Objective: It is well-known that holoprosencephalic malformations are related to midline defects in fetal face. The aim of this study is to analyze the frequency and types of the facial malformations in fetuses diagnosed with holoprosencephalic defects, evaluated by fetal MRI.

Methods: A review of 16 cases with central nervous system malformations referred for fetal MRI and diagnosed with holoprosencephalic malformations from 2006 and 2016 were included.

Results: The median age of the mothers was 34 years (20-50). The fetal MRI examination was performed at the 27th week (17-34). The cases of holoprosencephaly were identified as 12 Alobar, 2 Lobar and 2 Semilobar. Using the DeMyer classification of midline facial malformations, the cyclopia with/without ocular fusion with proboscide was the most common malformation (31%). Cebocephaly and median cleft with arhinencephaly had both 25%. Ethmocephaly was present in 12.5% and normal face in 6% of the cases. The most common form, Alobar holoprosencephaly, presented 36% of cases with the cyclopia with/without ocular fusion with proboscide, 9% of ethmocephaly, 36% presenting cebocephaly and 18% presenting median cleft with arhinencephaly.

Conclusion: In holoprosencephalies phenotypes, the most common malformation is cyclopia with/without ocular fusion with proboscide followed by Cebocephaly, median cleft with arhinencephaly and Ethmocephaly.