

The toothless molar tooth malformation

How to make the diagnosis on prenatal MR scans

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Objectives

The molar tooth sign on imaging studies is based on maloriented superior cerebellar peduncles, a deepened interpeduncular fossa, and vermian hypoplasia^{1,2}. However, this sign may be less clearly delineable prenatally. The aim of this study was to define other morphological characteristics in fetal MRI to allow for an accurate diagnosis of molar tooth malformations.

Methods

Retrospectively, fetal MRI scans of 66 fetuses with suspected ciliopathies were analyzed. Among them, 12 cases with an unclear cystic posterior fossa malformation and postnatal/postmortem and/or genetical proof of Joubert syndrome or molar tooth malformation underwent re-evaluation of MR-images. The following anomalies were screened for: Molar tooth sign, brainstem anomalies, thickened superior cerebellar peduncles with parallel course to the skull base with or without signal intensity changes, dysplastic cerebellum, celes, and extracerebral manifestations of ciliopathies.

Results

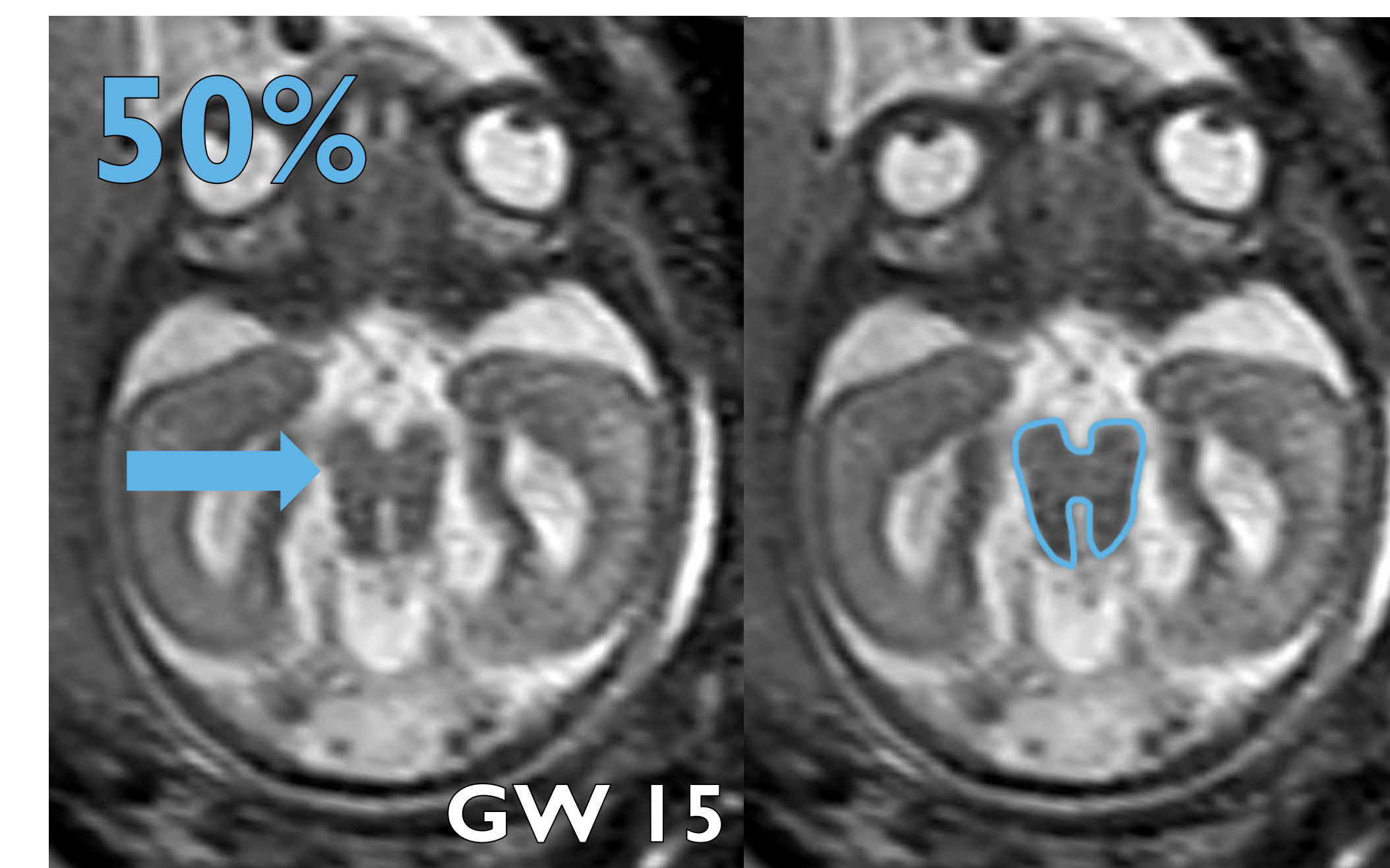
MTS was present in 50% of cases. Pathological appearance of the superior cerebellar peduncles could be proved in 80%, a dysplastic cerebellum and brainstem pathologies, mainly consisting of isthmic malformations and/or misproportion of the respective segments were present in 100%. All cases showed a hypo- and dysplastic vermis. (Abortive) celes were seen in 9, extracerebral anomalies, especially involving the kidney, occurred in 8 cases.

Conclusions

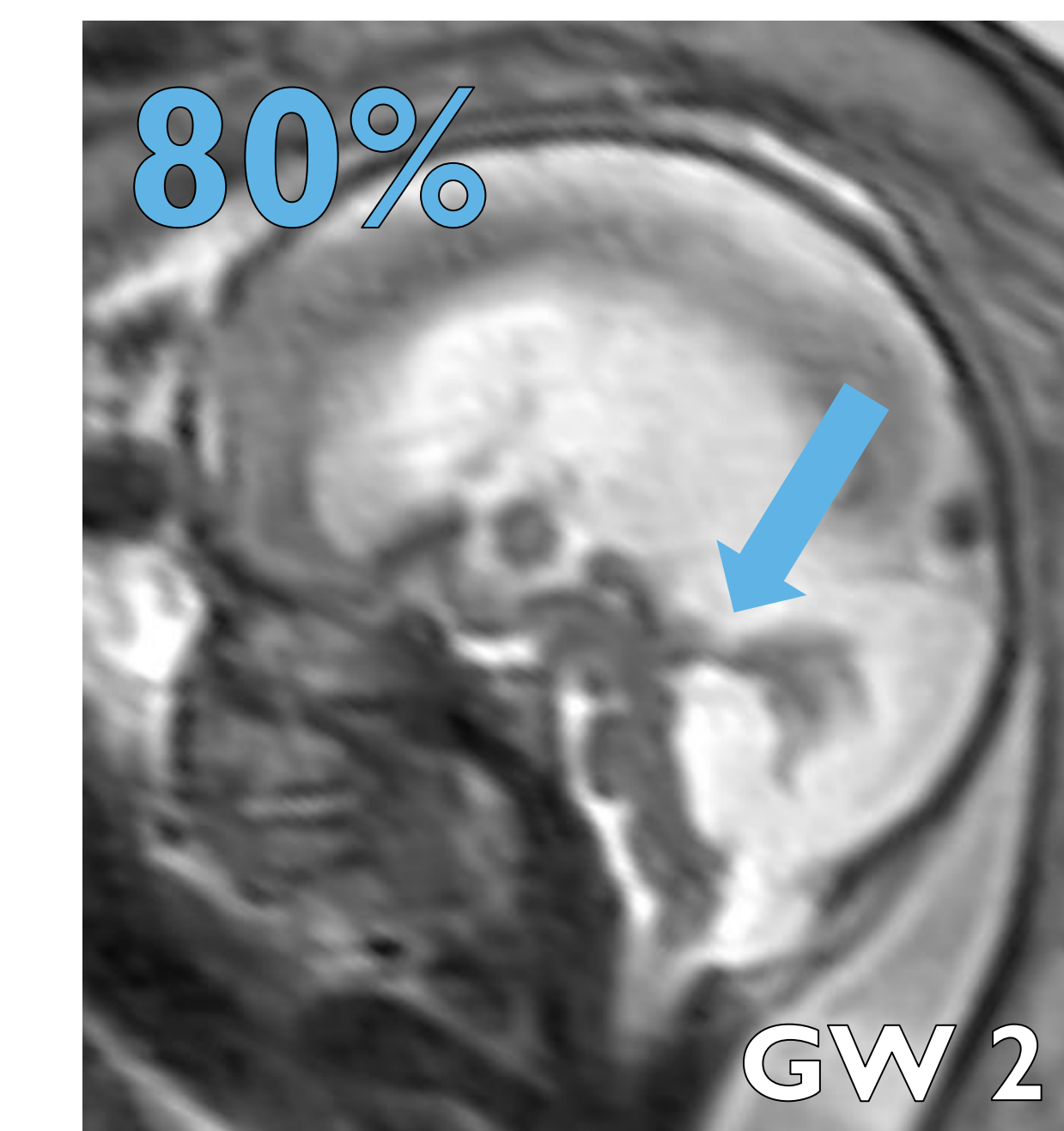
On prenatal MR-images, molar tooth malformations can be diagnosed even in absence of a MTS by characteristic anomalies of the superior cerebellar peduncles, the brainstem, and the cerebellum with certain extracerebral anomalies supporting this diagnosis.

References

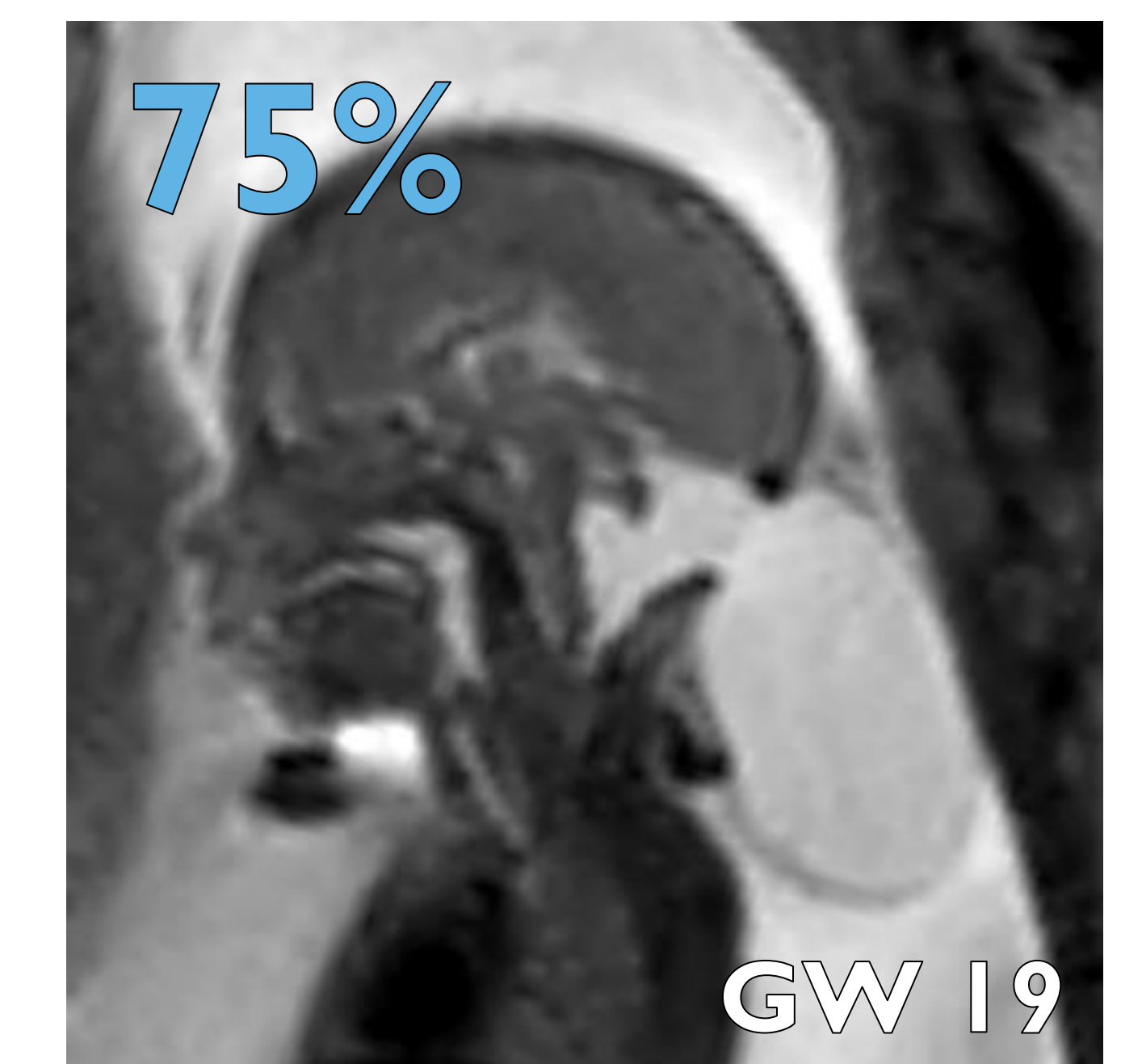
1. Romani M et al. Lancet Neur. 2013 Sep;12(9):894-905.
2. Pugash D et al. UOG. 2011 Nov;38(5):598-602.



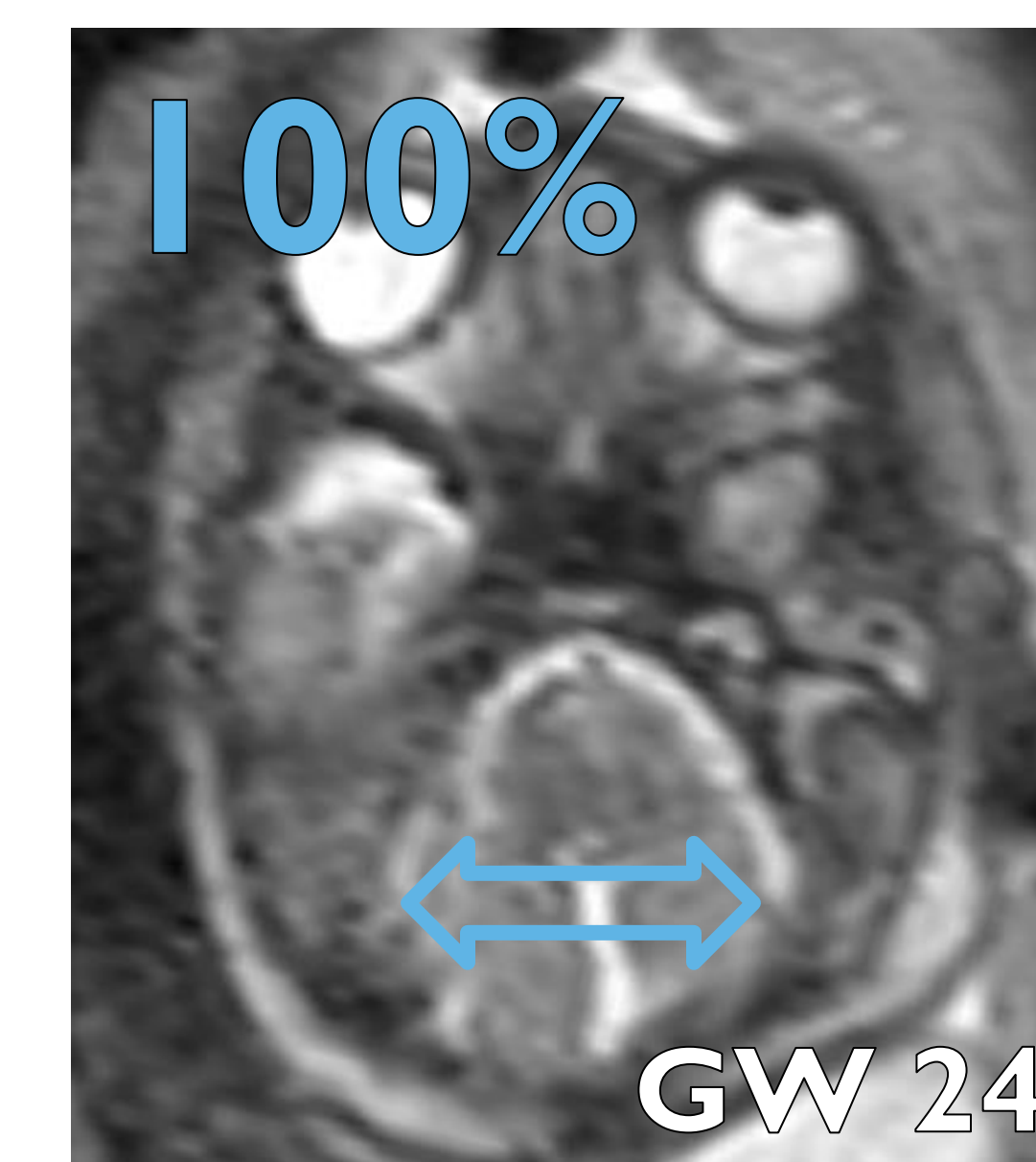
Molar tooth sign



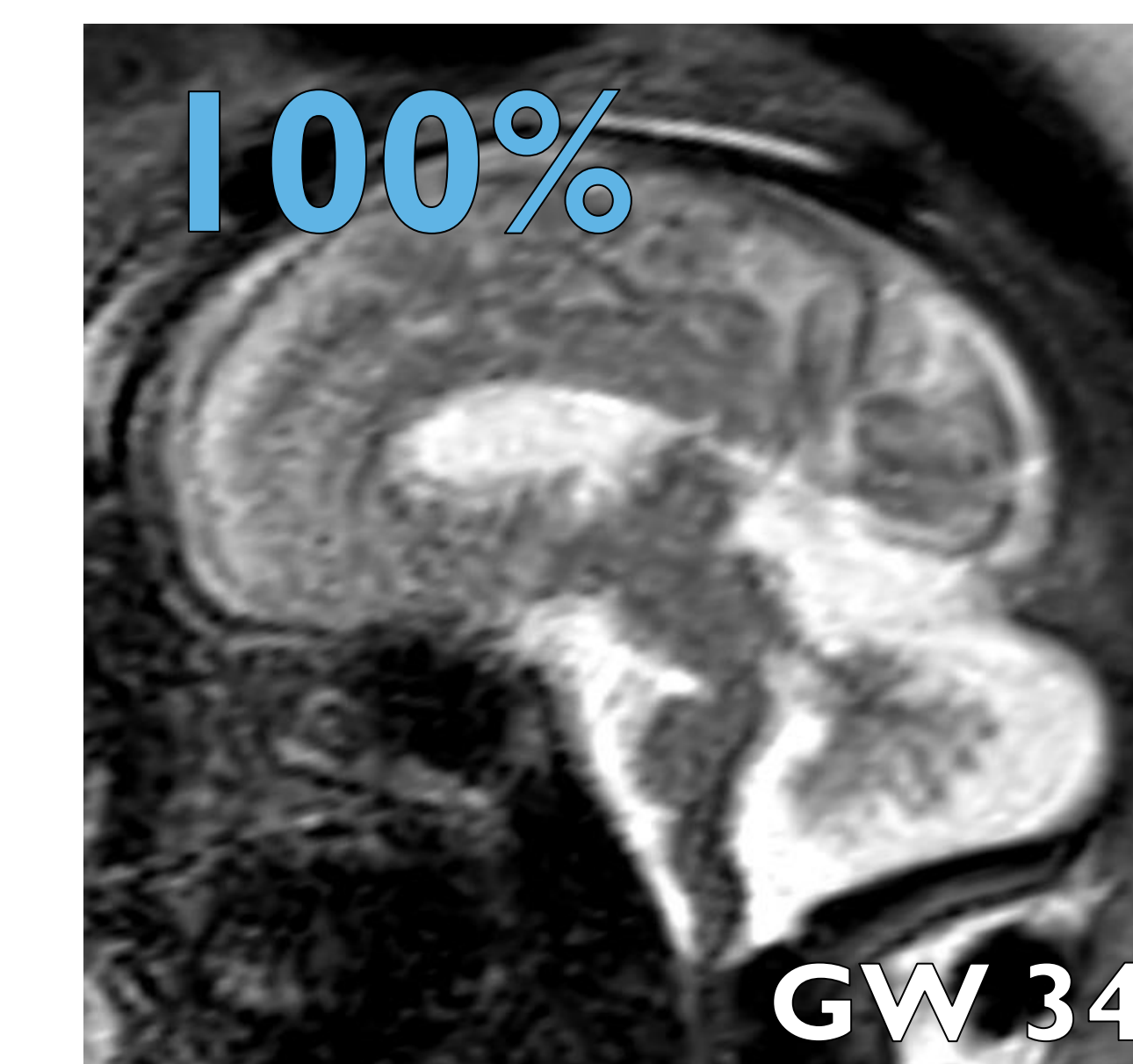
abnormal superior
cerebellar peduncles



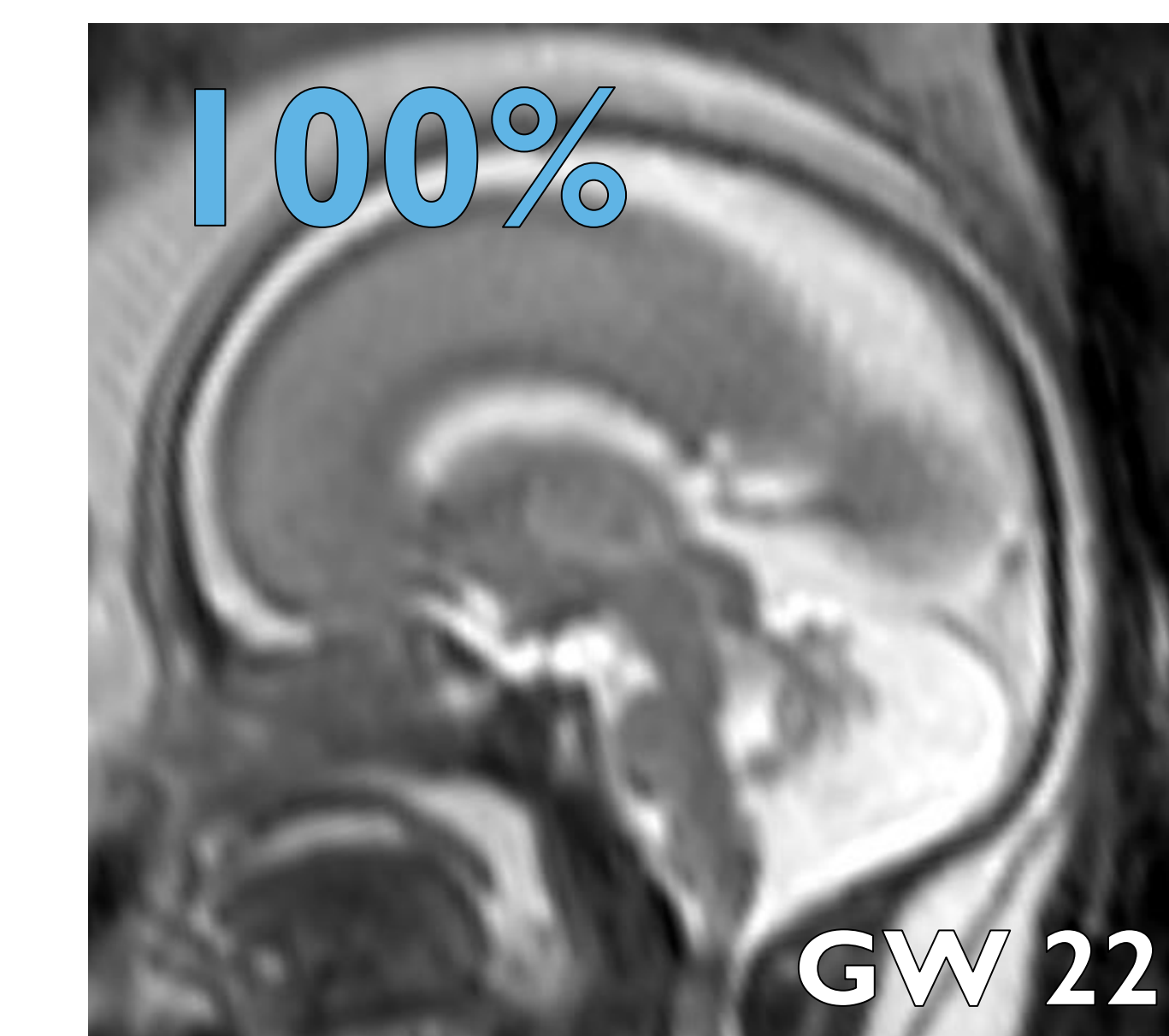
(abortive) celes



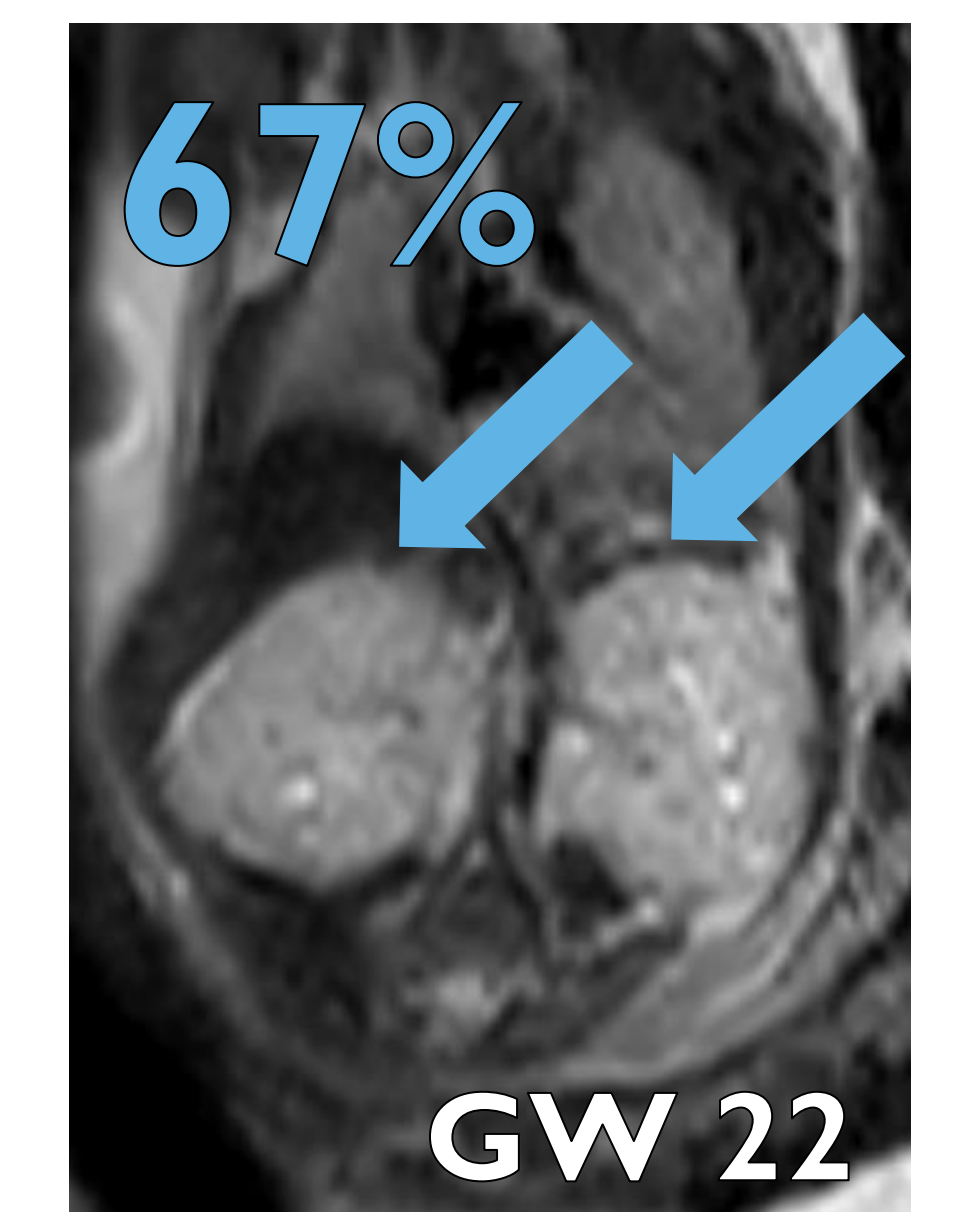
hypo-/dysplastic
cerebellum



hypo-/dysplastic
brainstem



hypo-/dysplastic
vermis



extracranial
anomalies