



Supp. Fig. 7: Characterization of iPSC derived neurons with and without 1q21.1 CNV . **A** The expression of NESTIN (NES), PLZF and ZO1 mRNA at day 20 and 50 of neuronal differentiation. The time point samples were taken had a significant effect on expression of the three markers (Control n=3, $F_{1,30}=23.2$; $P<0.0001$;). **B** Fold change of NPC markers in 1q21.1 deletion and duplication cell lines after 20 days of differentiation. Data was analyzed using multiple T-Tests. Stars represent Holm-Sidak corrected p-values. **C,D** Expression of Ki67 mRNA after 20 and 30 days of neuronal differentiation (n≥3). **E** The expression of DCX, NCAM and MAP2 mRNA at day 20 and 50 of neuronal differentiation. The time point samples were taken had a significant effect on expression of the three markers (Control n=3; $F_{1,30}=98.4$; $P<0.0001$). Data sets were analyzed by two-way ANOVA with post hoc comparisons comparing to day 20 samples. Stars above points represent Sidak-corrected post hoc tests. **F** Fold change of neuronal markers in 1q21.1 deletion and duplication cell lines after 50 days of differentiation. Data was analyzed using multiple T-Tests. Stars represent Holm-Sidak corrected p-values. **G** Expression of cortical layer markers in control day 50 cultures (n=3). **H** Expression of cortical layer markers in 1q21.1 deletion and duplication day 50 cultures (n=3). Fold change is normalized to control day 50 differentiations. Data was analyzed using multiple T-Tests. Stars represent Holm-Sidak corrected p-values. All data presented as means \pm SEM, * $P<0.05$, *** $P<0.001$, **** $P<0.0001$. **I** The expression of GFAP and S100β was significantly minimal in comparison to MAP2 in D40 neuronal cultures. The level of GFAP and S100β was comparable across the experimental group. **J** Similarly, the expression of GFAP and S100β in comparison to MAP2 was significantly reduced in D50 neuronal cultures (n=3). Data was analysed using Students T-Tests. **K** Bar graph showing level of TUJ1 expression across group at day 50. All data presented as means \pm SEM, n=3, * $P<0.05$, One way ANOVA, Dunnett multiple comparison between group was not significant. **L-N** Representative confocal fluorescence images of neurons across groups co-immunostained for MAP2 (green) and TUJ1 (red) after 50 days of differentiation. Nuclei were counterstained by DAPI (blue). Scale bar: 50µm.