Introduzione

Ultrasound examination of the foetal cranium can diagnose fetal cranial defects and abnormal skull shape and its aim is to detect in prenatal age most various abnormalities of the skull, brain and foetal face. The changes of each of these components frequently determine the growth of the other two. The abnormalities of the foetal head frequently appear and can associate serious pathological sceneries of high foetal and perinatal morbidity and mortality. Anomalies of the head have been observed using ultrasound study during the early second trimester. To establish diagnosis and long term prognosis is particularly important and helpful correct detection of the nature of the malformation.

The ultrasound studies of the foetal head other than the evaluations on the cranial head biometry, measured by biparietal diameter (BPD), fronto-occipital diameter (FOD) and head circumference (HC), valuate the bone structure, the presence of sutures between these and the fontanelle which, in prenatal age, result open to permit the correct growth of the skull.

The cephalic index (CI) is the ratio of the BPD to FOD diameters reported as a percentage (short axis/long axis x 100). It has been proposed as a discriminating parameter to recognize alterations in the growth of the skull. Its valuations is indicated when one of the two cephalic parameters is estimated above the +2 SD or below the -2 SD. The index to distinguish between normocephaly (normal value 70÷90) from the dolichocephaly (CI < 75 with a long and thin head, also known as scaphocephaly or boat-shape) and the Brachycephalic (CI > 80 with rounded head due to increased BPD and reduction of the FOD).

Several recent obstetrical sonographic examinations demonstrated that variations in the shape of the fetal skull (dolicocephaly, brachicephaly) may adversely affect the accuracy of the BPD measurement in estimating fetal age.

CI was proposed originally as a means of recognizing altered head shape and confirming the validity of BPD measurements. Two previous studies found cephalic index to be gestational age-independent.

The association between variation in outline and chromosomal abnormalities is source of controversies not yet resolved. Although Borenstein and colleagues found an association between brachycephaly and trisomy 21 at 11 plus 0/7 to 13 plus 6/7 weeks of gestation, such association was not confirmed by others.

Borrel and colleagues determined the cephalic index in 555 consecutive chromosomally normal fetuses and in 38 chromosomally abnormal fetuses before amniocentesis. A cephalic index greater than 0.85 was observed in 14% of fetuses who had Down syndrome and in 11% of normal fetuses. The authors concluded that brachycephaly is not a useful marker for Down syndrome in early mid-trimester fetuses. Preliminary experience indicates that a cephalic index greater than 1 SD from the mean (less than 74% or greater than 83%) may be associated with significant alteration of the BPD measurement expected for a given gestational age, and therefore the head circumference can be used effectively as an alternative and more accurate means of establishing gestational age.

We can also correlate the measures of the foetal head with other parameters such as abdominal circumference (AC) and femur length (FL). Infact, abnormal head circumference/abdominal circumference ratio (HC/AC) or femur length/biparietal diameter ratio (FL/BPD) can suggest cranic abnormalities. Fetuses affected with Down syndrome demonstrate normal BPD but high FL/BPD ratio, secondary to shortened femur length.

Tran et al. (2005) in a multiple logistic regression model demonstrated that second trimester BPD/NBL ratio was
a significant and independent predictor of trisomy 21, and such parameter may increase the detection rate for trisomy 21, if integrated with current ultrasound and laboratory protocols of prenatal screenings.

### Foetal skull abnormalities

#### Craniosynostosis

During ultrasound examination of fetal cranium, it's opportune to check the suture each time to obtain the best scan for the measuring of the skull, to the premature joining of one or more sutures with consequent block of bone growth in perpendicular position of the concerned suture and the compensatory development of the normal suture. Craniosynostosis, the premature fusion of one or more cranial sutures is an etiologically heterogeneous and common malformation occurring in approximately 1 in 2500 live birth.

Craniosynostosis may present as an isolated finding or in association with other anomalies and, according to the involvement of the sutures, they can be simple (premature fusion of a single sutures) or complicated (when there are premature fusion of more sutures). This last type can turn into structural deficiencies called syndromic craniosynostosis.

In the primitive craniosynostosis, the most frequent are scaphocephaly or dolichocephaly, due to the premature fusion of the sagittal suture (the skull grows long relative to it’s width), plagiocephaly due to the premature closure of the coronal suture or of a lambdoidal suture with asymmetric growth of the skull (“oblique skull”), trigonocephaly following the premature fusion of metopic suture (with narrow triangular forehead with concavity of the temples), the pachycephaly caused by the premature joining of the lambdoidal sutures. Dolichocephaly is quite frequent in foetus, in the podalic situation, but needs following with attention because in some cases, in particular if noted prematurely, can camouflage the presence of craniosynostosis of the sagittal suture (scaphocephaly).

In the complicated craniosynostosis, due to the early closure of more sutures, brachycephaly which fused coronal sutures (skull grows wide relative to it’s length) the oxicephaly or turicephaly due to premature fusion of both the coronal and sagittal sutures with abnormal high cervical skull shape, and the kleeblattschädel (enlarged thilobed skull) for premature closure of the front sagittal sutures, coronal and lamboidal, which is the most frequently ultrasound diagnosed malformation due to the typical shape of the foetal skull. Craniosynostosis occurs in isolation in 85% of cases but is in general descrive in association with several syndrome, including cranial shape abnormalities and severe malformations of the digits and grouped together under the term acrocephalosyndactyly (ACS).

The Online mendelian Inheritance in man database contains 120 entries with craniosynostosis as an abnormal feature. The syndromic craniosynostosis is the hereditary form of craniosynostosis, which is associated with extracranial phenotypes such as limb, cardiac, central nervous system and tracheal malformations.

For different of these syndromes, among which Crouzon, Apert, Pfeiffer and Saethre-Chotzen, the genetic bases are been identified.

Syndromic craniosynostosis has been reported to be associated with mutations of several gens including FGFR1, FGFR2, FGFR3, FBN1, TWIST, and MSX2 genes.

Among these identified mutations, mutations in the genes of fibroblast growth factor receptors (FGFRs), particularly FGFR2, are most common and have been implicated in syndromic craniosynostosis. The molecular mechanisms and genotype-phenotype correlations of FGFR2 mutations have been shown to be very diverse.

In a recent study on patients with syndromic craniosynostosis, carried out phenotype analysis, the Authors founded the sequent data. Proptosis coexisted with small midface in 11 of 12 patients. Syndactyly and broad digit were found in all of the patients with Pfeiffer syndrome and severe webbing with syndactyly in the second, third and fourth digits were found in the patient with Apert syndrome di tipo 1. All patients with Crouzon syndrome showed the typical craniofacial characteristics of this syndrome such as abnormal head shape, small midface, proptosis and fused sutures. Vertebral anomalies were found in the patient with Beare-Stevenson cutis gyrata syndrome di tipo 1. Patient with Muenke syndrome 1 showed mild midfacial hypoplasia with coronal suture fusion, severe sensorineural hearing loss and strabismus.

Craniosynostosis can normally be diagnosed during the observation of the anatomic abnormalities. Trough ultrasonic studies, turibrachy cefaly was noticed on a fetuses between the 16th and 17th week, who suffered Apert syndrome, as the mother. A significant sign in these cases. It was also reported at the 32nd week of pregnancy a unilateral coronal suture synostosis with asymmetric multilobulated skull, ultrasound is also used for diagnosing sagittal suture craniosynostosis (scaphocephaly).

Prenatal ultrasound images of patients with craniosynostosis where studied by Miller and colleagues to determine the extent of prenatal diagnosis. The Authors concluded that diagnosis of these malformations was not possible in the first trimester, while, abnormalities such as kleeblattschädel, trigonocephaly, brachycephaly (bilateral coronal suture craniosynostosys) and plagiocephaly (unilateral coronal suture craniosynostosys), is possible during the second and third trimesters.

#### Discussion

Anomalies of the foetal cranium have been amenable to early second trimester ultrasound finding study. To establish diagnosis and long term prognosis is particularly important and helpful correct detection of the nature of the malformation.

Echotomography of the foetal head is based on biometry and evaluation of the bone structure, the presence of sutures between these and the fontanelle to prenatal diagnosis of foetal skull abnormalities, such as craniosynostosis.

Ultrasound examination represent an important instrument to prenatal check the greatest part of the foetal skull abnormalities and also 3D ultrasound can be useful be-
Echotomography of craniosynostosis: review of literature

sides in to show the entity of the malformation and in parental counselling.

References