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*Brief Report*

# Ventricular septal defect closure in a patient with achondroplasia

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**Abstract** Achondroplasia with co-morbid CHD is rare, as are reports of surgical treatment for such patients. We present the case of a 13-year-old girl with achondroplasia and ventricular septal defect. Her ventricular septal defect was surgically repaired focussing on the cardiopulmonary bypass flow, healing of the sternum, and her frail neck cartilage. The surgery and recovery were without complications.

Keywords: Ventricular septal defect; achondroplasia; congenital heart surgery

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Achondroplasia is characterised by a lack of growth of cartilage. This condition causes short stature, depression of the nose, and spinal deformities. Achondroplasia is often caused by a genetic anomaly and is the most common cause of dwarfism. The estimated incidence of achondroplasia is approximately 0.36–0.60/10,000 live births.[1] Cases of achondroplasia and CHDs are rare, as are reports of the surgical treatment for such patients. Very few reports exist on cardiac surgery in patients with achondroplasia. In addition, to the best of our knowledge, there is no report of ventricular septal defect repair using cardiopulmonary bypass in a patient with achondroplasia. We therefore present the first known case.

## Case report

The patient was a 13-year-old girl (weight, 27 kg; height, 110 cm) who had been diagnosed at birth with achondroplasia on the basis of short stature, lateral curvature of the spine, and depression of the base of the nose. She was diagnosed as having a small ventricular septal defect at birth and was admitted to the hospital for the surgical treatment of this defect to prevent infective endocarditis and aortic regurgitation due to right coronary cusp prolapse at 13 years of age. On echocardiography, a ventricular septal defect of $4 \times 5$ mm was detected, along with a membranous septal aneurysm. Catheter examination revealed no pulmonary hypertension, small shunt ratio, $Qp/Qs = 1.16$, right coronary cusp prolapse without aortic regurgitation, and no other cardiac anomaly. Her body surface area was 0.91 $m^2$.

Surgical repair was performed via a median sternotomy. Cardiopulmonary bypass was established after heparinisation just before cannulation, and the right atrium was opened under mild hypothermic chemical cardiac arrest. The ventricular septal defect was observed to be surrounded by membranous fibrous tissue, and the origin of the large hole was below the membranous aneurysm. Patch closure was performed using a 0.4-mm polytetrafluoroethylene patch with eight stiches. Total bypass time was 72 minutes, and the cross-clamp time was 50 minutes. After re-warming, the patient was easily weaned from cardiopulmonary bypass without arrhythmias, and the duration of the surgery was 192 minutes. The postoperative haemodynamic state was stable. The postoperative course of the patient was uneventful, and early postoperative mobility was applied. She discharged on postoperative day 5.

## Discussion

In our patient, surgical repair for the ventricular septal defect was performed with the following three points in mind: fixation and repair of the sternum,

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cardiopulmonary bypass during surgery, and maintenance of anaesthesia.

Achondroplasia is considered to be caused by a mutation affecting the fibroblast growth factor receptor gene 3.[1] This gene affects only cartilage formation, not osteogenesis, which made operating on the sternum easier in our case. Median sternotomy was performed with two wires fixed at the top of the sternum and four CV-0 sutures. After surgery, healing of the sternum was uneventful and was completed within 2 months.

Praveen et al reported on successful atrial septal defect closures using cardiopulmonary bypass. The flow during cardiopulmonary bypass was based on the body surface area, which was calculated using the Mosteller formula, age, and weight nomograms.[2] We calculated the patient's body surface area on the basis of her height and weight only, and we did not take age into consideration. The necessary flow rate was determined using an index from 2.6 to 2.8 × (body surface area) L/min. The primary diagnosis did not influence the management strategies; however, this did not cause any complications related to the cardiopulmonary bypass.

Ivol et al stated that in patients with achondroplasia neck manipulation, particularly flexion, should be avoided because of the potential risk of cervical cord compression and instability.[3] We therefore performed a two-step ventilation: one jaw thrust and one mask-to-face ventilation were performed. A monitoring line was attached, as per normal procedure. Camera-supported intubation was used to avoid neck extension. In the postoperative phase, the neck position was carefully monitored and early extubation was performed.

In conclusion, we performed surgical repair of a ventricular septal defect in a patient with achondroplasia syndrome without any complications.

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## Conflicts of Interest

None.

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