Author’s response to reviews

Title: Identification of a Novel CTCF Mutation Responsible for Syndromic Intellectual Disability - A Case Report

Authors:

Fatma Bastaki (FABastaki@dha.gov.ae)
Pratibha Nair (pratibha.nair@gmail.com)
Madiha Mohamed (mfmohamed@dha.gov.ae)
Ethar Malik (emmalik@dha.gov.ae)
Mustafa Helmi (mahelmi@dha.gov.ae)
Mahmoud Al-Ali (mtalali@gmail.com)
Abdul Rezzak Hamzeh (abdul.hamzeh@hmaward.org.ae)

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Dear Editor,

Thank you for the latest decision letter with the two points that need to be further clarified. Below are our responses to these points and the corresponding changes were made in the manuscript in red.

Best regards,
Comments and [Responses]:

1. Usually, filtering for population frequencies is not sufficient … Please specify your used filter criteria in more detail. [This point is clarified at the end of paragraph 1 in Materials & Methods].

2. Although the mutation in CTCF is convincing this does not exclude a pathogenic contribution of an additional variant. As mutations in SETD5… You should test it in the parents. If it is inherited it can be excluded. If it is de novo, too, a contribution to your patient's phenotype has to be discussed. [The patient inherited the mutation (c.3221G>A; p.R1074Q) from the father and this is added to the penultimate paragraph in Discussion].