Reviewer's report

Title: Genotype-phenotype correlation in 22q11.2 deletion syndrome

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Reviewer: Anne Slavotinek

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The authors describe testing 142 patients referred for evaluation for 22q11 deletion syndrome and analyze the phenotypic features in the 110 individuals who were 22q11 deletion positive by MLPA/FISH. This is a well written paper, although not exceedingly novel, and I have only minor comments.

Minor points:
“Mean age 16.1±9.7;” - you should state months or years
“We elaborate below on the clinical features of this patient because it carried a rare deletion with only two other similar cases in the literature” – she rather than it
“T. was referred to our clinic” – patient initials should not be used.
“Already in preschool she was diagnosed with…” – please state age
“VSD” – please spell out; same for “VIQ”
“Thus the MLPA is clearly superior to FISH as a diagnostic tool and a useful method for deletion characterization, essential for genotype/phenotype analysis.” – you should mention that array is another technique that would have detected an atypical deletion although costs of performing it maybe higher.
“….and the high throughput methods of the near future e.g..” – can remove near future, as the methods are already used.