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Inferior myocardial infarction after blunt chest trauma

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A 44 year-old male presented with substernal chest pain a few minutes after falling on his chest while playing hockey. He thought that his pain was due to the fall so he stopped playing. The pain only worsened. The patient has a history of hypertension controlled with 50 mg daily of lisinopril and hyperlipidemia treated with fenofibrate 200 mg daily. His family history is significant for coronary artery diseases in his brother and his grandfather who died at 42 and 50 years of age, respectively, due to myocardial infarction. When his pain became unbearable, the patient drove himself to an urgent care where a 12-lead EKG showed a STEMI on inferior leads. The patient was transferred to our hospital where an emergent coronary angiogram showed a hyperdominant right circulation with a fresh clot completely obstructing the mid RCA, for which throbectomy trials have failed. The patient then underwent an emergent CABG with thrombus removal and a dissection was noted. The patient was admitted to the coronary care unite for observation, his hospital course was complicated by right ventricular failure (EF=40%), atrial fibrillation and hypotension. He was stabilized and discharged on post operative day 11. Trauma induced myocardial infarction is a rare clinical entity. The diagnosis and management is oftern delayed, therefore, outcomes are suboptimal. Every effort should be made to avoid such a delay, and this condition should be kept in mind, even in the light of polytrauma.

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Truncation and microdeletion of EVC2 with a novel EFCAB7 missense mutation in rare syndromic congenital heart defects: Ellis-Van Creveld syndrome

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Ellis-van Creveld syndrome (EvC) belongs to ciliopathy with cardiac anomalies, disproportionate short stature, polydactyly, dystrophic nails and oral defects. Approximately 60% of EvC patients had severe Congenital Heart Defects (CHD), in which more than half are atrio-ventricular septal defect and common atrium, which leads to early childhood mortality. However, the EvC phenotype overlaps with other ciliopathies, which hampers the accurate diagnosis. To elucidate the genetic characteristics we screened *EVC/EVC2* mutations in 8 Vietnamese EvC patients. All showed CHD. One had compound heterozygous *EVC2* mutations: a novel mutation c.769G>T-p.E177X in exon 6 inherited from father and another previously reported c.2476C>T-p.R826X mutation in exon 14 inherited from mother. The *EVC2* mRNA expression level was significantly lower in the patient and her parents compared to those in the controls. Another case had a novel heterozygous *EVC* mutation (c.1717C>G-p.S572X) in exon 12, inherited from his father. Of note, the mother without any *EVC* mutation on Sanger sequencing showed a lower expression level of *EVC* mRNA compared with controls. SNP array analysis revealed that the patient and mother had a heterozygous 16kb deletion in *EVC*. This patient also had a heterozygous novel variant in exon 9 of *EFCAB7* (c.1171T>C-p.Y391H), inherited from his father. The patient had atypical phenotype, which suggested that EFCAB7 may modify the cardiac malformations by tethering with EVC. In conclusion, we detected two novel nonsense mutations and a partial deletion of *EVC/EVC2* in two families diagnosed as EvC. The relative expression of *EVC/EVC2* mRNA was reduced in these members, which revealed that these mutations were disease-causative. Moreover, we showed a possible modifier gene mutation in one family with EvC.

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