OBJECTIVES: The tubulinopathies are the congenital CNS malformations related to abnormal cortical development caused by a mutation in one of the seven genes encoding different isotypes of tubulin protein responsible for neuronal migration in the developing fetal brain.

METHODS: Primigravida with unremarkable history at 26 + 2/7 weeks was referred to our unit due to ventriculomegaly on the 2nd trimester anatomy scan. Further investigation was done by fetal MRI that revealed complex brain malformations. The pregnancy was terminated at 28 weeks without genetic testing. However, all the characteristic neuroimaging signs conformed with TUBB2B mutation.

RESULTS: Dedicated neurosonographic examination identified mild ventriculomegaly (13 mm) with abnormal shape, slightly asymmetric, which were dilated and angulated and also 3rd ventricle dilatation. The ventricular walls were mildly irregular, a defused brain anomaly was identified with callosal dysgenesis (short and thin CC lacking the rostrum, part of the genu and the splenium) and brain asymmetry was observed, not in size but regarding to the gyri and sulci distribution on the brain surface. The Sylvian fissures were quite asymmetric and operculization was abnormal in both sides. The calcarine sulci were absent and the parieto-occipital were shallow. In coronal view aberrant frontal sulcation was observed unilaterally. These findings were hard to classify, but met the criteria for disgyria with suspected areas of PMG. Additionally, the basal ganglia were very dysmorphic with enlarged caudates and a big massa intermedia. The brainstem was hypoplastic and kinked and the basilar pons prominence was flattened. The midbrain tegmentum was thin, the tectum was enlarged and the aqueduct obstructed distally. The TCD was normal, but vermis was dysgenetic or possibly absent.

CONCLUSION: These findings of abnormal gyral pattern, callosal dysgenesis, kinked brainstem and particularly persistent ganglionic eminences are compatible to tubulinopathy, possibly due to TUBB2B mutation as proposed by Mutch and coworkers.