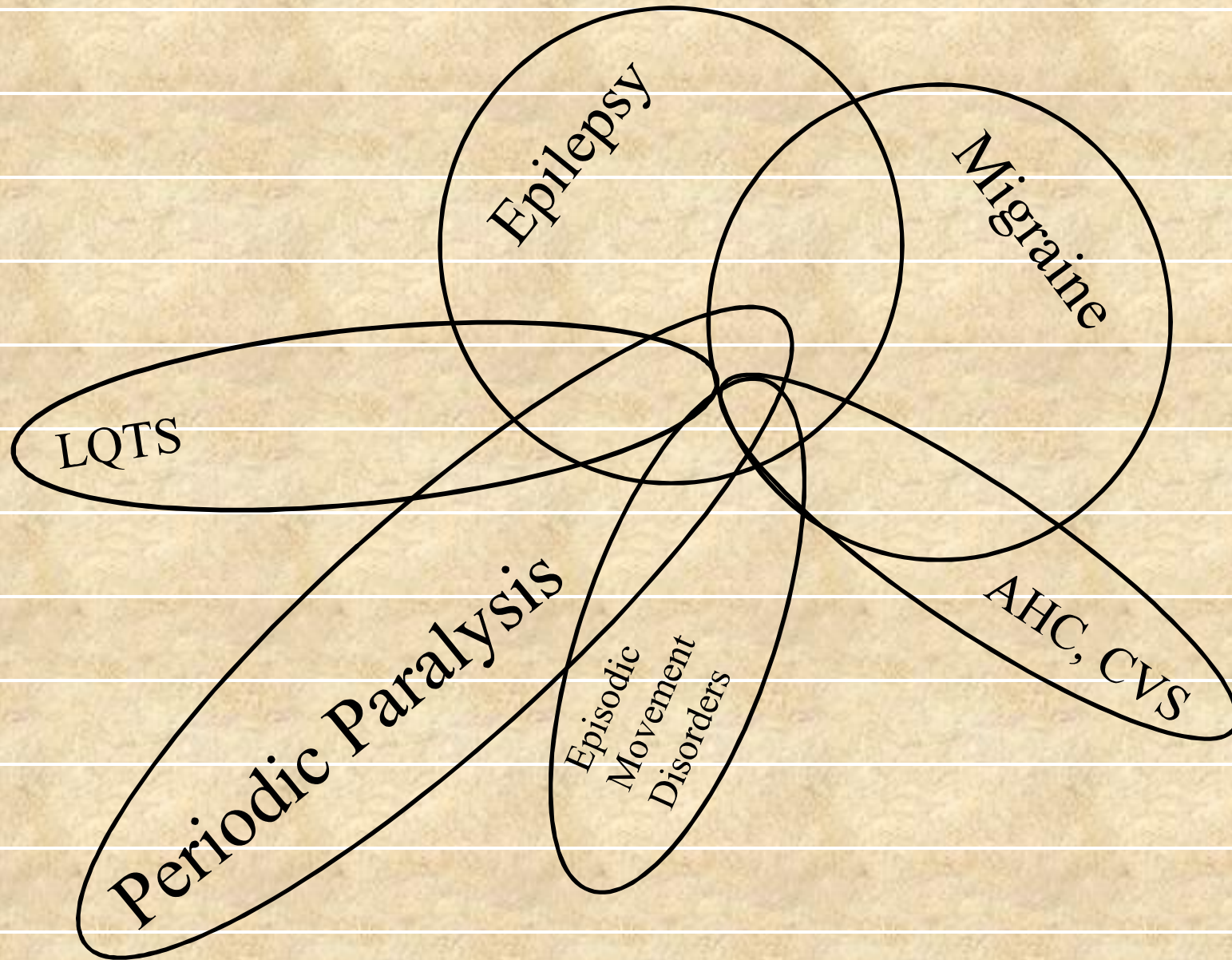
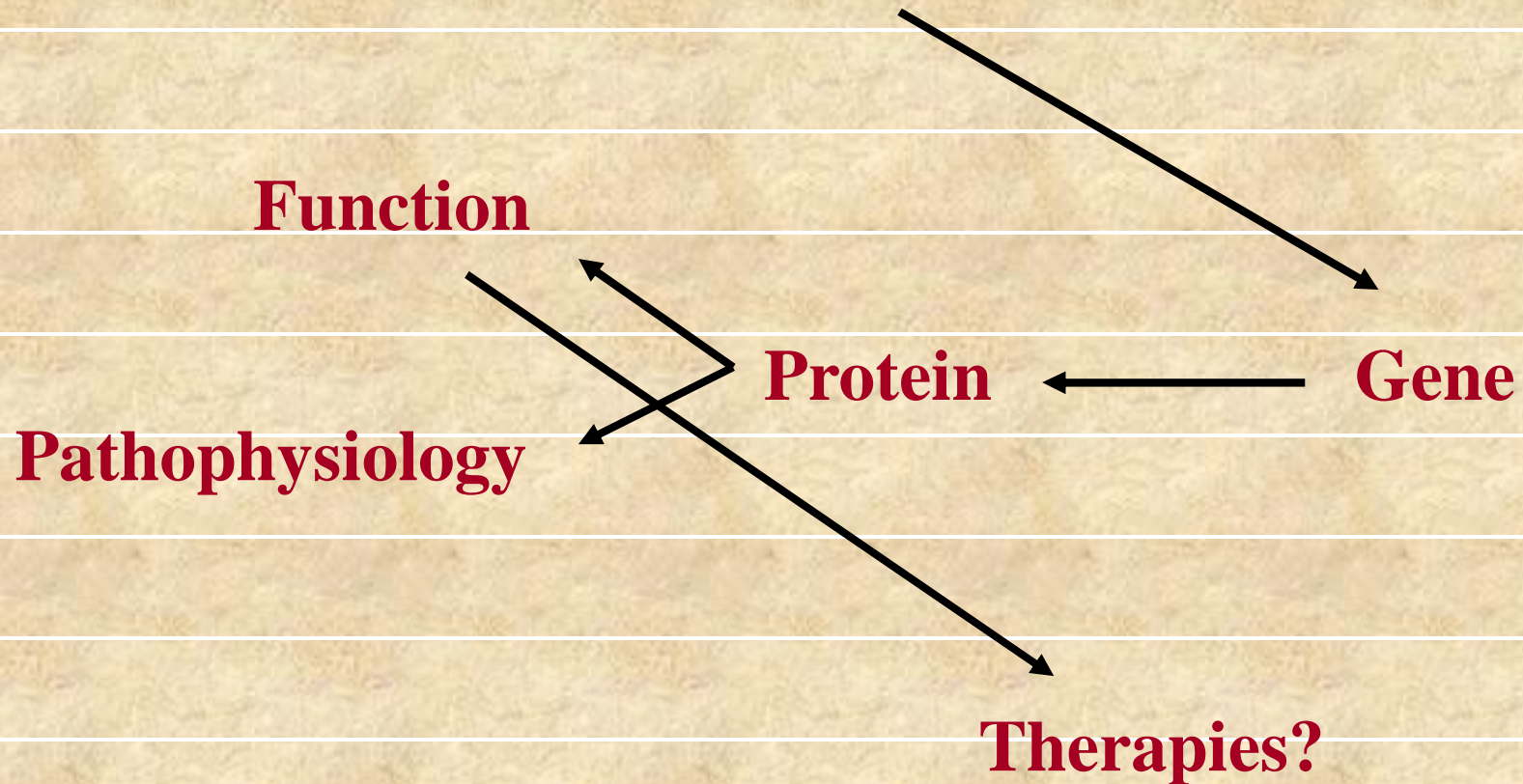


# Episodic Disorders



# Rare Monogenic Phenotype (FASPS)



# Non-dystrophic Myotonia/Periodic Paralysis

Hyperkalemic Periodic Paralysis → *SCN4A*

Hypokalemic Periodic Paralysis → *SCN4A*  
→ *CACNA1S*

Paramyotonia Congenita → *SCN4A*

Potassium Aggravated Myotonia → *SCN4A*

AR Myotonia Congenita → *CLCN1*

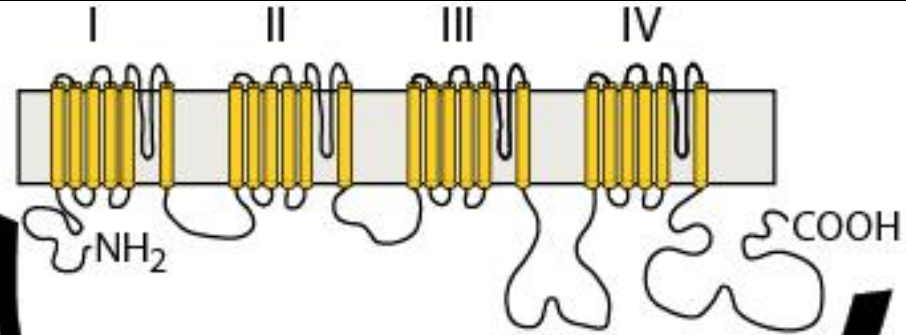
AD Myotonia Congenita → *CLCN1*

Andersen-Tawil Syndrome → *KCNJ2*

Thyrotoxic Hypokalemic PP → *KCNJ18*

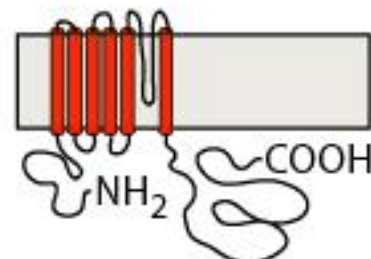
# NaCh

*Periodic Paralysis  
Epilepsy, LQT*

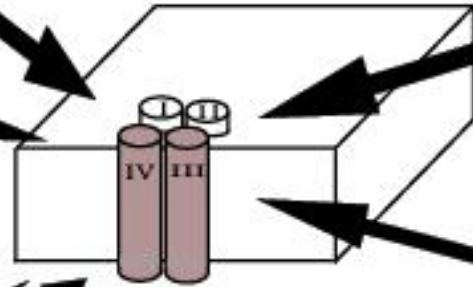


# KCh

*Periodic Paralysis  
Episodic Ataxia  
Epilepsy, LQT*

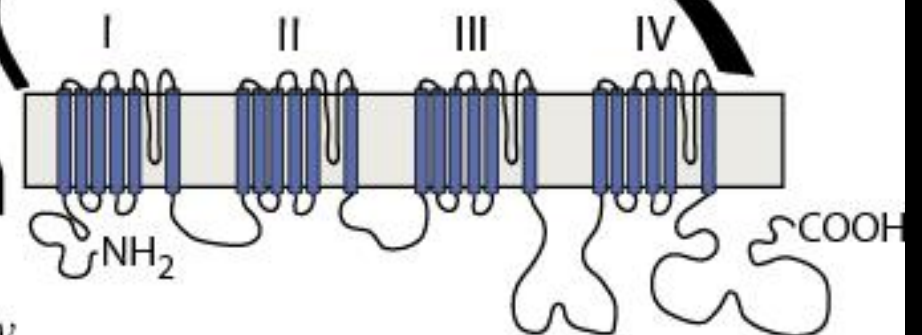


X 4



# CaCh

*Periodic Paralysis,  
Episodic Ataxia, Epilepsy  
Familial Hemiplegic Migraine*



# Phenotypes

**Muscle diseases—periodic paralyses**

**NMJ—Congenital myasthenic, Lambert-Eaton**

**Peripheral nerve—pain syndromes**

**Central nervous system—cortex**

**Migraine**

**Epilepsy**

**Familial Cortical Myoclonus**

**Cerebellum—Episodic ataxia**

# TC 25 Channelopathies

Monday, September 23 14:45 - 18:15

Chairs: Louis Ptacek, Angela Vincent

Autoantibodies to ion channels, receptors and associated proteins  
Angela Vincent

Channels in Familial hemiplegic migraine and episodic ataxia  
Arn Van den Maagdenberg

Channels in epilepsy  
Ingrid Scheffer

Paroxysmal dyskinesias—disorders of the synapse  
Louis Ptacek

**Autoantibodies**



**Ion**



**Transcriptional  
Regulation**

**Channels**



**Synaptopathies**



**Post-translational  
Modifications**

***Stress!***  
***Redox?***

**Trafficking and  
Cellular Localization**

**Membrane  
Excitability**