II. NEONATOLOGY

ETIOLOGIC AND POSITIV DIAGNOSIS OF CRANIOSYNOSTOSIS

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Abstract

In this material authors aim to make a short presentation of most frequent shape and structure modifications of cephalic extremities.

The start point of this study was, on one hand, the growing addressability, in pediatric rooms, of patients with modifications of head shape and dimensions, and on the other hand, diagnosis errors that could appear; particular aspect could be seen as pathologic. We would like to present these anomalies from the clinical and paraclinical point of view and to get them into a diagnosis entity.

Key words: cranyostenosis, newborn, sutures

Introduction

The head perimeter of a healthy, term newborn is about 34 – 36 cm. In the first year of life perimeter grows with approximately 10 cm, so that at the age of 1 year it reaches about 46 cm and in the following 20 years the perimeter grows with another 10 cm, so that at one adult it has about 56 – 57 cm. The modifications in plus (macrocephaly) and in minus (microcephaly) from these dimensions could be pathologic. The most frequent cause of microcephaly is craniosynostosis.

Craniosynostosis represents the premature fusion of one or more cranial sutures leading to modifications of head shape and/or dimensions. Craniosynostosis can be classified as: primary and secondary. Primary craniosynostosis may result from a primary defect of ossification; secondary craniosynostosis is characterized by a failure of brain growth and secondary ossification.

Craniosynostosis can also be classified by the number of sutures involved: simple craniosynostosis (one suture involved) and complex craniosynostosis (multiple sutures involved).

Incidence

By some authors the incidence is 0,4 / 1000 birth, sagital suture being involved mostly.

Causes

The most frequent causes, from literature, are: idiopathic, endocrine (hyperthyroidism, hypophosphatemia, vitamin D deficiency, renal osteodystrophy, hypercalcemia), hematologic (thalassemia), genetic (Apert syndrome, Crouzon syndrome, Pfeiffer syndrome).

There are also mentioned some risk factors as: caucasian mother, maternal age, male infant, maternal tobacco abuse, fertility treatments, treatments with nitrofurantoin, chlorpheniramine.

Clinical forms

Ossification of the cranial vault is circulary, starting from central region of each cranial bone and extending outward toward the cranial sutures and fontanelles (fig.1).

- The metopic suture separates the frontal bones;
- The coronal suture separates the frontal bones from the parietal bones;
- The sagittal suture separates the parietal bones;
- The lambdoid suture separates the occipital bones from the parietal bones.

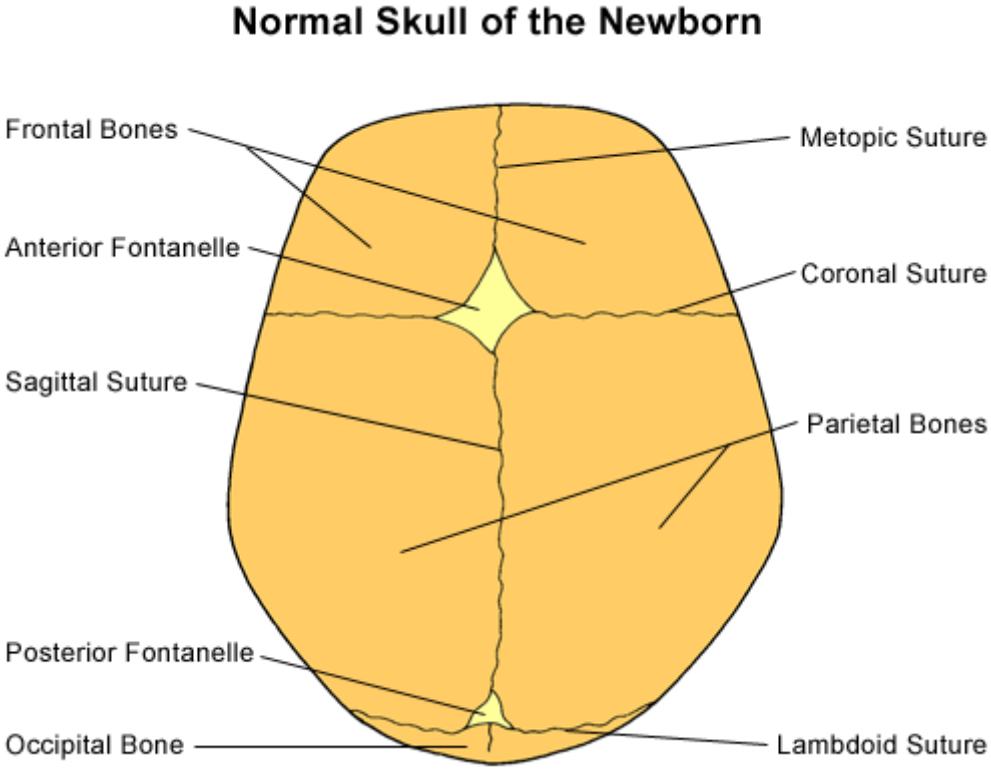
Clinical diagnosis

Consists of several steps: anamnesis – to emphasize the presence since birth, physical exam which reveals: asymmetry, particular aspect, ossification of sutures and fontanelles, measurements of the cranial perimeter to evaluate Hydrocephalus and Microcephaly, evaluation for any other musculoskeletal abnormalities (ex.: torticolis, fingers and toe abnormalities)

Ossification of Metopic Suture (Metopic Synostosis)

The ossification of metopic suture determines the appearance of trigonocephaly (fig. nr. 2)

The shape of the shead is pointed, as a triangle; anteroposterior diameter is small, eyes appear closer together, forehead more pointed. It is, usually, a mild affection and does not need surgery.



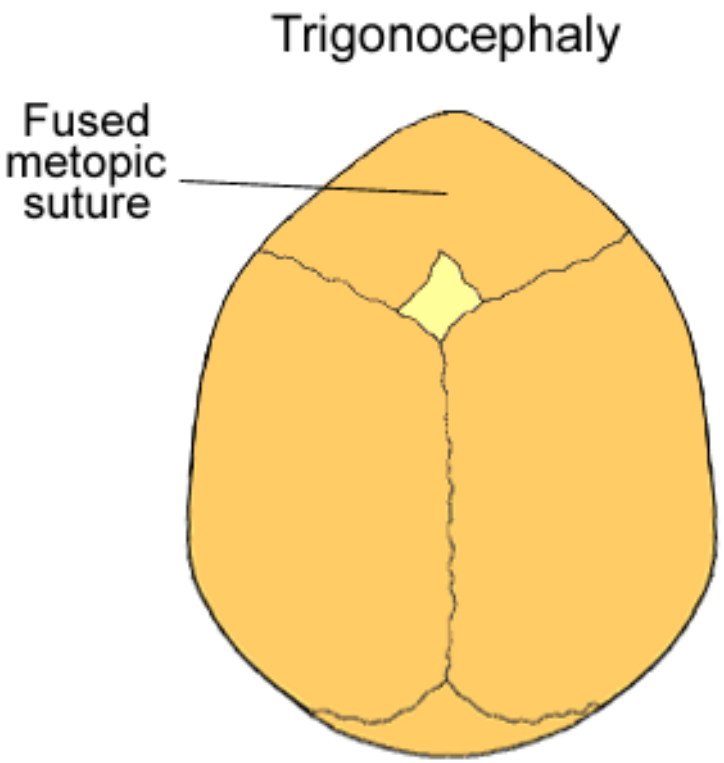


Fig. nr.2 Trigonocephaly.

Bicoronal Ossification (Bicoronal Synostosis)

The determined affection is brachycephaly (fig. nr.3), through premature ossification of both coronal sutures. Biparietal diameter is widened and the anteroposterior one is smaller. It is a severe disease associated with premature ossification of metopic suture with modifications of skull bones, face hypoplasia, flattened nose. Frequently associated with Crouzon syndrome and Apert syndrome. It is a serious affection and needs surgery, usually in the first year, to increase anteroposterior diameter.

Unilateral coronal synostosis = Frontal Plagiocephaly (fig. Nr.4).

Occurs by premature ossification of coronal suture; it is characterized by decreased anteroposterior diameter, flattened forehead.

Fig. nr.3 Brachycephaly.

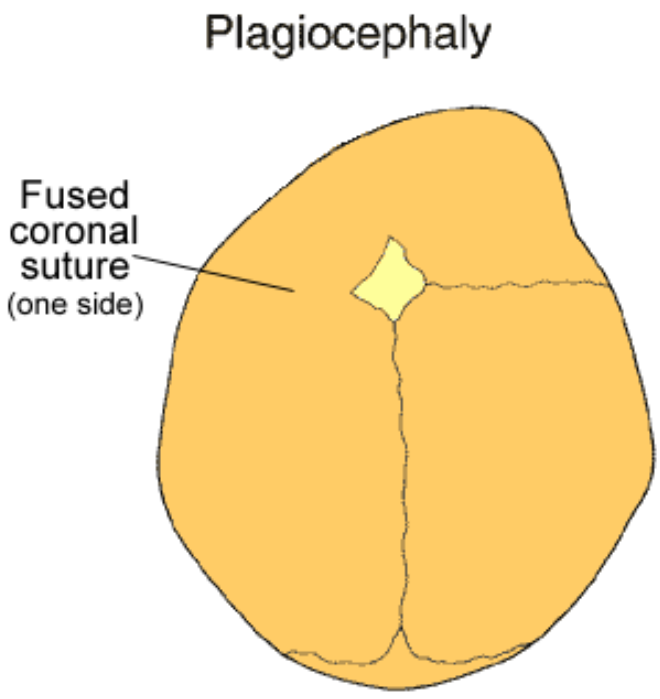


Fig.nr.4 Plagiocephaly.

Unilambdoid synostosis = Occipital Plagiocephaly (fig. Nr. 5).

Occurs due to premature ossification of lambdoid suture.

It is characterized by decreased anteroposterior diameter.

At this form may be taken into consideration positional modifications

Sagittal synostosis = Scaphocephaly

Early ossification of sagittal suture is the most frequent craniosynostosis (1/4000 newborns) especially at male infants. It occurs more at premature; anteroposterior diameter is longer and the biparietal narrower.

Primary craniosynostosis occurs when one or more sutures fuse prematurely, while the brain is still increasing. In this situation intracranial pressure can increase, with all its consequences.

Secondary craniosynostosis is more frequent than the primary type and results from primary failure of brain growth (microcephaly). Usually is not accompanied by increased intracranial pressure; does not compromise normal brain growth and does not need surgery.

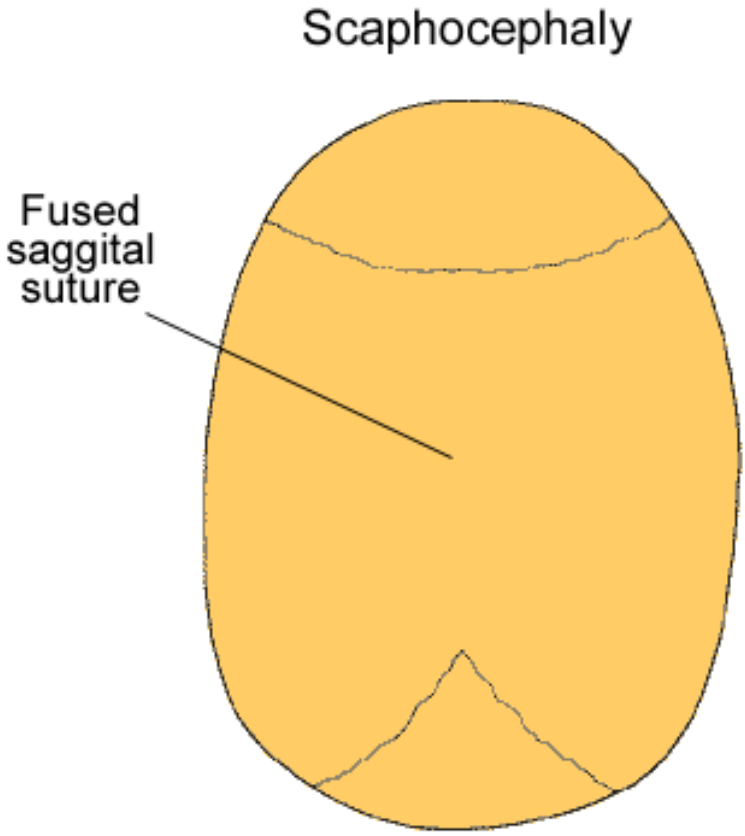


Fig.nr.5 Scaphocephaly.

Diferential diagnosis

Several affections can be taken into consideration, with modifications of cefalic extremity: cerebral tumors, hydrocephaly, hidranencephaly, endocrine diseases, craniovertebral dysraphism.

In the etiologic diagnosis must be considered: Apert syndrome, Crouzon syndrome, Pfeiffer syndrome, and all other affections mentioned at causes.

Paraclinic investigations

For a precise diagnosis and in order to localize the premature ossificated suture there are needed:

- Skull radiography with anterior-posterior and lateral views
- CT and RMN especially when surgery is being considered
- Transfontanellar ultrasound (if anterior fontanella is open), shows ventricular dilatation (when craniosynostosis is associated with a malfomation syndrome), hydrocephalus, corpus callosum agenesis
- Endocrine evaluation

Complications

Severe, untreated craniosynostosis can lead to complications: increased intracranial pressure, face asymmetries, abnormal dental occlusion, orbit asymmetry associated with secondary strabismus

Treatment

Medical care

Practically there is no medical treatment, only hospitalization and prevention: monitor signs and symptoms of elevated intracranial pressure, measure head

circumference, ophthalmologic consult at patients with high intracranial pressure

Surgical care

It is the election treatment for infants with microcephaly (secondary craniosynostosis). Surgery is complicated and it is not performed currently.

In primary craniosynostosis with asymmetric head shapes surgery is performed for esthetic purpose.

Surgery supposes a complete etiologic diagnosis, a result of a team work: pediatric neurologist**,** geneticist**,** plastic surgeon**,** neurosurgeon and endocrinologist.

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14. Fig.1 Fig.2 Fig.3 Fig.4 Fig.5 www.gfmer.ch/genetic_diseases_v2/gendis_detail_list.php?cat3=86

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