

Prenatal diagnosis of tubulinopathy: a case report

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Introduction

Prenatal diagnosis of tubulinopathies is challenging because tubulinopathies present a wide spectrum of brain anomalies caused by genetic variants in genes that encode tubulins. We report a fetus with tubulinopathy presenting with ventriculomegaly (VM) and confirmed by exome sequencing.

Case report

A 31-year-old pregnant woman, G2P1, was referred to our hospital for evaluation due to bilateral VM at 23 weeks of gestation. Fetal neurosonography (NSG) revealed deviation of the anterior part of the interhemispheric fissure, abnormal shape of anterior horn of bilateral lateral ventricles, corpus callosum hypoplasia, cerebellar hypoplasia, and abnormal Sylvian fissure operculum suspicious for cortical malformation. Fetal MRI confirmed the sonographic findings and also demonstrated flat pons suggesting pontine hypoplasia. After counseling, the family opted to terminate the pregnancy. Whole exome sequencing revealed a missense mutation in the TUBA1A gene compatible with tubulinopathy.

Conclusion

This case demonstrates that while fetal NSG or MRI may be able to detect a wide spectrum of fetal brain anomalies, the underlying etiologies still need sequencing for better prognostication and counseling.

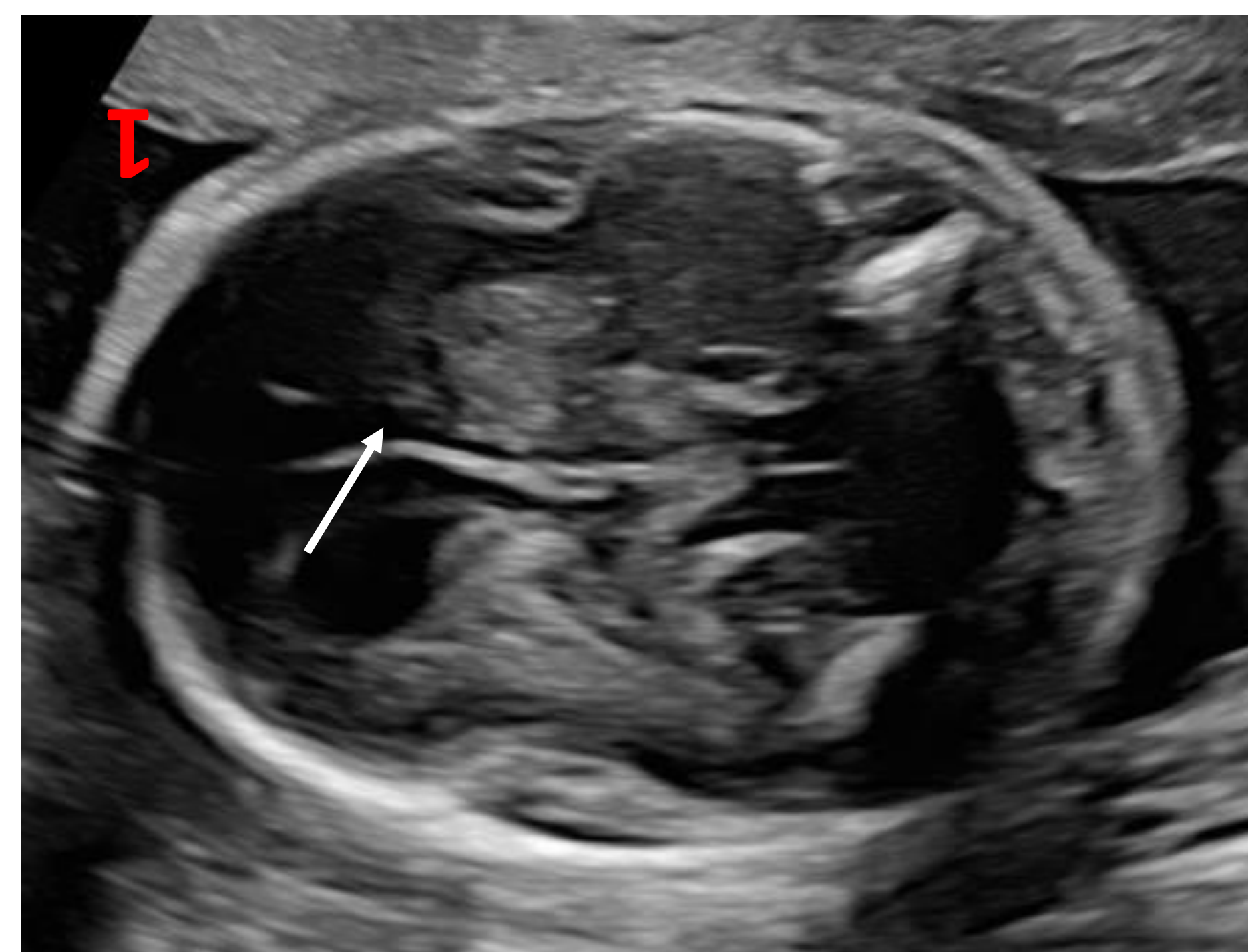


Figure 1
Deviation of the anterior part of IHF and abnormal Sylvian fissure operculum

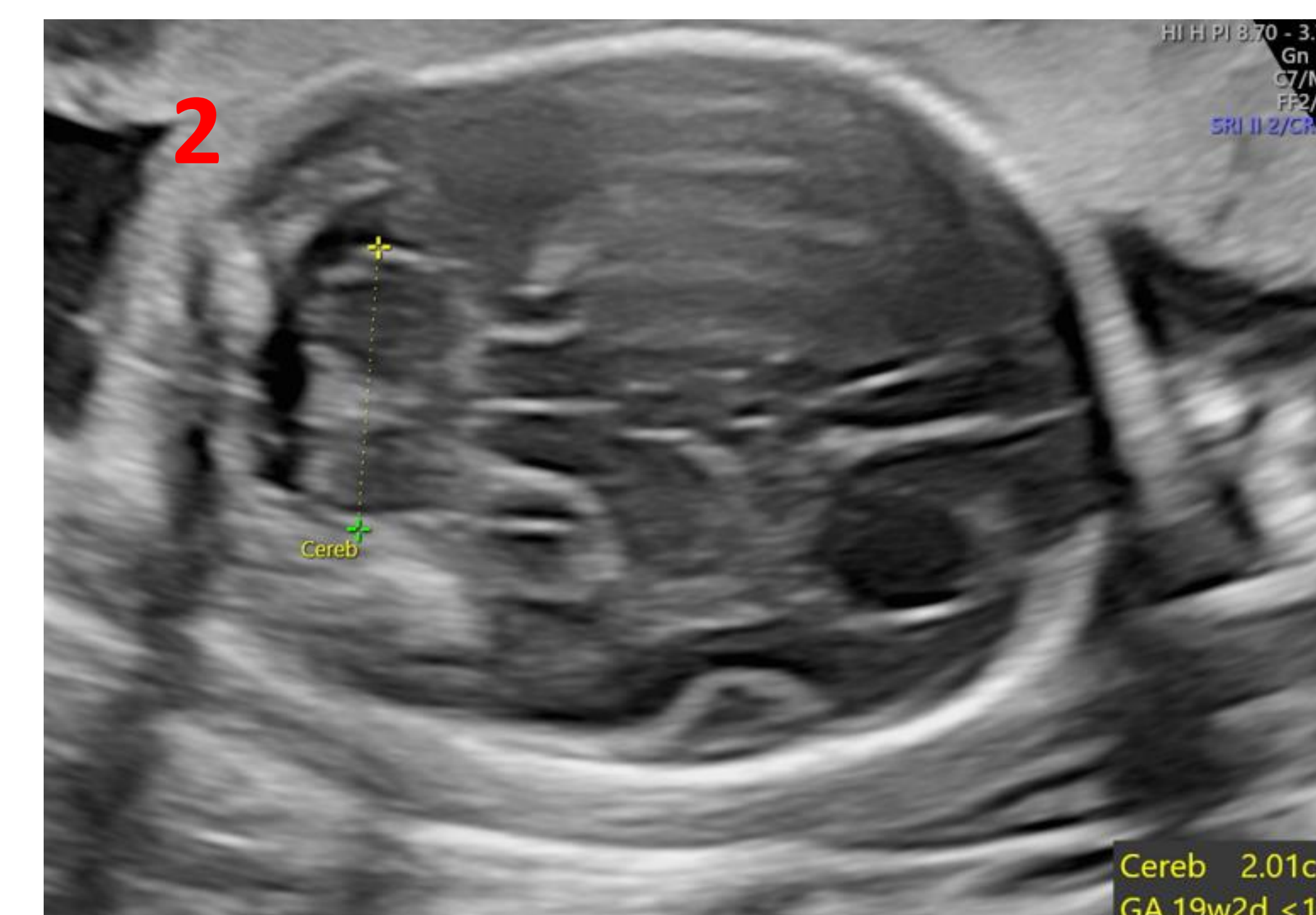


Figure 2
Small TCD (20.1 mm) compatible with 19w2d, suspected cerebellar hypoplasia

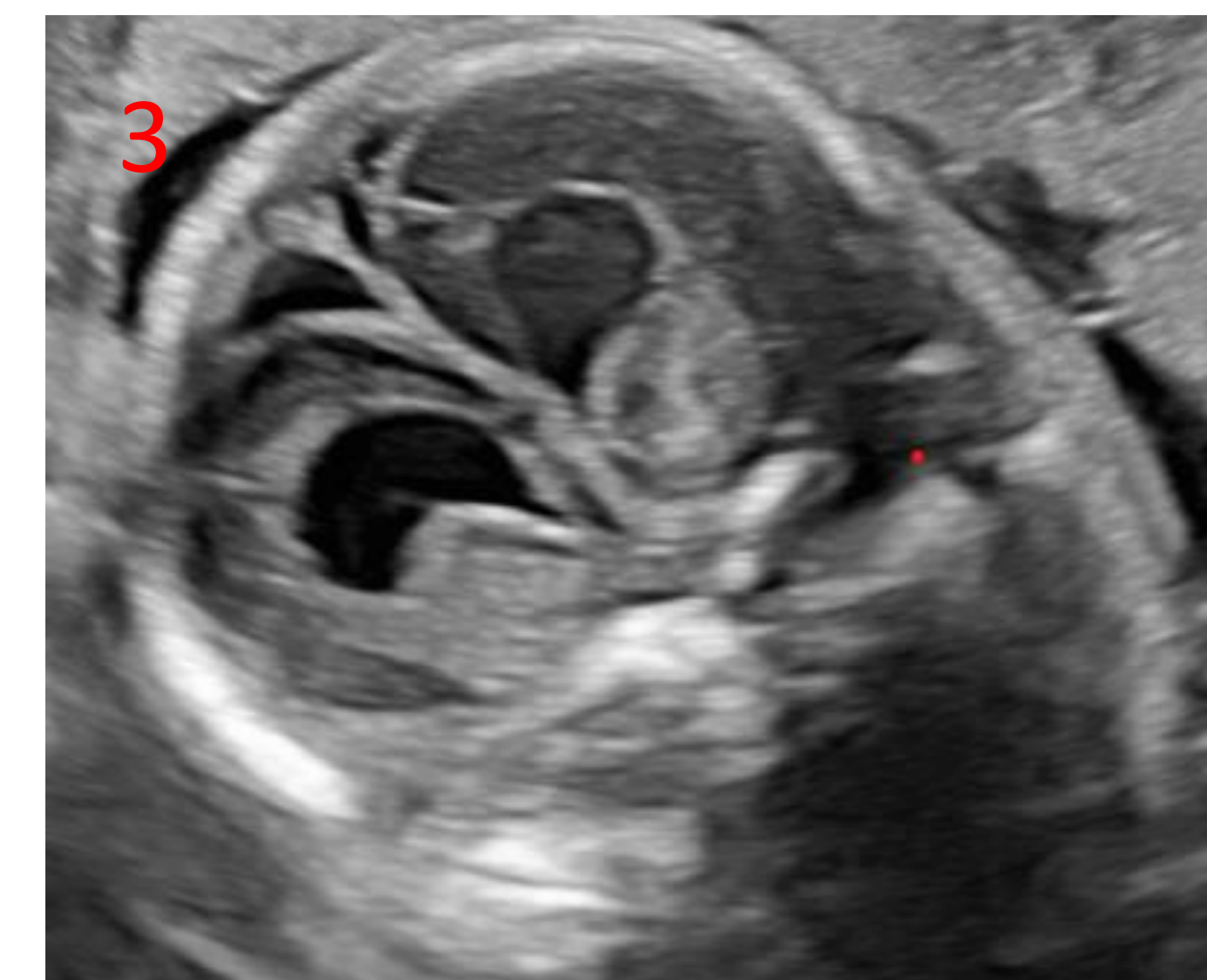


Figure 3
Abnormal shape of anterior horn of bilateral ventricle

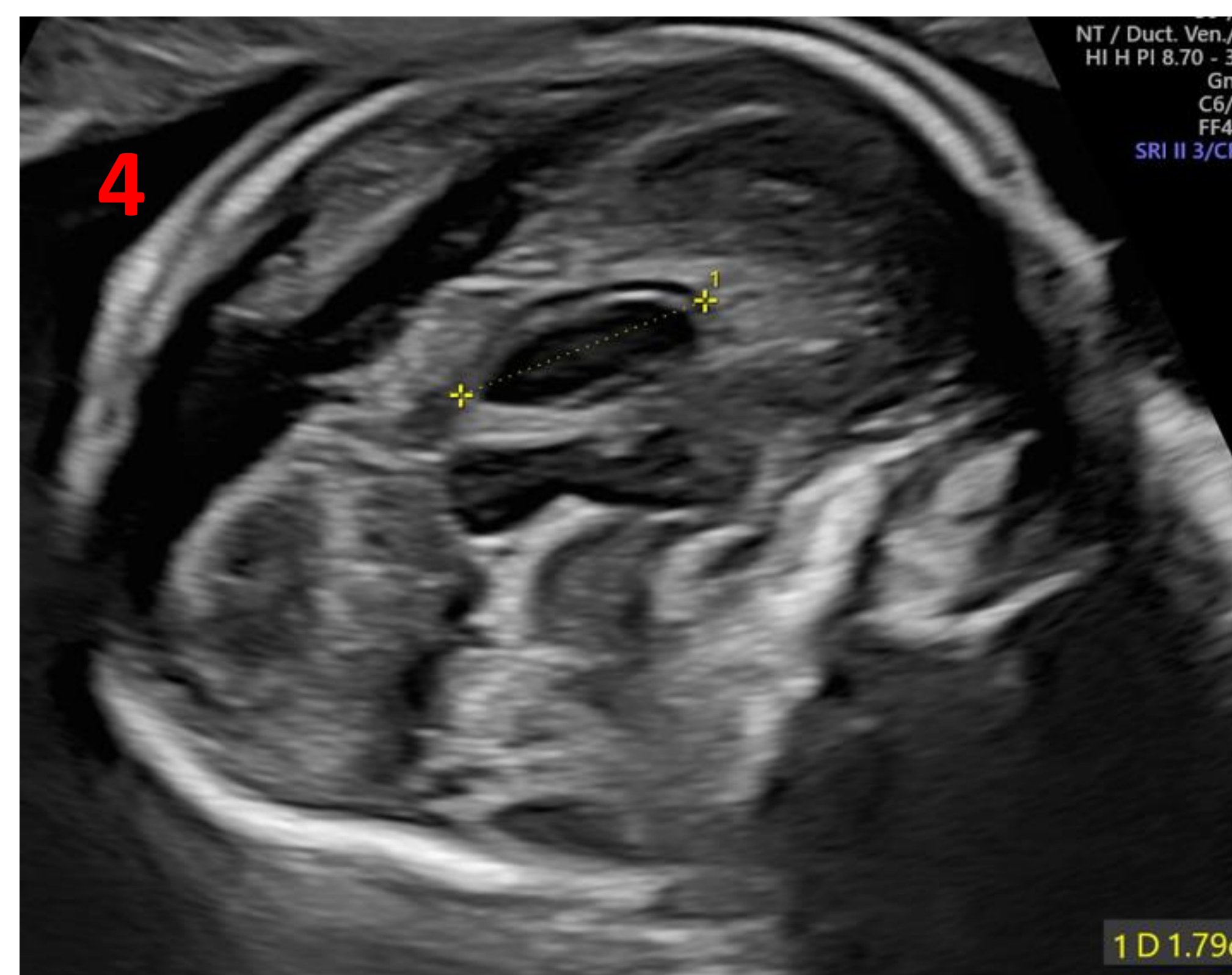


Figure 4
Short (17.9 mm) and thin corpus callosum suspected CC hypoplasia

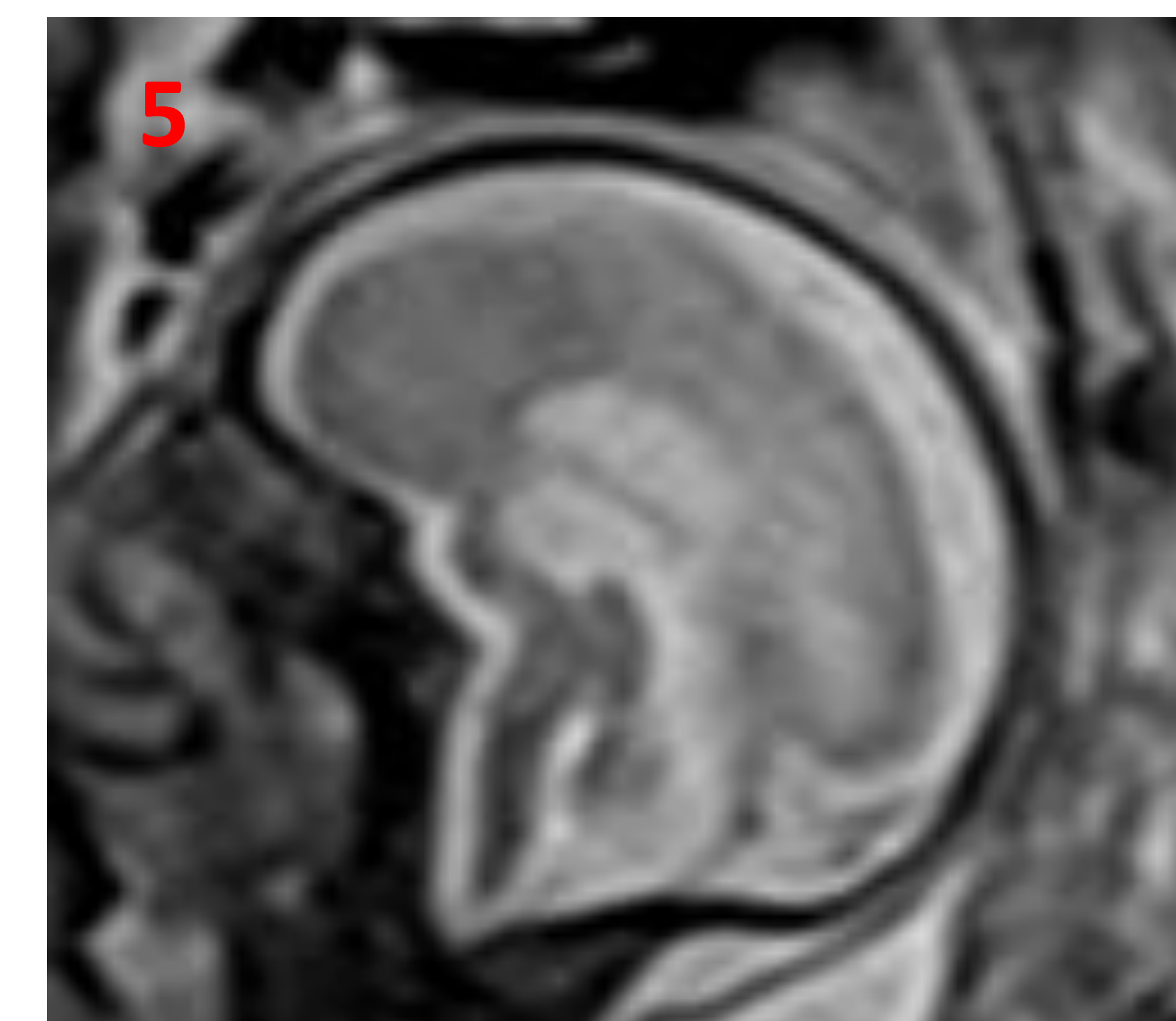


Figure 5
Flat pons suspected pontine hypoplasia