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Mixed Gonadal Dysgenesis

Mixed gonadal dysgenesis (MGD) is an intersexual genetic abnormality caused by a defect in the sex chromosomes (gonosomes) associated with dysgenetic gonads and retained Müllerian structures. The most common gonosomal aberration in MGD is 45 X0/46 XY mosaic karyotype. The external genitalia could be normal looking female and these children will present later in life with primary amenorrhea. Otherwise, it could be ambiguous: clitoromegaly and urogenital sinus to a sizable phallus with hypospadias. A uterus and one or both fallopian tubes may also be present. MGD is characterized by a streak gonad and a contralateral testis (that is typically cryptorchid) or bilateral streak testes. The testis might show prepubertal tubules lined by a few spermatogonia and immature Sertoli cells. Female gender assignment is usually preferred, but male assignment is an alternative in instances of extreme virilization. Dysgenetic gonads with the presence of a Y chromosome or a translocated fragment have a significant risk of developing malignant gonadoblastoma (though seminoma and dysgerminoma can occur). Routine early bilateral gonadectomy is advised in MGD. The child to be raised as a female will need clitoral recession and vaginoplasty in early infancy. If it is to be raised as male, then various types of hypospadias repair can be done, gonads can be replaced with prostheses, the prepenile scrotum reconstructed and Müllerian structures removed.

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Recurrent Intussusception

Intussusception, the most common cause of bowel obstruction in infants, occurs mostly in the ileo-cecal area (distal ileum invaginates inside the colon) and it is idiopathic (or caused by hypertrophied Peyer patches after Adenovirus III infestation). Initial management consists of hydrostatic or air enema reduction. Recurrent intussusception (RI) occurs in up to 10% of cases after hydrostatic reduction and to a lesser degree after surgical

reduction (3%). Patients with RI have fewer symptoms with a shorter duration. Most RI occurs within six months of the initial episode. With multiple recurring episodes a search for a pathological lead point (Meckel's diverticulum, polyp, ectopic gastric mucosa, duplication or lymphoid hyperplasia) is imperative. RI even in the face of previous surgery is not a contraindication to try radiological reduction since the success rate is still high and the morbidity low. The child with perforation, signs of peritonitis, irreducible intussusception or a diagnosed lead point should undergo immediate surgical reduction.

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Fibrous Hamartoma

Establishing a diagnosis on clinical or radiological grounds of a subcutaneous mass in an infant or child could be a challenging dilemma, not to mention poorly cost-effective work-up. Fibrous hamartoma is an uncommon benign, soft tissue, painless nodule of rapid growth usually seen during the first two years of life. Initially described in 1956, this lesion is most frequently found in boys, solitary, identified in the subcutis or lower dermis of the axillary region, upper arm, upper trunk, inguinal region, scalp, scrotum and perineum. The lesion varies in size, is firm, fixed to the underlying tissue and not encapsulated. A few cases show overlying skin changes (pigmentation, hair). Pathologically they show well-defined bundles of dense, uniform, fibrous connective tissue projecting into fat, primitive mesenchyme and mature adipose tissue. Diagnosis has been established by fine-needle aspiration technique rarely. Standard care consists of local excision to establish a diagnosis. Observation is not a good option given the uncertainty of missing a malignant lesion. Incomplete removal might result in recurrence and need of wider local excision. Mutilating surgery is unnecessary. The prognosis is excellent.

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