A Rare Case of Percutaneous Coronary Intervention in Achondroplasia

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ABSTRACT: Achondroplastic individuals are associated with increased cardiac risk when compared to the general population. Coronary interventions in patients with achondroplasia have not been studied previously. We report the case of a 32-year-old male smoker with achondroplasia who presented with acute chest pain of 3 hours duration. He was diagnosed with acute inferior and right ventricular myocardial infarction and thrombolyzed with streptokinase. Since the patient continued to have pain and hypotension, he was taken for rescue percutaneous coronary intervention (PCI). Because of short stature and kyphoscoliosis, difficulties were faced in cannulating the coronaries and performing intervention. He had total occlusion of proximal right coronary artery, for which angioplasty with stenting was done. To our knowledge, this is the first case of PCI conducted in an achondroplastic patient.

Key words: achondroplasia, percutaneous coronary intervention

Achondroplasia is an autosomal dominant dwarfing condition characterized by macrocephaly and rhizomelic shortening of the extremities.1 It is caused by a mutation affecting the fibroblast growth factor receptor (FGFR) gene 3. The estimated prevalence is 1:25,000 in the general population.2 The overall heart disease mortality rate is over two-fold greater than that of the general population.2 No literature exists about the coronary interventions on this group of patients. Hereby, we report the case of an achondroplastic male who presented with acute inferior and right ventricular myocardial infarction and underwent successful percutaneous transluminal coronary angioplasty with stenting to the right coronary artery (RCA).

Case Report. A 32-year-old Asian-Indian male with achondroplasia presented with acute-onset chest pain of 3 hours duration. His height was 87 cm, his weight was 44 kg, and he had kyphoscoliosis (Figures 1 and 2; Video 1). He was a chronic smoker, but had no history of diabetes mellitus, hypertension, or hyperlipidemia. The electrocardiogram showed ST elevations in inferior and right ventricular leads (Figure 2). Echocardiogram revealed inferior wall hypokinesia with biventricular dysfunction. He was diagnosed with acute inferior and right ventricular myocardial infarction and advised to undergo primary percutaneous coronary intervention (PCI). Initially, he did not give consent for the procedure, so he was thrombolyzed with 1,500,000 U of streptokinase. He was also treated with aspirin, clopidogrel, atorvastatin, intravenous fluid, and unfractionated heparin. In spite of thrombolysis, the patient continued to have chest pain with persistent ST elevation and hypotension requiring inotropic support. He was then taken for rescue PCI after explaining the risk and getting consent. He was preloaded with 600 mg of clopidogrel. During coronary angiogram, there was difficulty in cannulating the coronaries due to short stature and kyphoscoliosis (Figure 3; Video 1). Angiogram was done via right femoral artery approach using 6 Fr, 3.5 curve Judkins left and right catheters. After several manipulations, cannulation was done keeping the major portions of the catheters outside the body. Angiogram showed total occlusion of the proximal RCA with normal left coronary system (Figure 4; Videos 2 and 3). The RCA was recannulated with 6 Fr, 3.5 curve Judkins right guiding catheter. Initial crossing of the lesion with guidewire alone was unsuccessful. 0.014” Floppy II extra-support guidewire with the support of a 2.5 x 12 mm semicompliant balloon was then used to cross the lesion. The lesion was predilated with the same 2.5 x 12 mm balloon at 6 atm (Figure 5; Videos 5

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Figure 1. Achondroplastic patient measuring 87 cm.
A 3 x 30 mm bare-metal stent was deployed across the lesion at 12 atm (Figure 5; Videos 7 and 8). Postdilatation was done sequentially using 3.5 x 10 mm non-compliant balloon from 14-20 atm (Figure 6; Videos 9). Distal TIMI 3 flow was achieved at the end of the procedure (Figure 6; Videos 10 and 11). Postprocedure chest pain subsided and resolution of ST elevation was noted. Blood pressure stabilized and inotropes were gradually tapered off. Ventricular function improved. Aspirin 150 mg and clopidogrel 75 mg were given as daily maintenance antiplatelet drugs. Later course in the hospital was uneventful and he was discharged in a stable condition on day 5 of hospitalization. After discharge, the patient was placed on aspirin, clopidogrel, statin, beta-blocker and ACE inhibitor. The patient was followed regularly; after 6 months, he was asymptomatic with normal ventricular function.

Discussion. Achondroplasia is the most common disproportionate skeletal dysplasia seen in human beings. The risk of heart disease-related death is increased two-fold in these individuals and is highest in the male population, where it is nearly three-fold higher than the general population. Apart from the traditional risk factors, there seems to be other factors responsible for this increased risk. This could be genetic in nature and no large cohort studies have been done to delineate these additional factors. No studies have been done regarding the technical aspects of coronary intervention in this group of individuals. There are a few reports of coronary artery bypass surgery in these patients, but reports on PCI have not been documented in the literature. To our knowledge, this is the first case report of PCI in an achondroplastic patient. Because of the short stature of the patient and associated kyphoscoliosis, we encountered technical difficulties when first attempting to cannulate the coronary arteries. Crossing the lesion, which was to-
tally occluded, was also challenging. Further studies need to be done regarding the appropriate development of hardware and new techniques in this subgroup of patients. Our case demonstrates that patients with achondroplasia can safely undergo coronary angioplasty without additional risk.

Conclusion. PCI can be safely performed in patients with dwarfism due to achondroplasia, despite the fact that little information dealing with this issue is available in international literature. It is interesting to note that no similar case reports have been published. Further studies need to be done to improve the technical aspects of the procedure in these patients.

References