Prenatal MR imaging of focal cortical gyration anomalies at early stage of development

A. Righini[1], C. Parazzini[1], C. Doneda[1], L. Avagliano[2], F. Arrigoni[1], M. Rustico[1], G. Bulfamante[2], and F. Triulzi[1]
[1]Radiology and Neuroradiology, Children's Hospital V. Buzzi, Milan, Italy, Italy, [2]Pathology, San Paolo Hospital, Milan, Italy, Italy

Synopsis
We report the MRI patterns of focal cortical gyration anomalies, as they appear at a very early stage of the sulcation process (when fetal brain is almost "lyssencephalic"). 22 cases (gestational age between 21 and 24 weeks) showed focal gyration anomalies, which could be divided in four basic patterns of cortical rim distortion: *"wart-like"*, *"saw-tooth"*, *major aberrant invaginating sulcus/i*, *single or multiple bumps*. Most of these cases presented similarities to the rat model of experimentally induced polymicrogyria. The present cohort shows how focal cortical gyration anomalies can be detected even at very early sulcation process stage

Introduction
Scarce information are available about early stage development of cortical gyration anomalies, while there are extensive data on experimental cortical dysplasia (rat model). We show the magnetic resonance (MR) imaging patterns of focal cortical gyration anomalies, as they appear at a very early stage of the sulcation process; that is when human fetal brain is basically as smooth ("lyssencephalic") as the rat brain.

Methods
We reviewed 950 fetal brain MR imaging studies acquired between 2000 and 2009 within the 24th week of gestation. All exams had been performed using 1.5 Tesla scanner with surface or phase array abdominal coils and multiplanar ss-FSE T2-weighted sequences (TR/TE 3000/180 ms, 3-4 mm thick slices, 1.16 to 1.25 mm in plane resolution). In a minority of cases, T1-weighted sections could be obtained free from motion artefacts in order to be evaluated. The cases showing focal anomalies of cortical rim were selected and classified according to the type of cortical ribbon distortion, by two senior pediatric neuroradiologists in consensus. 22 cases were selected according to the above criteria, gestational age ranged between 21 and 24 weeks (average 22.3, sd 1.07 weeks). In 11 cases postnatal MR imaging, MR autopsy, or pathology follow-up was available, while in 11 cases no follow-up data on brain structure could be collected (mostly because impossibility to collect the brain after pregnancy termination); however, in one of the latter cases molecular genetic analysis confirmed the diagnosis to be tuberous sclerosis.

Results
We identified four basic patterns of cortical rim anomaly: *"wart-like"* (8 cases), *"saw-tooth"* (6 cases), *major aberrant invaginating sulcus/i* (6 cases), and *single or multiple bumps* (3 cases) . All anomalies were already detectable when the brain was still smooth ("physiological lyssencephaly"). The *"wart-like"* and *"saw-tooth"* patterns were confirmed to be focal polymicrogyria when follow-up was available. In *"wart-like"* and in *aberrant sulci* cases the anomaly became more complex along with gyration process. A *bump* anomaly turned to be a cortical tuber at very early stage of development. All cases were associated with additional brain anomalies: callosal agenesis, focal parenchyma volume reduction, septum pellucidum agenesis, periventricular nodular heterotopias, focal thinning of subplate layer or intermediate layer, germinal matrix-ganglionic eminence hypertrophy, cerebellar hypoplasia, micro or macrocephaly, borderline ventriculomegaly. Four cases were probably on genetic basis, four very probably of hypoxic-ischemic and infective (3) origin; 11 cases were of unknown cause.

Conclusions
The present cohort shows how focal cortical gyration anomalies can be detected even at very early sulcation process stage. Most of these cases presented similarities to the rat model of experimentally induced polymicrogyria. Moreover, these cases showed how polymicrogyria and aberrant sulci grow along with brain maturation. The detection of migrating cells focal alterations suggests a migratory defect playing a role in the genesis of some gyration anomalies. Finally, the thinning of the subplate and of intermediate layer of cerebral mantle, noticed in some fetuses, suggests that also failure in the framework sustaining the cortical plate may be relevant in the pathophysiology. One of the reasons of the presence of associated anomalies in all cases may be related to ultrasound findings prompting MR imaging investigation.

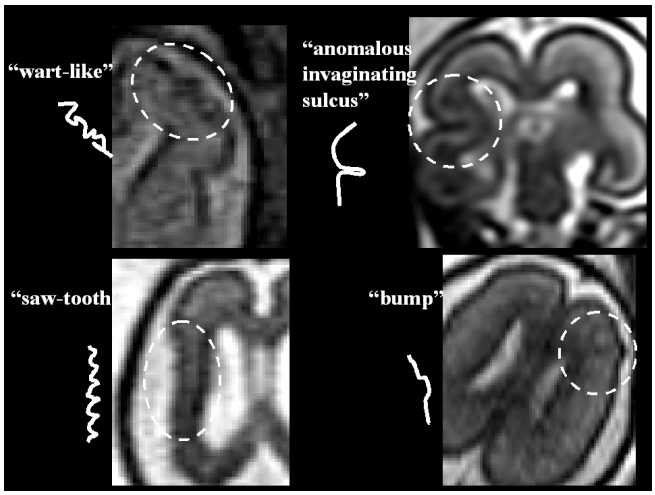

Figure 1
ss-FSE T2-weighted coronal and axial sections showing four patterns of focal cortical gyration anomaly identified in different exemplificative cases..