PP31. CRI DU CHAT INDUCED PLURIPOTENT STEM CELLS: NEW FRONTIERS IN DISEASE UNDERSTANDING

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Abstract: Cri du chat syndrome is a genetic disease caused by partial or total loss of the short arm of chromosome 5. A group of genes (1.6 %) was classified as dosage sensitive leading to haploinsufficiency. TERT. (Semaphorin) 5A and CTNND2 genes are known to be expressed in the brain, in neurons, in the developing nervous system and play a role in neural migration, but the function of the most genes on 5 p are still unknown. From two CdC patients' peripheral mononuclear blood cells, we generated induced pluripotent stem cells (IPSCs); positive clones for TRA 1-60 were expanded. We investigated the Stemness expression and pluripotent potential through spontaneous differentiation capacity in the three germ layers genes expression using q-PCR. A Taqman real-time q-PCR showed that expression of TERT, CTNND2 and (Semaphorin) 5A are about half, fifth and quarter respectively compared to control donor. Hereafter, these clones are valuable to recapitulate in neurons and into Mesenchymal Stromal Cells for diagnostic, prognostic and therapeutic targets detection.