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Hypomyelination with tooth and bone involvement: New insights

Brain MR imaging and *in vitro* models

Ferdy K Cayami

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VRIJE UNIVERSITEIT

Hypomyelination with tooth and bone involvement: New insights

Brain MR imaging and *in vitro* models

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List of Abbreviation

4H	hypomyelination, hypodontia, hypogonadotropic hypogonadism
ADDH	ataxia, delayed dentition and hypomyelination
AIFM1	apoptosis inducing factor mitochondria – 1
ALP	alkaline phosphatase
ARS	alizarin red S
BCR	bicaudate ratio
CNS	central nervous system
CT	computer tomography
DMP1	dentin matrix acidic phosphoprotein 1
DNA	deoxyribonucleic acid
FLAIR	fluid-attenuated inversion recovery
FOP	fibrodysplasia ossificans progressiva
GMF	gross motor function
GMFCS	gross motor function classification system
H-ABC	hypomyelination with atrophy of basal ganglia and cerebellum
HBSL	hypomyelination with brainstem and spinal cord involvement and leg spasticity
HCC	hypomyelination with congenital cataracts
HEMS	Hypomyelination of early myelinated structure
H-SMD	hypomyelination with spondylometaphyseal dysplasia
ICC	intraclass correlation coefficient
iPSCs	induced pluripotent stem cells
IRR	Inter rater reliability
MBP	myelin basic protein
MLD	metachromatic leukodystrophy
MLPA	multiple ligation-dependent probe amplification
MRI	magnetic resonance imaging
MSCs	mesenchymal stem cells

OC	osteocalcin
ODDD	oculodentodigital dysplasia
OPN	osteopontin
PCR	polymerase chain reaction
PCWH	peripheral demyelinating neuropathy, central dysmyelination, Waardenburg syndrome and Hirschsprung disease
PLIC	posterior limb of the internal capsule
PLP1	proteolipid protein 1
PMD	Pelizaeus-Merzbacher disease
PMLD	Pelizaeus-Merzbacher like disease
POLR1	RNA Polymerase I
POLR3	RNA Polymerase III
qPCR	quantitative polymerase chain reaction
RNA	ribonucleic acid
RUNX2	runt-related transcription factor 2
T1W	T1-weighted
T2W	T2-weighted
TE	echo time
TI	inversion time
TOP	terminal oligopyrimidine tract
TR	repetition time
TRPV4	transient receptor potential cation channel subfamily V member 4
UCBSCs	umbilical cord blood-derived stem cells
VLCFA	very long chain fatty acids
WES	whole exome sequencing
WM	white matter
X-ALD	x-linked adrenoleukodystrophy

