otherwise normal individual, or it can be related to a congenital disorder, birth defect or genetic syndrome.

Dysmorphic features can vary from isolated, mild anomalies and minor cosmetic imperfections (such as polydactyly) to severe congenital anomalies (such as holoprosencephaly).

In some cases, dysmorphic features are part of a larger clinical picture, sometimes known as a sequence, association or syndrome.

A syndrome is a pattern of multiple anomalies thought to be pathologically related, particular combination of major (essental) and minor (may be absent) criteria.

So, why searching for fetal syndromes?

Early diagnosis is very important especially in the delineation of best care for the patient, prognosis, likelihood of other abnormalities, identifying correct recurrence risk and the best approach to monitor future pregnancies.

Being able to provide as clear information as possible is of great importance to avoid confusion in parents as well as healthcare providers, to make a management plan and to put everything in the perspective.

Recognizing the patterns of fetal malformations is extremely useful for sonologist, practitioners providing prenatal diagnosis.

Ultrasound findings of abnormalities and patterns of more common fetal syndromes as well as some less common fetal syndromes are lined up in this presentation.

Technological advances in ultrasonography, particulary the introduction of high definition 3D and 4D ultrasound allowed us to study fetal anatomy in great detail in very early stages of fetal life, which on their hand helped us to detect fetal abnormalities easier and even earlier than ever before.

Beside fetal anatomy, we are now even able to study a function of some systems and fetal behavior. Fetal behavioral patterns are directly reflecting development and maturational process of fetal CNS. KANET test is the first method that attempted to use 4D US in order to asses and combine different parameters of fetal behavior and form a scoring system in order to determine their neurological status. So, now we are beeing able to detect not only structural abnormalities, but also their functional and behavioral abnormality patterns releted to a fetal syndrome in era of prenatal diagnosis.

KÖ-38 [17:00]

Antenatal diagnosis of urinary pathology/practice cours

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Sonography is an extremely valuable technique for renal imaging. The congenital abnormalities of fetal kidney is subdivided in two dominant categories: those in which the fetal kidney appears hydro-nehrotic and those in which it does not. Among this in which the kidney appears hydro-nephrotic, the major task is to assign a level of obstruction (congenital uretero-pyelo-junction ,congenital megalo-ureter, bladder outlet obstruction and posterior valves)

The approach to non hydroneproic abnormalities is quite different. In this second categorie we have some devastating conditions Including renal agenesis, multicystic dysplasia kidney, which are lethal when seen bilaterally. They also include many group of disease commonly referred to polycystic disease associated or not to syndrome like Meckel Gruber syndroma or Bardet Biedel Syndroma.

On this topic we propose some clinical cases and demonstrate the approach to be followed to the analysis of different semiotic signs urinary and associated signs to make a diagnosis of the pathology. A family and of pregnancy women history can help to the approach of the diagnosis. We propose the follow up and the prognosis of different pathology.

KÖ-39 [17:15]

Obstructive uropathies

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All or some of the urinary system is dilated. If the obstruction is complete and in early fetal period, hypoplasia and dysplasia may occur (Potter type II).

If it occurs in the 2nd half of pregnancy hydronephrosis may develop.

Fetal urology society

Grade O: No dilatation, Grade I: Renal pelvic dilatation, Grade II: Pelvic dilatation and calyx are visible, Grade III: Renal pelvis and calyx are dilated, Grade IV: Grade III and parancime becomes thinner.

- <19 weeks: ≥5 mm.
- 20-29 weeks: ≥8 mm.
- >30 weeks: ≥10 mm(Mandell et al., 1991).

The risk of renal and urinary tract abnormality increases with:

- The severity of hydronephrosis,
- Persistence of hydronephrosis into the third trimester,
- Bilateral involvement, and
- The presence of oligohydramnios.

Hydronephrosis

There may be pelvicaliciel dilatation in 1% of all fetuses.

There may be transient hydronephrosis as a result of high maternal hormone levels or excessive maternal-fetal hydration.

In hydronephrosis cases there may be ureteropelvic obstruction or vesicoureteric reflux.

If the anterio-posterior pelvis renalis diameter is >10 mm and there is pelvicaliciel dilatation, there is moderate hydronephrosis.

If hydronephrosis is diagnosed, dilatation in ureters and uretra must be detected. The size of bladder must be evaluated.

Urinary system abnormalities are generally bilateral, other kidney and amniotic fluid must be evaluated.

If it is diagnosed in 2nd trimester chromosomal abnormalities must be searched.

Vesico amniotic Shunt

Lower urinary tract outflow obstruction may develop in a fetus from pathologies such as urethral atresia and posterior urethral valves, and can be partial or complete.

Severe obstruction may lead to oligohydramnios and pulmonary and renal dysplasia.

There is uncertainty about the criteria for appropriate selection of fetuses for treatment with vesico–amniotic shunting.

Fetal lower urinary tract outflow obstruction is usually managed expectantly or by repeat vesicocentesis.

Some cases are managed by termination of the pregnancy.

The aim of a fetal vesico–amniotic shunt for lower urinary tract outflow obstruction is to decompress the obstructed bladder and restore amniotic fluid dynamics and volume, thereby preventing oligohydramnios and consequent pulmonary and renal dysplasia.

Fetal blood is also sampled for chromosomal analysis to help diagnose or exclude concomitant chromosomal abnormalities that may influence management decisions or treatment choices.

12 Ekim 2014, Pazar

KÖ-40 [08:45]

Fetal hiperekojenik barsak

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Hiperekojenite bağırsağın çevredeki kemikle benzer veya daha fazla ekojenitede olması şeklinde tanımlanır. Bazı kaynaklar da akciğer veya karaciğer ekojenitesine göre karşılaştırma önermektedir. Normalin bir varyantı olabileceği gibi, primer gastrointestinal patoloji, konjenital viral enfeksiyonlar, kistik fibrozis, aneuploidi ve intraabdominal hemorajilerde de hiperekojen barsak karşımıza çıkabilmektedir. Rutin ikinci trimestır antenatal ultrasonografisinde fetal hiperekojen barsak %0.6-1.4 oranında görülmektedir.16. haftadan itibaren parlak mekonyum lümende birikerek ince barsakları daha görünür hale getirmektedir. Mekonyum peritonitinde görülen fetal asit, intraabdominal kalsifikasyon ve intestinal dilatasyonun hiperekojen barsakta görülmemesi ayırıcı tanıyı kolaylaştırmaktadır.

20. gebelik haftasından önce görülen izole hiperekojen barsak genellikle geçicidr, ilerleyen haftalardaki seri ultrasonografilerde gözden kaybolmaktadır. Bu durum çoğu infantta normal barsak fonksiyonuyla sonuçlanmakradır. 3. Trimestırde persiste eden hiperekojenik ince barsak daha çok altta yatan patolojiyi yansıtsa da normal bir sonuçla da karşılaşılabilinmektedir.

Prognoz: İkinci trimestirde hiperekojen barsak görülen fetusların %60'ında doğumdan sonra anomali görülmez. Geriye kalanlarda karyotip anomalisi, IUGR veya perinatal ölüm görülebilmektedir Aneuploidi insidansı %3-27 olup Down sendromu çoğunluğu oluşturmakla beraber, Turner ve triploidiler de görülebilmektedir. Nyberg ve ark. 2. trimestirde hiperekojen barsağın Down sendromlu hastaların %7'sinde bulunduğunu ve yarısının izole olduğunu vurguladılar. Yine de hiperekojen barsak 2. Trimestirde Trizomi 21 için sensitif veya spesifik bir markır değildir. Karyotip anomalileri dışlandığında %10'unda fetal ölüm görülür. Bu oran uteroplasental yetmezlik, prematürite ve fonksiyonel neonatal intestinal obstriksiyondan kaynaklanır. İntestinal atrezi, imperfore anüs, volvulus, CMV ve maternal lupus diğer nadir nedenlerdir. Kistik fibrozisten etkilenen fetusların %60'ında hiperekojen barsak tespit edilebilmektedir.

Yönetim: Karyotip anomalisi, intrauterin enfeksiyonlar ve kistik fibrozis açısından detaylı bir aile anamnezi alınmalıdır. Olası striktürel problemlerin dışlanabilmesi için intestinal dilatasyon ve fetal asit açısından bir kez daha ultrasonografik değerlendirme yapılmalıdır. Seri ultrasonografik değerlendirmeler hiperekojenitenin rezolusyonu, fetal büyümenin takibi ve plasental fonksiyonun değerlendirilmesi için gereklidir.

Daha invaziv araştırmalar; parental kistik fibrozis taşıyıcılığı ve fetal karyotip tayinidir. Persiste hiperekojen barsakta ve umblikal arter kan akımı bozulmuş olan IUGR'da fonksiyonel noenatal intestinal obstruksiyon riski bulunmaktadır. Parenteral nütrisyon, rektal yıkama, suda çözünür kontrast enema mekonyum tıkaçlarını açmak için gerekebilmektedir. Sonrasında ter testi yapılabilmektedir.

KÖ-41 [09:00]

Hiperekojenik böbrek

Ebru Tarım

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Prenatal dönemde yapılan ultrasonografide ekojenik böbrek görülmesi, hem doktor hem de hasta için tanı ve tadavi yö-