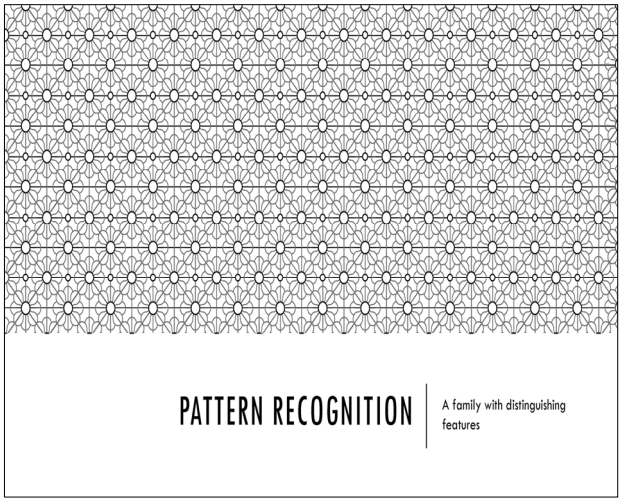
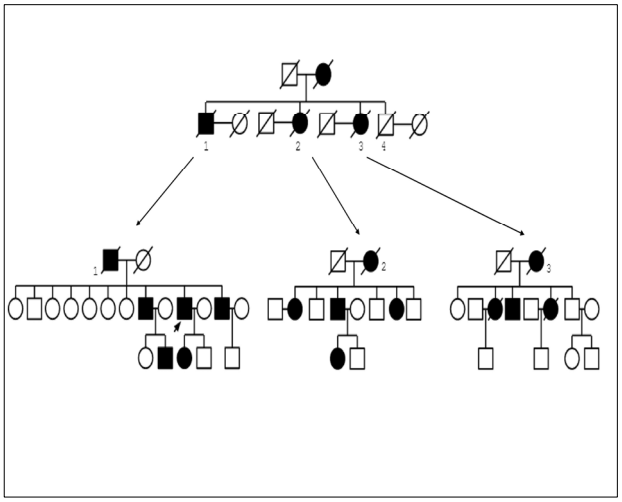
유전성 근육질환

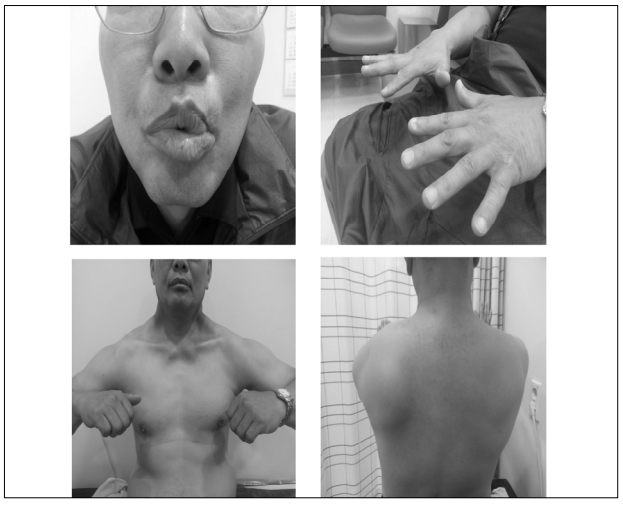
신 진 홍
양산부산대학교병원

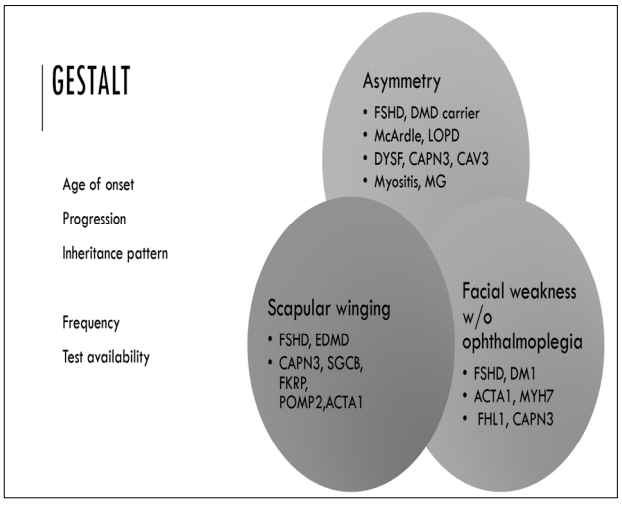
Genetic Muscle Disease

Jin-Hong Shin
Pusan National University Yangsan Hospital









MYOPATHY PLUS

A newborn with high CK, weakness and brain malformation

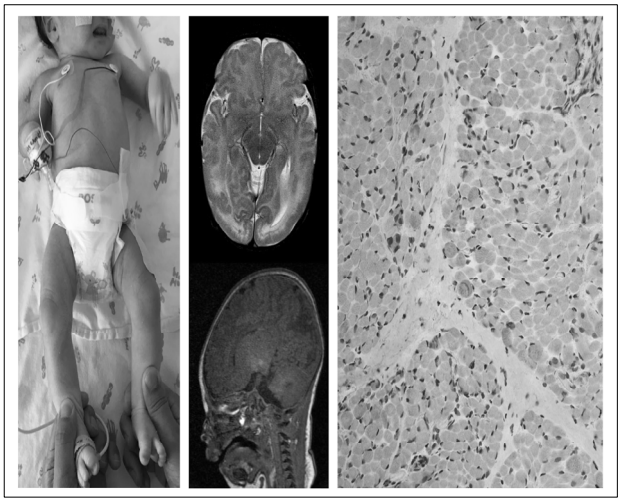


TABLE 12.4 **Genetically Identified Forms of Congenital Muscular Dystrophy (CMD)**

Common Disease Name	Abbreviation	Gene Symbol	Gene Locus	Protein	Protein Type
'Merosin deficient' CMD	MDC1A	*LAMA2*	6q	Laminin α2	Extracellular matrix
Ullrich CMD	UCMD	*COL6A1*	21q22	Collagen VI	Extracellular matrix
	UCMD	*COL6A2*	21q22	Collagen VI	Extracellular matrix
	UCMD	*COL6A3*	2q37	Collagen VI	Extracellular matrix
Integrin α7 deficiency		*ITGA7*	12q13	Integrin α7	Transmembrane (plasma membrane)
Fukuyama CMD	FCMD (MDDGA4)*	*FKTN***	9q31-q33	Fukutin	Possible substrate for glycosyltransferase
Muscle-eye-brain disease	MEB (MDDGA3)	*POMGNTI*	1p3	*O*-mannose β-1,2-*N*-acetyl-glucosaminyl transferase	Glycosyltransferase
Walker–Warburg syndrome	WWS				
	(MDDGA1) (MDDGA2) (MDDGA7)	*POMT1* *POMT2* *ISPD*	9q34 14q24.3 7p21	Protein-*O*-mannose transferases 1 & 2 Isoprenoid synthase containing domain	Glycosyltransferase ?
–	MDC1B	?	1q42	?	?
–	MDC1C (MDDGA5)	*FKRP*	19q1	Fukutin-related protein	Possible phosphosugar transferase
–	MDC1D (MDDGA6)	*LARGE*	22q12	LARGE	Possible glycosyltransferase
Rigid spine syndrome	RSMD1	*SEPN1*	1q36	Selenoprotein N1	Glycoprotein of endoplasmic reticulum

TREATMENT AVAILABILITY

Man with progressive proximal weakness and respiratory failure

M41 PROGRESSIVE ORTHOPNEA FOR 1 MO

- weight loss >10 kg for 2 years
- respiratory weakness and difficulty climbing stairs for 2 months
- proximal weakness 4 to 4+
- EMG myotonia (+), clinical myotonia (-)
- FVC 1.39 L
- An elder sister with recurrent respiratory failure, unknown cause

TREATMENT AVAILABLE BY ANY MEANS

Inflammatory myopathy

Genetic myopathy
- Muscular dystrophy
- Metabolic myopathy

NMJ disorder

Motor neuron disorder