoblastoma and cylindroma. A few cases of malignant changes in eccrine spiradenoma has been reported very rarely. Malignant changes may occur if the eccrine spiradenoma lasts long enough and clinically shows rapid cystic growth pattern. US study reveals a well-demarcated mass with lobulated contours and does not extend into the musculi-fascial tissues. Management consists of local excision removing the tumor completely to avoid recurrence. In cases of incomplete surgical removal a high risk of local recurrence has been reported. Early and complete surgical excision is curative in most cases.

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