Reviewer's report

Title: Adult phenotype and further phenotypic variability in SRD5A3-CDG

Version: 2 Date: 23 July 2013

Reviewer: Lynne A Wolfe

Reviewer's report:

Minor Revisions:
1. Mild intellectual disability seems inappropriate for a 40 yr old described as requiring assistance for ADL's. Was formal Neuropsyc testing accomplished?
2. Skeletal survey content would be important information since subtle bone abnormalities has been reported in other pts with SRD5A3-CDG. A Table comparing these sibs to other SRD5A3-CDG as well as to the Kahrizi cohort would improve ease of understanding the phenotype.