

## FoRiSIE Winter School in Clinical Endocrinology

7-11 gennaio 2019 - Roma

# Una nuova malattia rara endocrinologica: la lipodistrofia

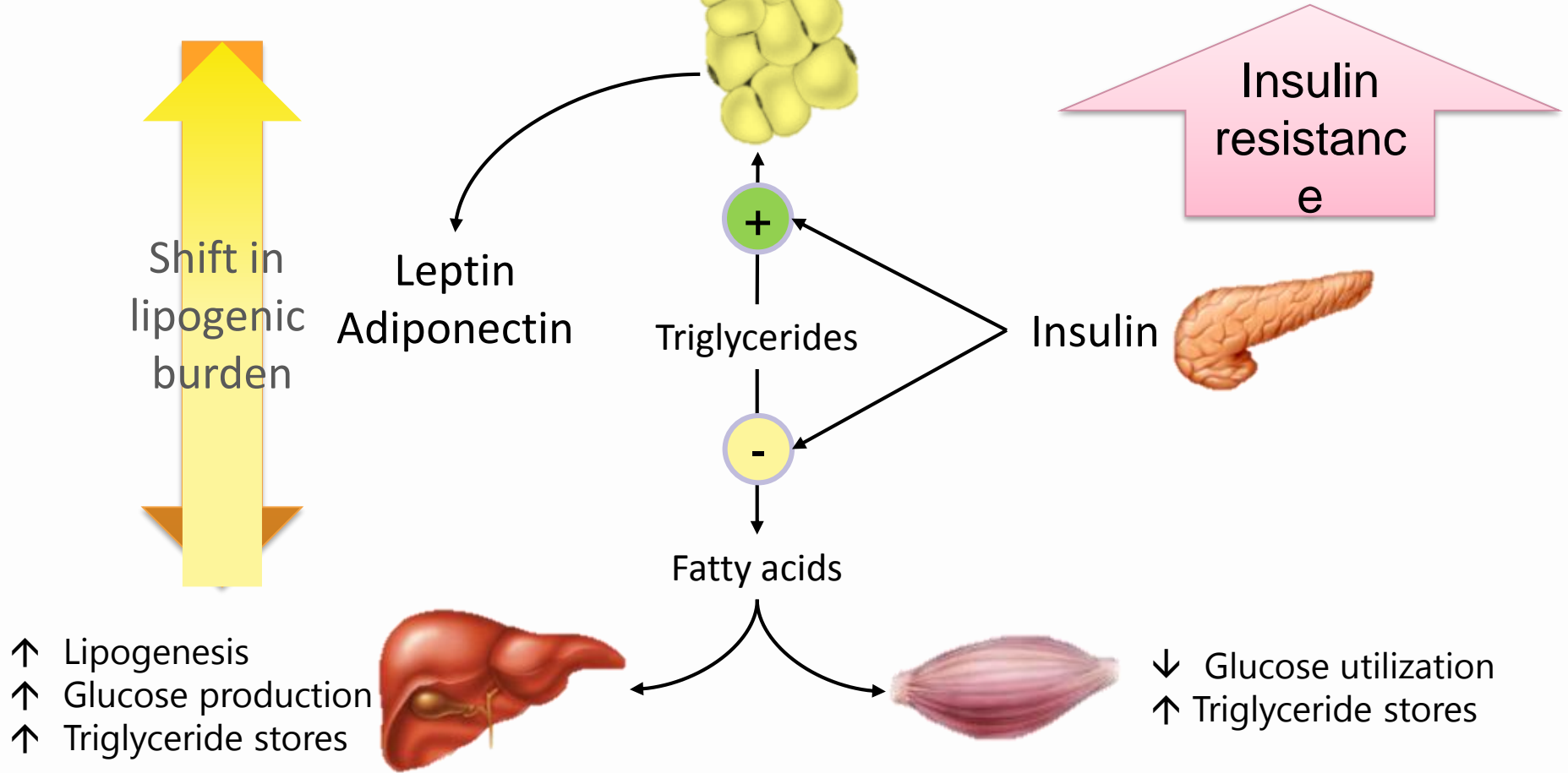
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Department of Emergency and Transplants

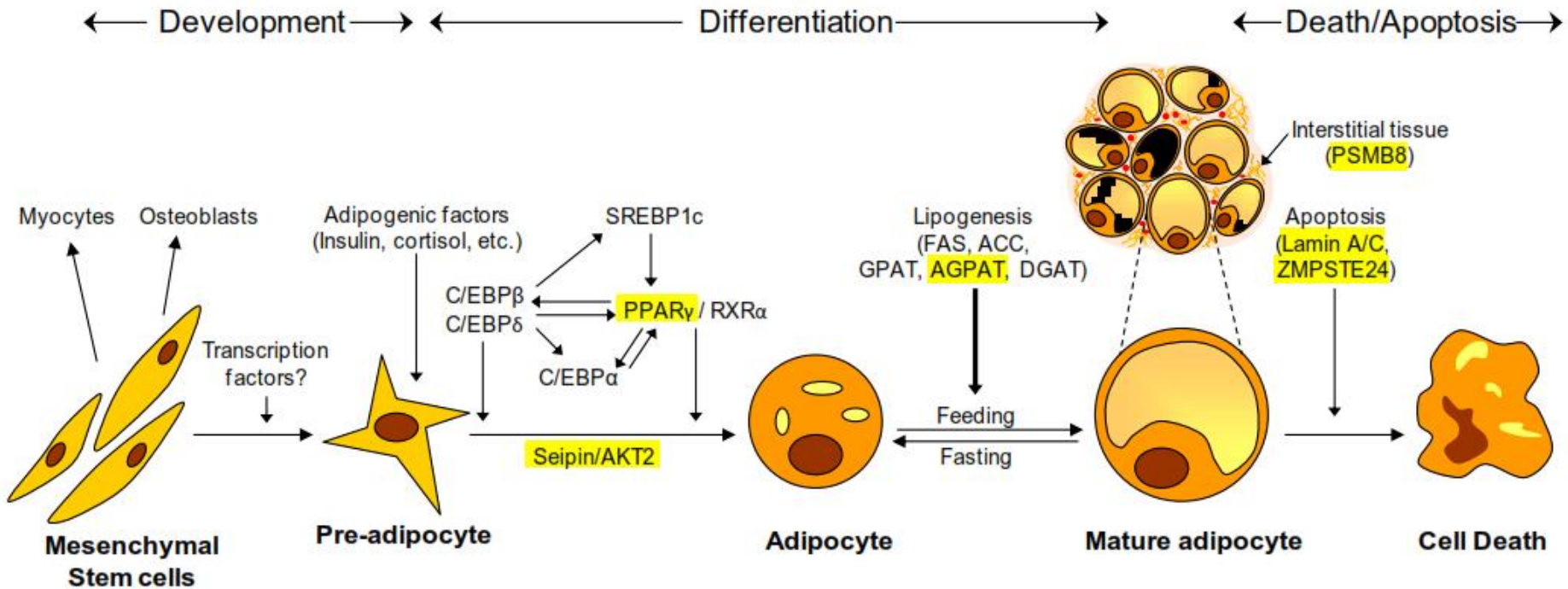
Section of Internal Medicine, Endocrinology, Andrology and Metabolic Diseases

University of Bari Aldo Moro, Bari, Italy

# The lipogenic burden ordinarily lies with adipose tissue

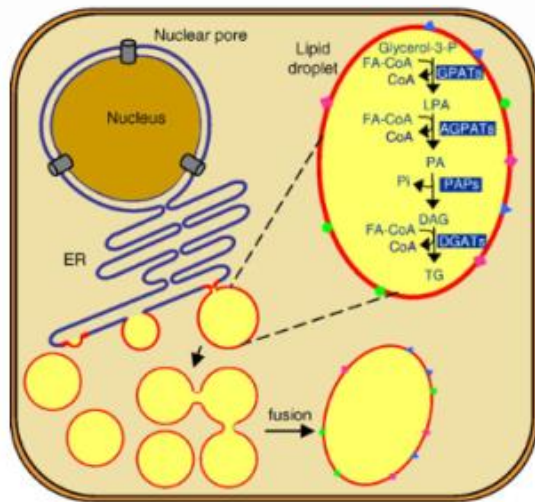


# Proteins mutated in human lipodystrophies

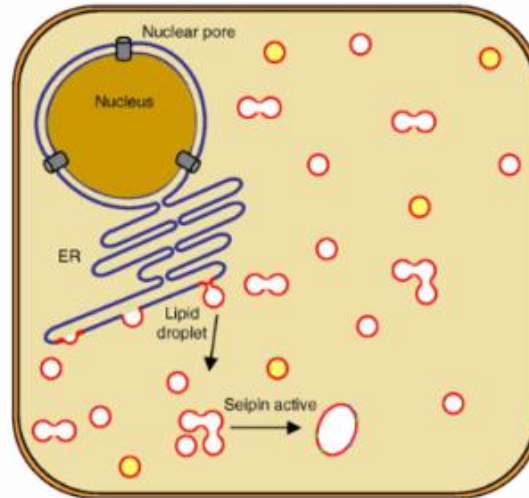


Garg A. *J Clin Endocrinol Metab* 96: 3313–3325, 2011

# Schematics of lipid droplet formation in adipocyte

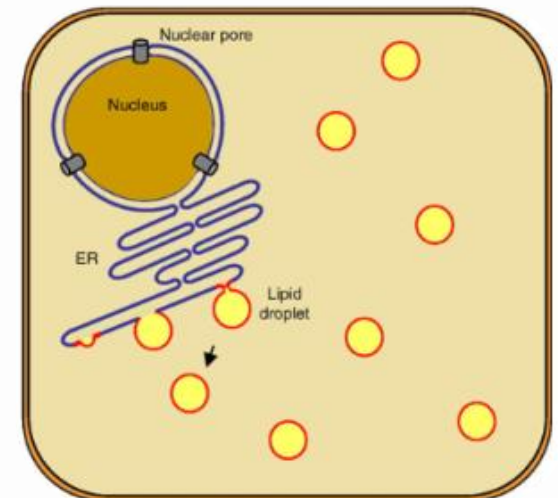


Normal



lipid synthesis or storage

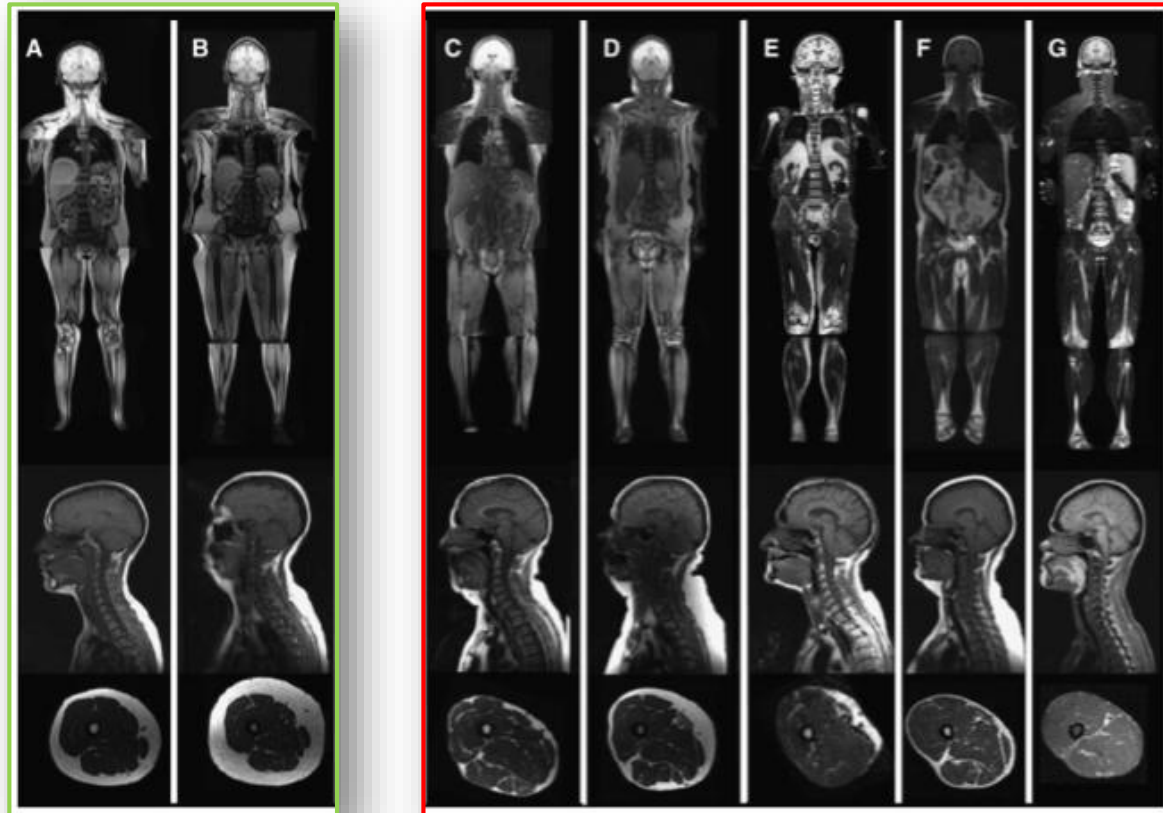
- lamins A/C through SREBP-1c
- AGPAT2
- caveolin 1
- cavin



lipid droplet

- Seipin
- Perilipin
- CIDEC

# Evaluation of adipose tissue deposition in females with various lipodystrophies



Hegele A, et al. *Journal of Lipid Research* Volume 48, 2007

# High prevalence of laminopathies among patients with metabolic syndrome

	Patients without nuclear abnormality, <i>n</i> = 77	Patients with nuclear abnormalities, <i>n</i> = 10	<i>P</i> -value
Age (years)	56.6 ± 1.0	50.2 ± 3.3	<b>0.03</b>
BMI (kg/m <sup>2</sup> )	36.6 ± 0.8	39.1 ± 1.2	0.28
WC (cm)	118.8 ± 1.5	125 ± 2	0.16
TC (cm)	54.8 ± 0.7	59 ± 1.3	<b>0.04</b>
Fat mass (%)	38.8 ± 0.7	40 ± 3	0.3
WC/TC	2.17 ± 0.02	2.13 ± 0.06	0.3
TC/BMI	1.53 ± 0.02	1.51 ± 0.02	0.7
Frequency of diabetes ( <i>n</i> , %)	62, 81	9, 90	0.49
Frequency of hypertension ( <i>n</i> , %)	59, 77	9, 90	0.45
Frequency of statin TRT ( <i>n</i> , %)	48, 62	6, 60	0.9
Number of criteria of MS	4.09 ± 0.09	4.4 ± 0.27	0.25
HDL cholesterol (mmol/l)	1.12 ± 0.04	0.97 ± 0.13	0.3
Triglycerides (mmol/l)	2.14 ± 0.16	3.39 ± 0.89	<b>0.03</b>
Alanine aminotransferase (UI/l)	43.1 ± 2.8	66 ± 20	<b>0.03</b>
Gamma glutamyl transpeptidase (UI/l)	63.9 ± 8.5	100 ± 43	0.2
Slight lipodystrophy (of lower limb) ( <i>n</i> , %)	(2) (2.6)	2 (20)	<b>0.006</b>
Muscular complaint ( <i>n</i> , %)	(2) (2.6)	2 (20)	<b>0.006</b>
Cardiac rhythm abnormalities ( <i>n</i> , %) <sup>b</sup>	(2) (2.6)	1 (10)	0.31

