# A Very Rare Case of Colpocephaly Associated With Trisomy 18

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We presented a very rare case of colphocephaly which is a term used to describe a congenital abnormal enlargement of the occipital horns of the lateral ventricules associated with normal frontal horns. A variety of cranial abnormalities and syndromes were associated with colphocephaly such as such as lissencephaly type 1, linear nevus sebaceous syndrome, Marden-Walker syndrome, tourette syndrome, Aicardi syndrome, trisomy 8 mosaic, trisomy 9 mosaic, Norman-Roberts syndrome, Zellweger syndrome, Nijmegen breakage syndrome, hemimegalencephaly, Chudley-McCullough syndrome; but association of colphocephaly with trisomy 18 case has not been identified up to now.

**Key Words:** Colphocephaly, Trisomy 18, Cranial-abnormalities, Syndrome

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## Introduction

Colpocephaly is a term used to describe a congenital abnormal enlargement of the occipital horns of the lateral ventricules associated with normal frontal horns.In 1940 Benda first recognized the "failure of decrease in the size of the primitive brain vesicles" in a mentally retarded boy with epilepsy and microcephaly who, on neuropathological examination, also had an absent corpus callosum, micro- and macrogyria, and gray matter heterotopia. He used the term "vesiculocephaly" for this ventricular configuration.[1] Afterwards In 1946 Yakovlev and Wadsworth used the term colpocephaly instead of vesiculocephaly (from the Greek "kolpos," meaning hollow) to "avoid the miscegenation of the Latin and Greek roots".[2] Later, the term "colpocephaly" was restricted to the persistence of a specific form of fetal ventricular configuration into postnatal life where occipital horns of the cerebral ventricles remain disproportionately large and dilated.[3]

It results from underdevelopment or lack of thickening of the white matter in the posterior cerebrum.The pathophysiology is attributed to the white matter development arrest that occuring between second and fifth month of intrauterine life.

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A variety of cranial abnormalities are seen in trisomy 18 fetuses however colpocephaly finding has not been identified until now.

## Case Report

A 23-year-old, gravida 2, para 1, woman admitted to our hospital at 17 weeks of gestation for routine prenatal visit.Obstetric ultrasonography revealed fetal bilateral colpocephaly, atrioventricular septal defect (AVSD), nasal bone hypoplasia.Other fetal intracranial structures were found to be normal at ultrasonography (Figure 1).

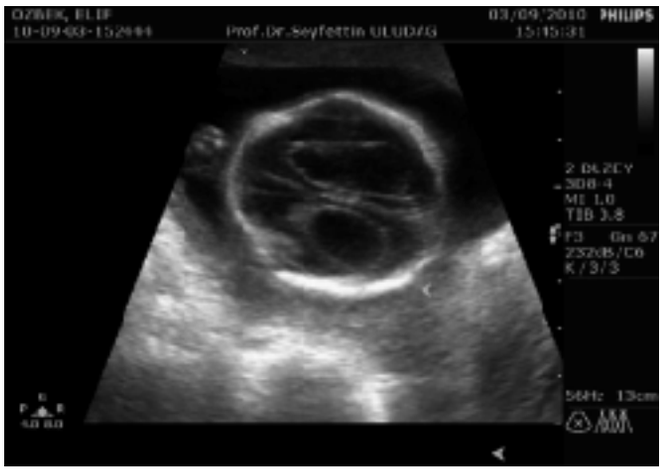


*Figure 1: Ultrasonographic view of cranial structures in colphocephaly(bilateral dilatation of posterior horns of lateral ventricule).*

Further investigation with fetal echocardiography and fetal magnetic resonance imaging (MRI) were performed. The pa-

tient was scanned for the intrauterine infections in terms of toxoplasmosis, rubella, cytomegalovirus and herpes virus.The infection panel was found to be negative. Fetal MRI findings confirmed colpocephaly and bilateral ventriculomegaly.Spina bifida was not found. Atrioventricular septal defect and atrioventricular valf insufficiency was identified on fetal echocardiography. Amniocentesis was performed and fetal karyotype was found to be trisomy 18. The patient was informed about the prognosis of trisomy 18 and associated anomalies, and medical abortion was performed.

## Discussion

Trisomy 18 is the second most common autosomal trisomy after trisomy 21.The incidence of trisomy 18 varies from approximately 1 in 3000 to 1 in 7000 livebirths.A variety of CNS malformation including choroid plexus cysts, meningomyelocele,cerebellar hypoplasia, absence of corpus callosum,microcephaly, enlarged cisterna magna has been defined.Beside from these anomalies the colpocephaly finding is the first identified case in association with trisomy 18.

It can be presented as a component of multiple disordes with diverse etiologies, such as chromosomal anomalies like trisomy 8 or trisomy 9 mosaicism; intrauterine infections like toxoplasmosis; maternal drug usage such as salbutamol,theophylline; central nervous system anomalies.[3,4]

Since the first description in 1940 of Benda, there have been approximately 50 cases have been reported. Colpocephaly, most commonly recognized on radiological investigations can occur as an isolated disorder or can be associated with several conditions, such as lissencephaly type 1, linear nevus sebaceous syndrome, Marden-Walker syndrome, tourette syndrome, Aicardi syndrome, trisomy 8 mosaic, trisomy 9 mosaic, Norman-Roberts syndrome, Zellweger syndrome, Nijmegen breakage syndrome, hemimegalencephaly, Chudley-McCullough syndrome.[3,4]

The clinical picture is highly variable, depending on etiology and associated conditions.It has also associations with several central nervous system malformations.Although the most common CNS malformation associated with colpocephaly is agenesis of corpus callosum; neuronal migration disorders (lissencephaly, pachygyria), macrogyria, microgyria, schizencephaly, enlargement of cisterna magna, optic nerve hypoplasia, meningomyelocele and hydrocephalus can be seen concomitant with this situation. It is usually seen bilaterally but in some cases it can be found unilaterally. Unilateral colpocephaly is a characteristic finding in the enlarged hemisphere in hemimegalencephaly.[5] Another cause of unilateral colpocephaly is an uncommon form of familial porencephaly.[6] The finding may be indirectly suggested on ultrasound by the so called lemon sign, which occurs due to depression of the calvarium at the bilateral frontal suture lines, giving the calvarium the appearance of a lemon.

Colpocephaly is often confused with hydrocephalus. In the fetus, it is commonly described as hydrocephalus or ventriculomegaly.[4] However, they are different anomalies and should be distinguished. Colpocephaly refers to the selective dilatation of the occipital horns with normal or small frontal horns and 3rd ventricle. The distinction is important because hydrocephalus usually requires shunting, affects all the ventricular system, and often is progressive and obstructive, while colpocephaly is an occipital selective, nonprogressive and nonobstructive condition that does not require surgical treatment.It can also be confused with the arachnoid cysts Arachnoid cysts located within the interhemispheric fissures are rare, frequently associated with agenesis or hypogenesis of corpus callosum.[7] Hence the treatment and associated anomalies are different; MRI can be valuable diagnostic tool for the differentiation of the intracranial cystic lesions.

Prenatal detection of the chromosomally abnormal fetus is an important clinical goal in modern prenatal care. Papp et al. aimed at their study to determine the incidence of ultrasound findings in common fetal chromosomal defects.During their study, 70 trisomy 18 fetuses were diagnosed prenatally. The total number of fetuses that showed sonographic signs (either 'major' or 'minor') was 66 (94.3%), whereas major anomalies were seen in 61 (87.1%) fetuses. Among the major fetal defects cardiac anomalies were the most common (47.1%, 33/70) with a 27.1% rate of VSD. CNS anomalies were seen in 35.7% (25/70) of the cases. Fossa posterior cyst (14.3%) and abnormal head shape ('strawberry' or 'lemon' shape) was the most frequently detected anomalies of this group (12.9%). [8] In our case, we identified atrioventricular heart defect and a concomitant CNS anomaly colpocephaly as lemon sing. More than 90% of patients with trisomy 18 have congenital heart disease (CHD). The most common cardiac lesions are atrial septal defect, large ventricular septal defect, patent ductus arteriosus, tetralogy of Fallot and polyvalvular disease.[9,10]

Although colpocephaly seems to be rare, when seen it must be taken into account and examination of the fetal heart is important.Morover prenatally chromosomal detection is also crutial. Colpocephaly can be an initial finding of trisomy 18 fetuses. Although the congenital heart defects seen most of the trisomy 18 patients, a rare cranial anomaly named colpocephaly must be evaluated in those population.

Our case; is the first case that an uncommon colpocephaly finding concomitant with trisomy 18 fetus.

## Trizomi 18 ile Birlikte Nadir Bir Kolposefali Vakası

Bu yazıda, lateral ventriküllerin arka boynuzlarının konjenital olarak anormal genişlediği ve ön boynuzların normal olduğu, nadir bir kolposefali vakasını sunduk.Kolposefaliye eşlik eden çeşitli kranial anormallikler ve sendromlar bulunmaktadır; bunlar arasında lissensefali tip 1, lineer nevus sebaceous sendromu, Marden-Walker sendromu, tourette sendromu, Aicardi sendromu, trisomy 8 mozaisizm, trisomy 9 mozaisizm, Norman - Roberts sendromu, Zellweger sendromu, Nijmegen kırılma sendromu, hemimegalencefaly, Chudley-McCullough sendromu sayılabilir, fakat kolposefalinin trisomi 18 ile birlikteliği şuana kadar tespit edilmemiştir.

**Anahtar Kelimeler:** Kolposefali, Trisomi 18, Kranial anormallikler, Sendrom

## References

1. Benda CE.1940. Microcephaly. Am J Psychiatry 97:1135-1146
2. Yakovlev PI, Wadsworth RC.1946.Schizencephalies: a study of the congenital clefts in the cerebral mantle: I. Clefts with fused lips. J Neuropathol Exp Neuro 5:116-130
3. Garg BP. 1982.Colpocephaly: an error of morphogenesis. Arch Neurol 39:243-246
4. Puvabanditsin S, Garrow E, Ostrerov Y, Trucanu D, Ilic M, Cholenkeril JV. Colpocephaly: a case report . Am J Perinatol 2006;23:295-7.
5. Sarnat HB, Flores-Sarnat L. Molecular genetic and morphologic integration in malformations of the nervous system for etiologic classification.Semin Pediatr Neurol 2002;9:335-44.
6. Aguglia U, Gambardella A, Breedveld GJ, Oliveri RL, Le Piane E, Messina D, Quattrone A, Heutink P. Suggestive evidence for linkage to chromosome 13qter for autosomal dominant type 1 porencephaly. Neurology 2004;62:1613-5.
7. Ulu MO, Kafadar AM, Dashti R, Işler C, Uludağ S, Erdinçler P. Treatment of symptomatic interhemispheric arachnoid cysts by cystoperitoneal shunting. J Clin Neurosci 2010;17:700-5.
8. Papp C, Szigeti Z, Tóth-Pál E, Hajdú J, Joó JG, Papp Z. Ultrasonographic findings of fetal aneuploidies in the second trimester--our experiences. Fetal Diagn Ther 2008; 23:105-13
9. Van Praagh S, Truman T, Firpo A, Bano-Rodrigo A, Fried R, McManus B, Engle MA, Van Praagh R. Cardiac malformations in trisomy-18: a study of 41 postmortem cases. J Am Coll Cardiol 1989;13:1586-97.
10. Musewe NN, Alexander DJ, Teshima I, Smallhorn JF, Freedom RM. Echocardiographic evaluation of the spectrum of cardiac anomalies associated with trisomy 13 and trisomy 18. J Am Coll Cardiol 1990;15:673-7.