Background X-linked lissencephaly with abnormal genitalia (XLAG) is established as one disease entity. XLAG, showing severe neonatal seizure and developmental delay, is a rare disorder caused by mutations in the aristaless-related homeobox (ARX) gene, located in Xp22.13. Arx-null mice for human XLAG model showed loss of tangential migration of GABAergic interneurons.

Objectives We investigated subpopulation of GABAergic interneurons in the brain of an infant with XLAG, who had a nonsense mutation of the ARX gene, compared with those of age-matched normal control, Miller-Dieker syndrome (MDS) as a type I lissencephaly, and polymicrogyria of Fukuyama type congenital muscular dystrophy (FCMD) as a type II lissencephaly.

Methods We used paraffin-embedded brain tissues of two XLAG, three MDS and four FCMD, with an informed consent of their parents. We performed immunocytochemistry for interneuron and migration markers.

Results Glutamic acid decarboxylase (GAD) and calretinin (CR) containing (+) cells were significantly very few in the neocortex and located in the white matter and neocortical subventricular zone. In the neocortical subventricular region, the GAD+ and CR+ cells had Mash3 protein, like a radial migration marker, and nestin protein. On the contrary, MDS showed relative low concentration of GAD+ cells. FCMD revealed random distribution of these marked cells.

Conclusions ARX controls not only tangential migration of GABAergic interneurons from the ganglionic eminence, but also may serve to induce radial migration from the neocortical subventricular zone. MDS and FCMD also demonstrated abnormal distribution of neocortical interneurons, but those severities are different in each type of lissencephaly.

CEREBRAL PERFUSION FROM INFANT TILL ADOLESCENCE ASSESSED WITH MR PSEUDO CONTINUOUS ASL

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Background and aim Arterial spin labelling (ASL) is a MR technique to assess brain perfusion without necessity of intravascular administered MR contrast [1]. Our aim was to obtain age dependent normal paediatric values of brain perfusion.

Methods In this retrospective study we included children aged 1–14 years recruited from our MRI database, collected from 2012–2014. In each age group 6 individuals were included having a normal MRI scan. All children were scanned on a 1.5 T MRI scanner (GE). A pseudo continuous ASL technique was