Reviewer's report

Title: A new case of de novo 6q24.3 Deletion on Paternal Chromosome 6 with Growth Hormone Deficiency: a twelve years follow-up

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Reviewer: Pawel Stankiewicz

Reviewer's report:

In the manuscript “A new case of de novo 6q24.3 Deletion on Paternal Chromosome 6 with Growth Hormone Deficiency: a twelve years follow-up”, Stagi et al. describe an apparently de novo 11 Mb deletion on paternal chromosome 6q24.2q25.2 identified in a 6-year-old boy with IUGR, postnatal growth failure with growth hormone deficiency, mild developmental delay, delayed bone age, hypophyseal hypoplasia, bilateral pseudopapilledema, hyperopic astigmatism, dysmorphic features, hypoplastic left colon, heart defects, GERD, and bilateral inguinal hernia. The Authors correlate the patient’s phenotype with the maternally imprinted PLAGL1 (ZAC1) gene on 6q24.2 and review the literature. They also propose to name this deletion “Nowaczyk syndrome”.

The manuscript adds to the literature. The data sound and well controlled.

Major Compulsory Revisions

The Authors should also discuss the overlapping deletions from Decipher.

Was the paternal sample analyzed to exclude a balanced insertional translocation?

Title
6q24.3 should be changed to 6q24.2-q25.2

Abstract
Deletions on the long arm of chromosome 6 are relatively uncommon, and only a small number occur on the paternal copy, causing growth deficit.

This sentence can be misleading. The Authors refer only to the distal portion of 6q.

Birth weight was 1740 gr (-1.6 SDS), the length was 43.5 cm (-1.1 SDS), and the head circumference was 32.2 cm (0.0 SDS).

At the age of 3.75 years, his height was 88.2 cm (-2.98 SDS), weight 11.500 kg (-3.34 SDS), and his body mass index was 14.78 (-0.81 SDS).

Please delete these sentences from Abstract.

Figure 1
The facial phenotype is not well shown with the bars over the eyes, so the similarities with the reported cases cannot be well judged.

Minor Essential Revisions

Mental retardation should read intellectual disability.

Case presentation

“Prenatal ultrasounds … paternal disomy.”

Please move this para before “His birth was…”

Was cardiomegaly followed postnatally?

Satellite analysis of chromosomes 2, 11, and 16 showed biparental inheritance, The Authors probably mean microsatellite analysis. Why was UPD testing performed for chromosomes 2, 11, and 16?

At 2 years of age, he underwent a left inguinal herniotomy.

What about the right hernia?

Figure 2 is poorly explained in its legend. I suggest showing a schematic representation of this chromosomal region with other reported overlapping deletions depicted.

Also, please provide the deletion coordinates in the text or figure legend. It is unclear whether PLAG1 is deleted in the patient.

Figure 3 should be moved to Supplement.

Discretionary revisions

Abstract

Please spell out STR

Background

Kumar and Nowaczyk should read Kumar et al. [13] and Nowaczyk et al. [14]

Case presentation

1740 gr

Change to

1740 g

Level of interest: An article whose findings are important to those with closely related research interests

Quality of written English: Needs some language corrections before being published
Statistical review: No, the manuscript does not need to be seen by a statistician.

Declaration of competing interests:
I declare that I have no competing interests