Böbreklerin değerlendirilmesinde longitüdinal ve transvers planlar kullanılır. Longitüdinal planda böbrekler eliptik şekilde, transvers planda ise spinaların her iki tarafında yuvarlak yapılar halinde görülürler. Renal uzunluk büyümenin bir belirteci olarak kullanılabilir; uzunluk, genişlik, kalınlık, ve çevre için standartlar geliştirilmiştir. Böbrek için basit bir kural; haftada 1,1 mm kadar büyümesidir. Gebelik boyunca böbrek çevresinin abdomen çevresine oranı 0,27-0,30 şeklinde sabittir. Ultrasonda mesane 30-45 dakikada bir dolar ve boşalır.

KÖ-46 [11:00]

Preterm doğum: TVS/TAS ile tarama ve yönetim

Alev Atış Aydın

Kanuni Sultan Süleyman Eğtim ve Araştırma Hastanesi Kadın Doğum Kliniği, İstanbul

Serviks ne kadar kısa ise preterm doğum yapma riski o kadar fazladır. Servikal uzunluğun transvajinal USG ile ölçülmesi preterm doğum öngörüsünde kullanılır. Düşük riskli gebeliklerde üniversal transvajinal servikal ultrason taraması kost-efektif bir strateji gibi gözükmektedir. Bu yüzden 18-24.gebelik haftalarında tek bir TVUS ile servikal uzunluk tayini tüm tekiz gebelere önerilebilir.

Daha önce erken doğum yapmış tekiz gebeler 16-23.gebelik haftalarında rutin öyküye dayalı serklaj yerine, güvenli bir şekilde TVusg ile 2 haftada bir (25-29 mm ise haftalık) serviks ölçülerek takip edilebilir.

TVusg ile servikal taramada saptanan kısa servikslerde (<25 mm) vajinal progesteron ile tedavi, y.d morbiditesi ve 34 haftadan önceki preterm doğum riskini %45 oranında azaltmaktadır. Çalışmalarda önceden erken doğum yapmış tekiz gebeliklerde serklaj, pesser ve vajinal progesteron benzer etkinlik gösterirken,çok kısa servikslerde (<15 mm) olduğunda serklaj daha üstün görünmektedir.

Düşük riskli popülasyonlarda TAUSG ile serviks taraması kost-efektif gözükebilir. TAusg ile serviks <30 mm saptananlarda TVUsg ile servikal tarama önerilebilir ancak üniversal tarama önermek için henüz yeterli kanıt bulunmamaktadır.

İkizlerde bu yöntemlerin hiçbiri etkin bir tedavi olmadığı için önerilmemektedir.

KÖ-47 [11:15]

Skeletal system dysplasies

Selahattin Kumru

Düzce University Medical School, Department of Obstetrics and Gynecology, Düzce, Turkey; Department of Obstetrics and Gynecology, Health Ministry Antalya Education and Research Hospital, Antalya, Turkey

Skeletal System Dysplasies are relatively rare conditions.

Dysplasies may be classified as: Achondrogenesis, Achondroplasia, Amelia, mycromelia, Campomelic Dysplasia, Hypophosphatasia, Osteogenesis Imperfecta, Short rib Polydactyly, Thanatophoric Displasia

Extremity Malformations classified as: Clubfoot, Rockerbottom Foot, Sandal Gap Foot, Radial Ray Malformation, Polydactyly, Syndactyly, Clinodactyly, Ectrodactyly, Artrogryposis Akinesia Sequence, Multiple Pterygium Syndrome.

Achondrogenesis

Achondrogenesis are a group of lethal Group of lethal osteochondrodisplasias due to failure of cartilaginous matrix formation and characterized by severe micromelia, unossified spine, short trunk, and disproportionally large head. There are 3 types of the disease: Type IA, Type IB and TypeII. The incidence of disease is reported about 1/40,000-1/50,000 livebirhts and 1/650 perinatal death. Prognosis, IUFD, neonatal death, longest survivor less than 1 month

Achondroplasia

Most common heritable, non-lethal skeletal dysplasia. It is characterized by disproportionately short limb (rhisomelia), frontal bossing, depressed nasal bridge, and short digits. Autosomal dominant single gene disorder. The cause of the disease is Fibroblast Growth factor receptor-3 (FGFR-3) mutations and 80% of cases are new mutations (sporadic). The prevelance of the disease about 1/20,000-28,000 livebirths. While normal lifespan and normal intelligence are expected, neurological and orthopedic complications are common.

Amelia, micromelia, phocomelia

Amelia: Absence of 1 or more limbs.

Micromelia: Shortining of both proximal and distal segment of limbs.

Phocomelia: Shortining of the limb with hand/foot arising near trunk.

Hemimelia: Absence of distal limbs.

Disease is usually diagnosed with missing or severely shortened extremities 1st or 2nd trimester. Roberts Syndrome characterized by orofascial clefting with phocomelia/amelia.

Most chondrodistrophies with severe micromelia lethal in perinatal period and prenatal treatment is not possible. Survivers need orthopedic surgery spinal, limb surgery.

Campomelic dysplasia

Campomelia: Bowed limbs.

Rare, semi-lethal osteochondrodystrophy. It is characterized by bowed extremities with absence of fractures, cutaneous dimpling, hypoplastic scapulae, sex reversal in males (Ambigius genitalia XY sex reversal (male to female)

The incidence of disease is 0.05-1.6/10,000 live births. It is sporadic autosomal dominant and caused by Haploinsufficiency of

SRY gene related gene (SOX9). The is no prenatal treatment change and most of the cases die in infancy due to respiratory insuffucuency.

Hypophosphatasia

Rare osteochondrodysplasia. Deficient mineralisation of the bones and caused by deficiency of tissue nonspesific alkaline phosphotase (ALP). Three types of the disease classified: Perinatal lethal, infantile and Late onset (adult) type.

Imaging findings are; micromelia (perinatal lethal type), Severe undermineralisation of bones and calvarium on mid-trimester USG, multiple rib fractures and short and thick tubuler bones.

The cause of the disease is a mutation in tissue nonspesific ALP (TNSALP) gene and decreased enzyme activity impairs bone minaralization, dentinogenesis.

Prenatal diagnosis is possible. Aggressive dental care, calcium restricted diet, intramedullary rods may helpful.

Osteogenesis imperfecta (OI)

OI is inherited connective tissue disorder with many phenotypic presentations. The estimated incidence is approximately 1 per 20,000 births. Most commonly caused by mutations in genes encoding the alpha-1 and alpha-2 chains of type I collagen. However, proteins involved in posttranslational modification of type I collagen are also cause the disease.

A usefull calisification of OI is presented as; Mild (radiologic type I), Moderate to severe (radiologic types III-IX), and Lethal perinatal form (radiological type II)

The clinical diagnosis of OI is based on the signs and symptoms; bone fragility and a positive family history or several extraskeletal manifestations (hearing loss, dark or bluish sclerae).

The fetal DNA is analyzed for gene mutations in a fetus known to be at risk for having skeletal dysplasia, preimplantation genetic diagnosis is also possible. However, genetic defects have been identified for approximately 70 percent of skeletal dysplasias and there is no definitive, readily available lab test for OI.

The structure and quantity of type I collagen can be determined in vitro from fibroblast culture using a small skin biopsy and abnormalities either in quantity or quality of type I collagen are present in about 90 percent of OI cases. So, negative studies do not exclude the diagnosis, because of the OI types that are not associated with type I collagen mutations (types II B and types V through IX) and the false negative rate of about 10 percent.

Artrogryposis and polydactyly

Arthrogryposis: two or more fixed joint contractures in multiple areas of the body.

Polydactyly indicates that one or more supernumerary digits are present in the hand or foot.

Prognosis of arthrogryposis and polydactyly depends on associated abnormalities and whether this finding is part of a syndrome. Perinatal morbidity and mortality related to an isolated limb defect is low.

Clubfoot

The incidence of clubfoot is approximately 1-3/1000 live births; the male-to-female ratio is 2:1 and two thirds of cases are bilateral. Clubfoot deformity is diagnosed when both the tibia and the fibula are seen in coronal plane, with the sole of the foot visible in the same plane, and this persists during the course of the ultrasound examination. However, a false-positive rate for prenatal diagnosis of clubfoot is up to 30%.

Repeated ultrasound examinatios to reassess for other associated anomalies and to confirm the finding of clubfoot and prenatal pediatric orthopedic consultation may be helpful to discuss postnatal management and prognosis. Approximately 90% of clubfeet are found postnatally to have structural defects requiring orthopedic treatment; 10% are positional defects requiring no postnatal treatment.

Long-term prognosis of clubfoot depends on associated abnormalities; however, the prognosis for normal function with isolated clubfoot is excellent.

Neural tube defects (NTD)

Open neural tube defect is an embryologic defect of the formation of the posterior vertebral arches of the spine, exposing the neural elements. Open NTD occurs in approximately 1/1000 live births.

- Myelomeningocele-sac containing spinal cord or other neural elements
- Meningocele-sac containing only protruding meninges and cerebrospinal fluid
- Myeloschisis-wide splaying of the vertebral arch with no visible covering (neural tube completely exposed)

Disease may be isolated or a part of multiple problem (such as VACTERL).

Prenatal neonatology and pediatric neurosurgery consultation to discuss postnatal management and prognosis. Fetoscopic intrauterine treatment is possible. Delivery in a tertiary care facility is recommended. Cesarean delivery is suggested before the onset of labor to improve functional neurologic outcome. Prognosis of ONTD depends on the level and size of lesion, sssociated anomalies, ventriculomegaly, and type of surgical closure. Periconception folic acid supplementation can reduce the risk of recurrence by 70%.