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Lab resource: Stem Cell Line

Generation of an iPSC line of a patient with Angelman syndrome due to an imprinting defect

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ABSTRACT

Angelman syndrome (AS) is a neurodevelopmental disorder with leading symptoms of happy demeanor, intellectual disability, ataxia and seizures. AS can be caused by genetic and epigenetic aberrations, resulting in the absence of functional UBE3A protein in the brain. *UBE3A* is an imprinted gene, which is, in neurons of the brain, expressed exclusively from maternal chromosome 15. The generated iPSC line was derived from skin fibroblasts of a patient with AS, who, due to an imprinting defect, lacked DNA methylation at the chromosome 15 imprinting center, which controls maternal-specific expression of *UBE3A*.

Resource table

Unique stem cell line identifier	ZIPi015-K
Alternative name(s) of stem cell line	AS_ID, ZIP15
Institution	Zentrum für integrative Psychiatrie, University Hospital Kiel, Kiel, Germany
Contact information of distributor	Franz-Josef Müller, franz-josef.mueller@uksh.de Laura Steenpass, laura.steenpass@uk-essen.de
Type of cell line	iPSC
Origin	human
Additional origin info	Age: 12 Sex: female Ethnicity if known: caucasian
Cell Source	skin fibroblasts
Clonality	clonal
Method of reprogramming	episomal/transgene-free
Genetic Modification	epigenetic aberration – imprinting defect
Type of Modification	lack of DNA methylation establishment or maintenance in the germ line of the patient's mother
Associated disease	Angelman syndrome (OMIM #105830)
Gene/locus	Prader-Willi/Angelman syndrome locus, chromosome 15q11q13
Method of modification	NA
Name of transgene or resistance	NA
Inducible/constitutive system	NA
Date archived/stock date	14.04.2017 (Essen)
Cell line repository/bank	NA
Ethical approval	Ethikkommission der medizinischen Fakultät der Christian-Albrechts Universität zu Kiel, Approval number A145/11 A145/11

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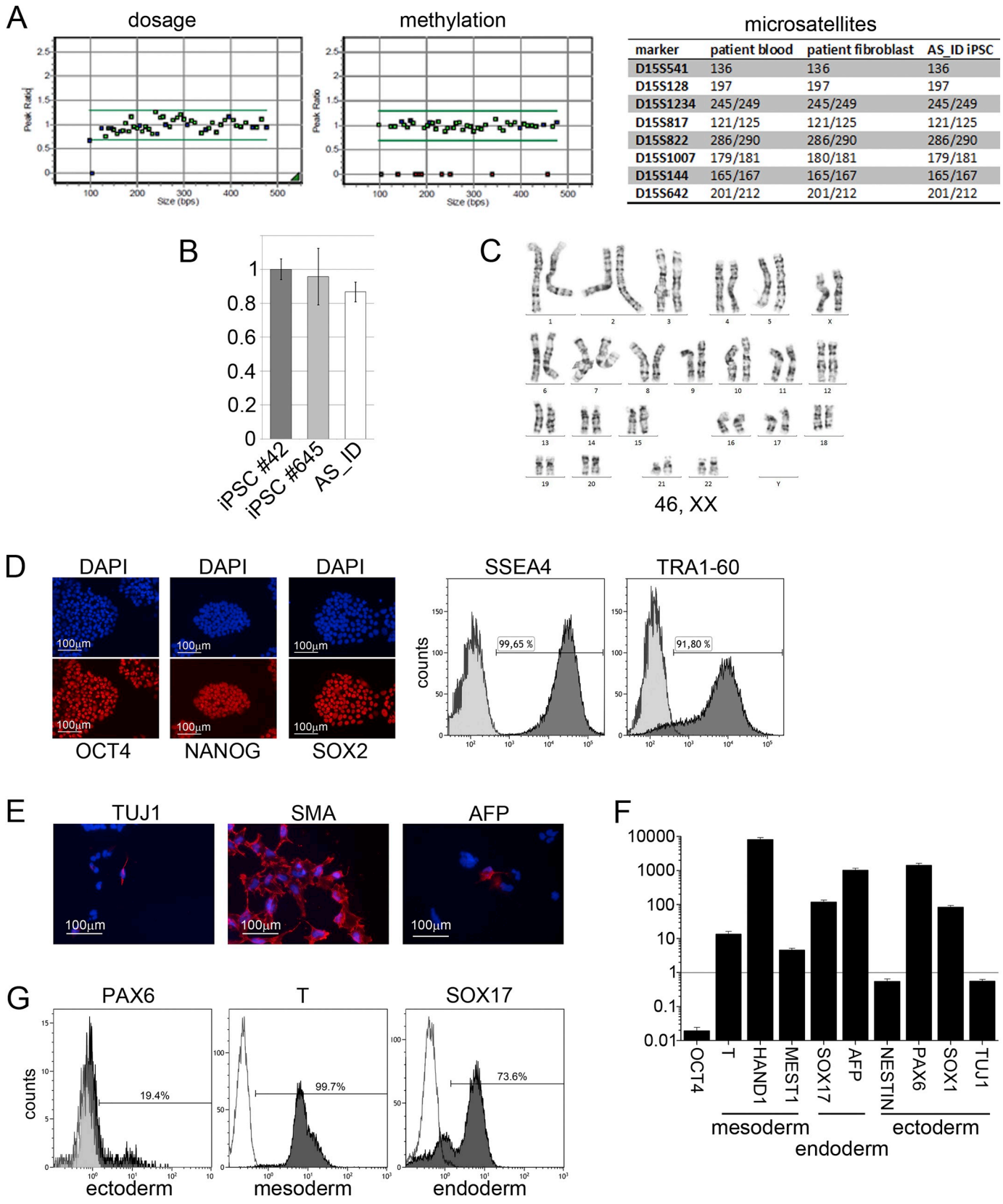

marker	patient blood	patient fibroblast	AS_ID iPSC
D15S541	136	136	136
D15S128	197	197	197
D15S1234	245/249	245/249	245/249
D15S817	121/125	121/125	121/125
D15S822	286/290	286/290	286/290
D15S1007	179/181	180/181	179/181
D15S144	165/167	165/167	165/167
D15S642	201/212	201/212	201/212

Fig. 1. Characterisation of patient-derived AS_ID iPSCs (ZIPi015_K) iPSCs carrying an imprinting defet.

1. Resource utility

The unavailability of neuronal tissue of patients hampers AS research. iPSC lines reprogrammed from patient cells and differentiated into neurons in vitro are an alternative. AS can arise from different genetic and epigenetic causes and it is of interest to establish iPSC lines covering all possible causes, including imprinting defects.