

## CONSULTATION DIAGNOSTIQUE

### INTERROGATOIRE ET EXAMEN CLINIQUE

- Histoire de la maladie (premiers symptômes, évolution, antécédents familiaux...)
- Arbre généalogique (mode de transmission)
- Examen neuromusculaire
- Tests analytiques et fonctionnels
- Examen général

### PREMIÈRE ÉVOCACTION D'UNE PATHOLOGIE

#### EXAMENS COMPLÉMENTAIRES

- ANALYSES BIOLOGIQUES SELON LE CONTEXTE**
- CPK, aldolase... (dystrophies musculaires)
  - Anticorps anti-récepteurs (myasthénie)
  - Autres bilans biologiques

#### Scanner et IRM

#### EXAMENS CIBLÉS EN FONCTION DES PREMIERS ÉLÉMENTS CLINIQUES

#### Biopsie Musculaire

- Histologie
- Microscopie électronique
- Immunohistochimie

#### Electromyogramme (EMG)

- Maladie de Charcot-Marie-Tooth
- Myasthénie auto-immune
- Syndrome myasthénique congénital
- Syndrome myotonique
- Dystrophie myotonique de Steinert
- Dystrophie musculaire facio-scapulo-humérale
- Amyotrophie spinale
- ...

#### - Test d'effort - Bilans métaboliques

- Myopathies métaboliques
- ...

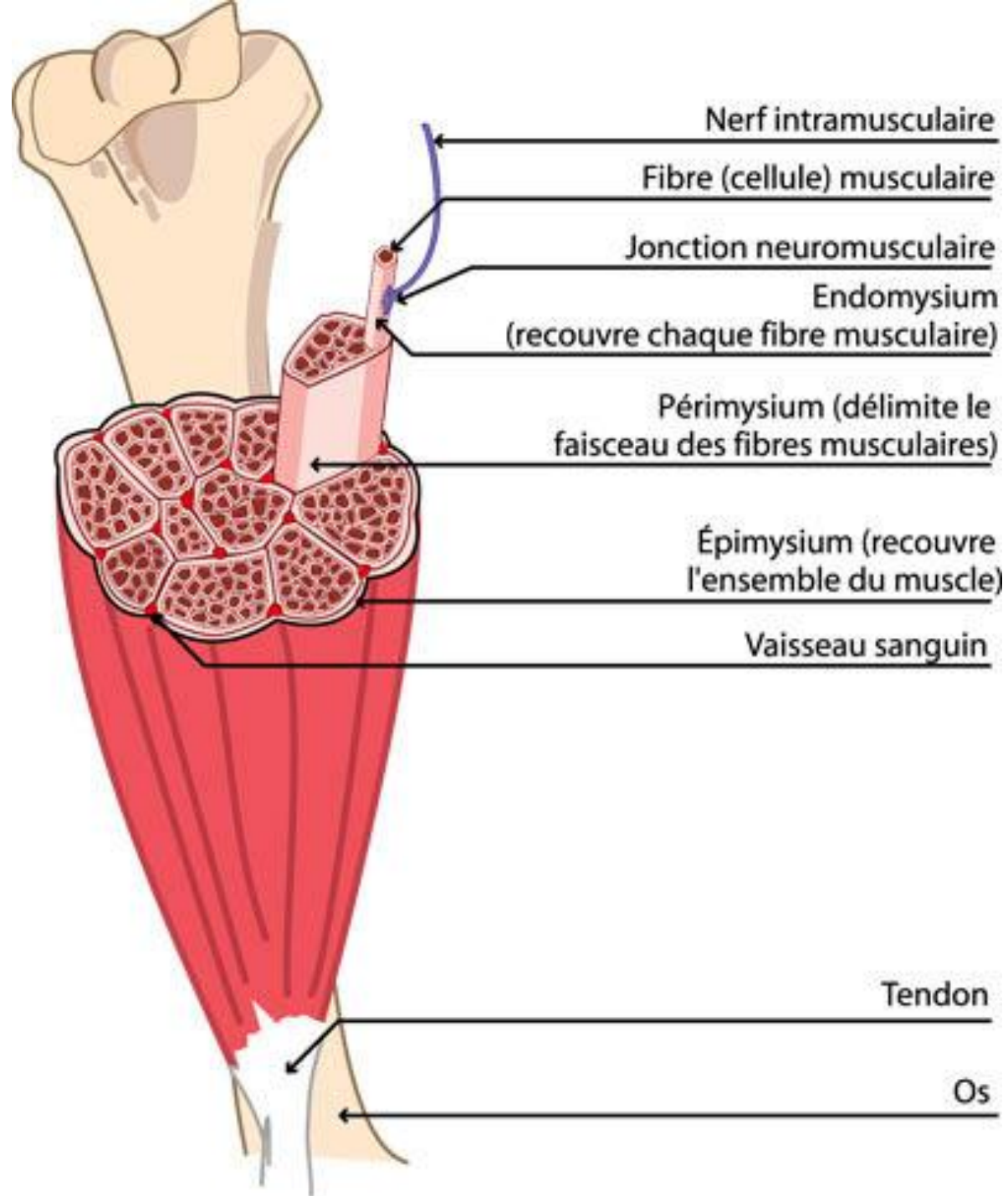
#### Bilans inflammatoire et immunologique

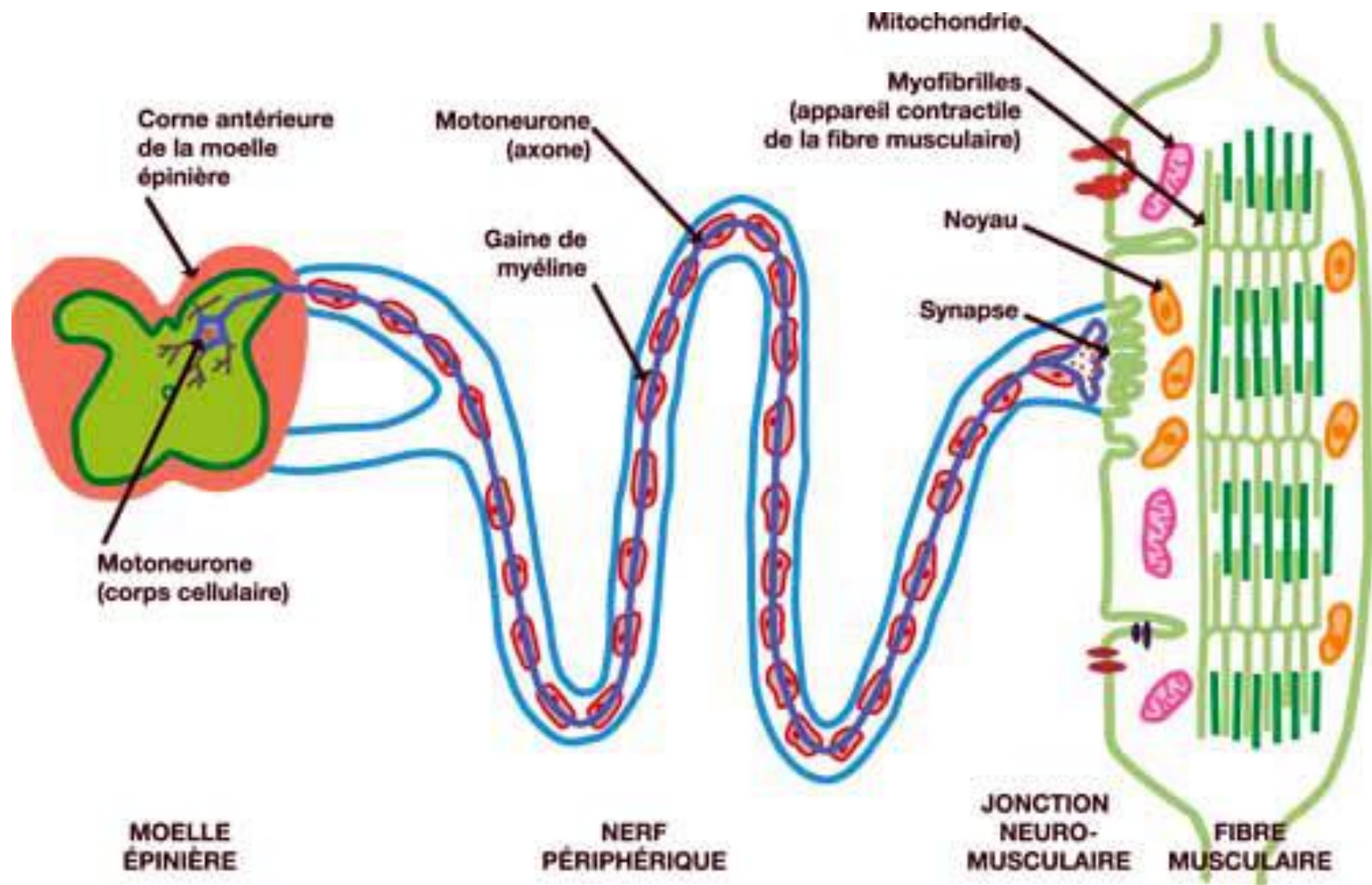
- Dermatomyosites,
- polymyosites
- ...

### DIAGNOSTIC CLINIQUE

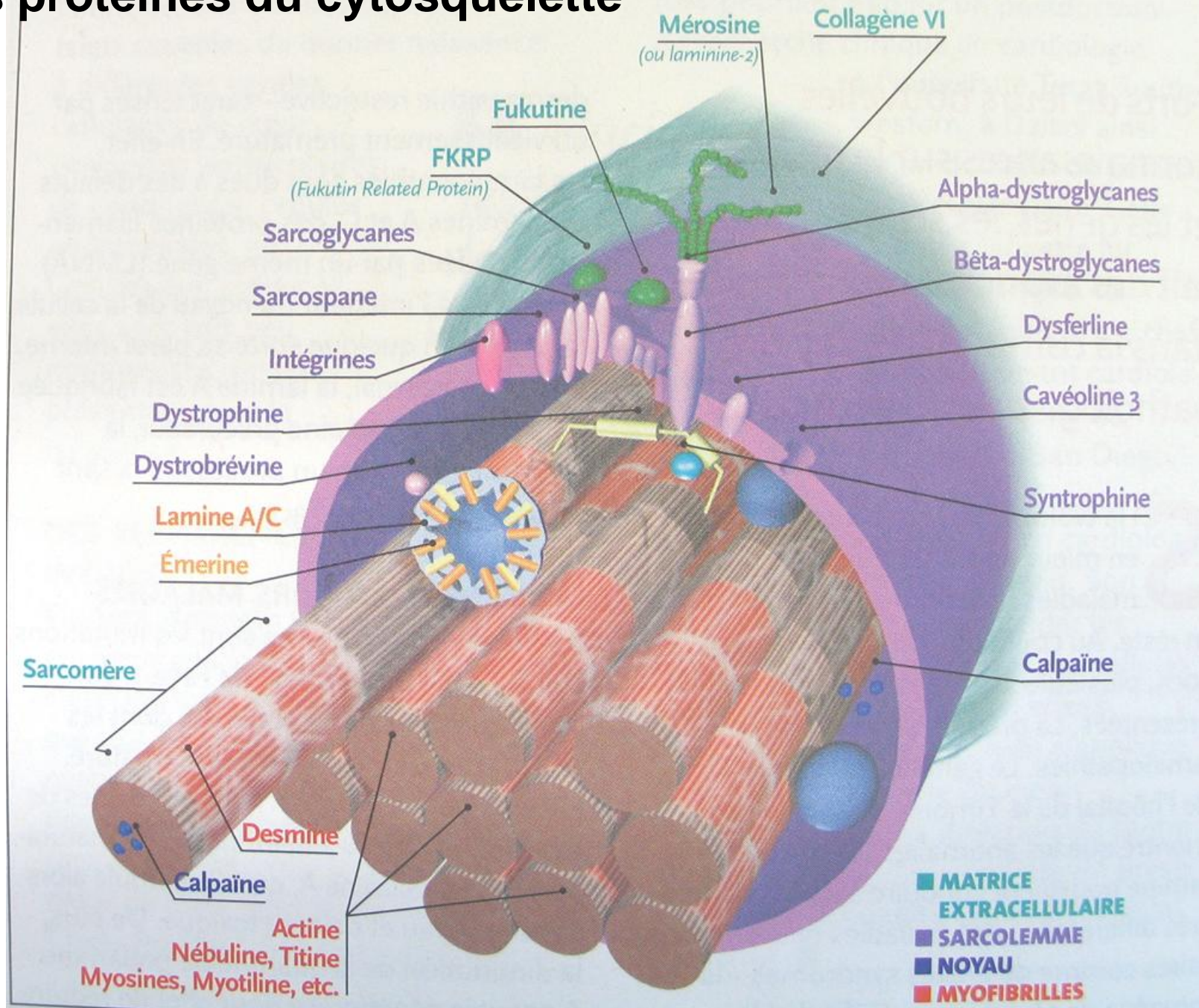
- Analyses génétiques**
- Recherche de l'anomalie génétique primaire (mutations, analyse du génome...)
  - Analyses de liaison

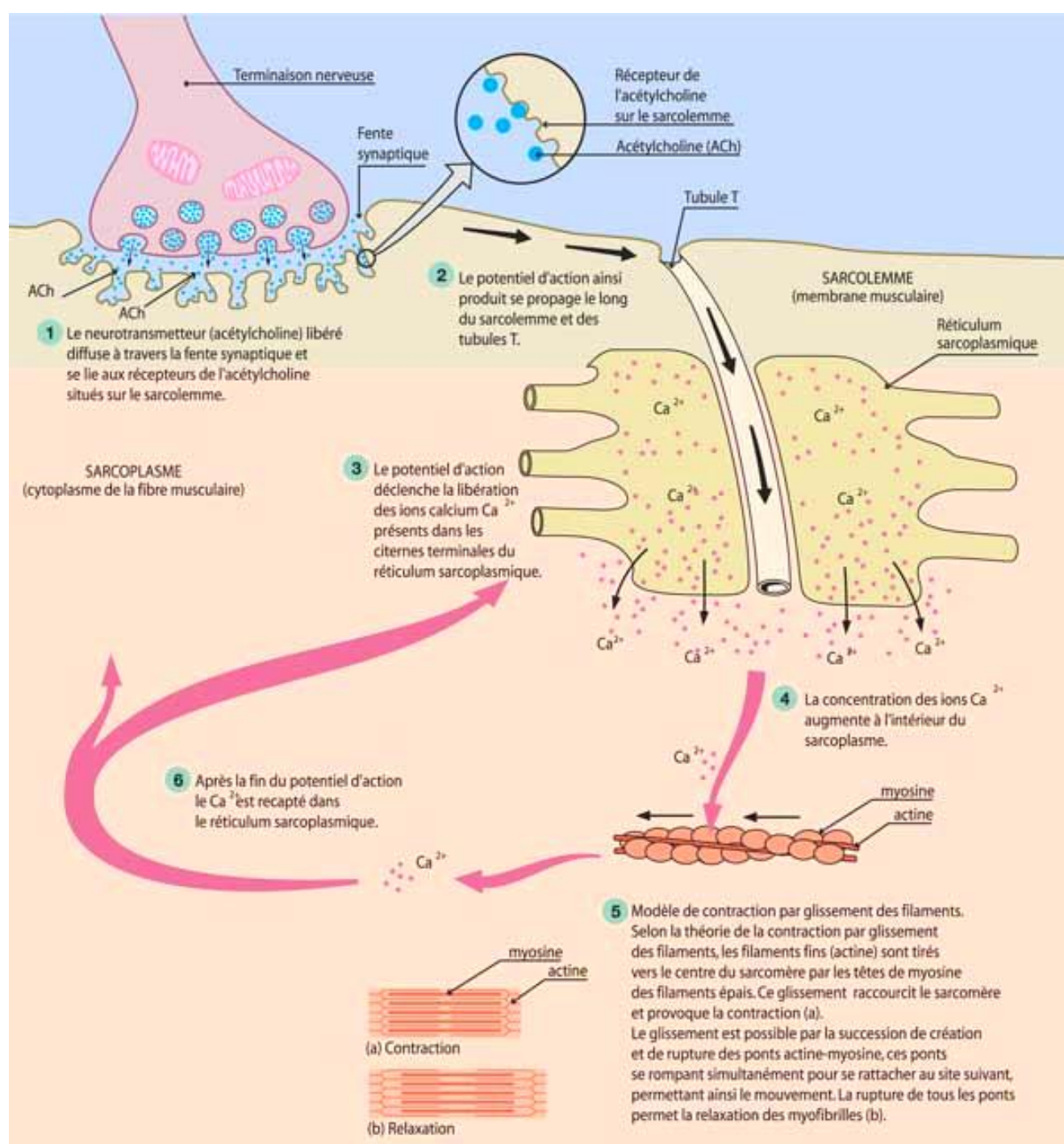
### DIAGNOSTIC GÉNÉTIQUE



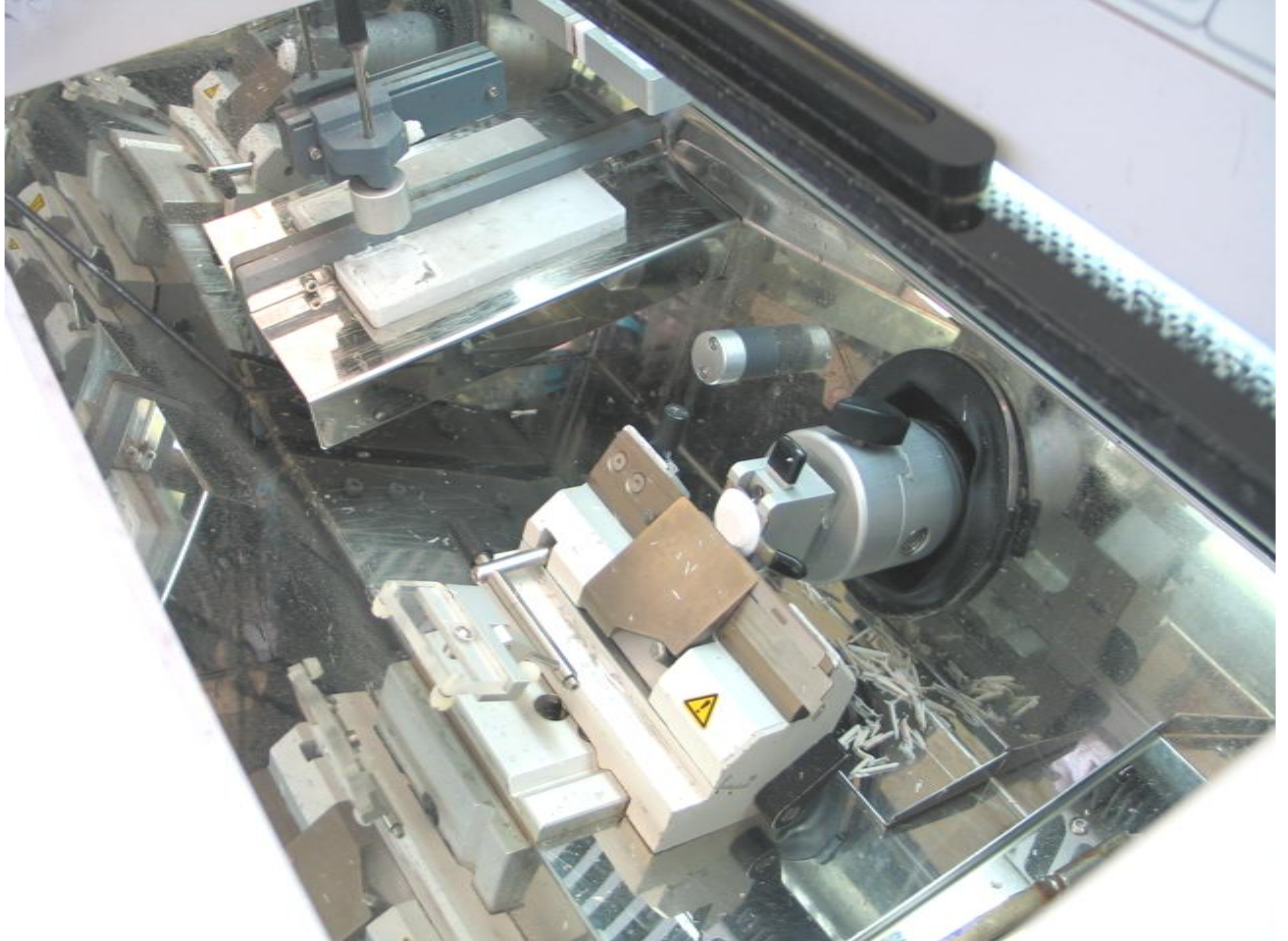


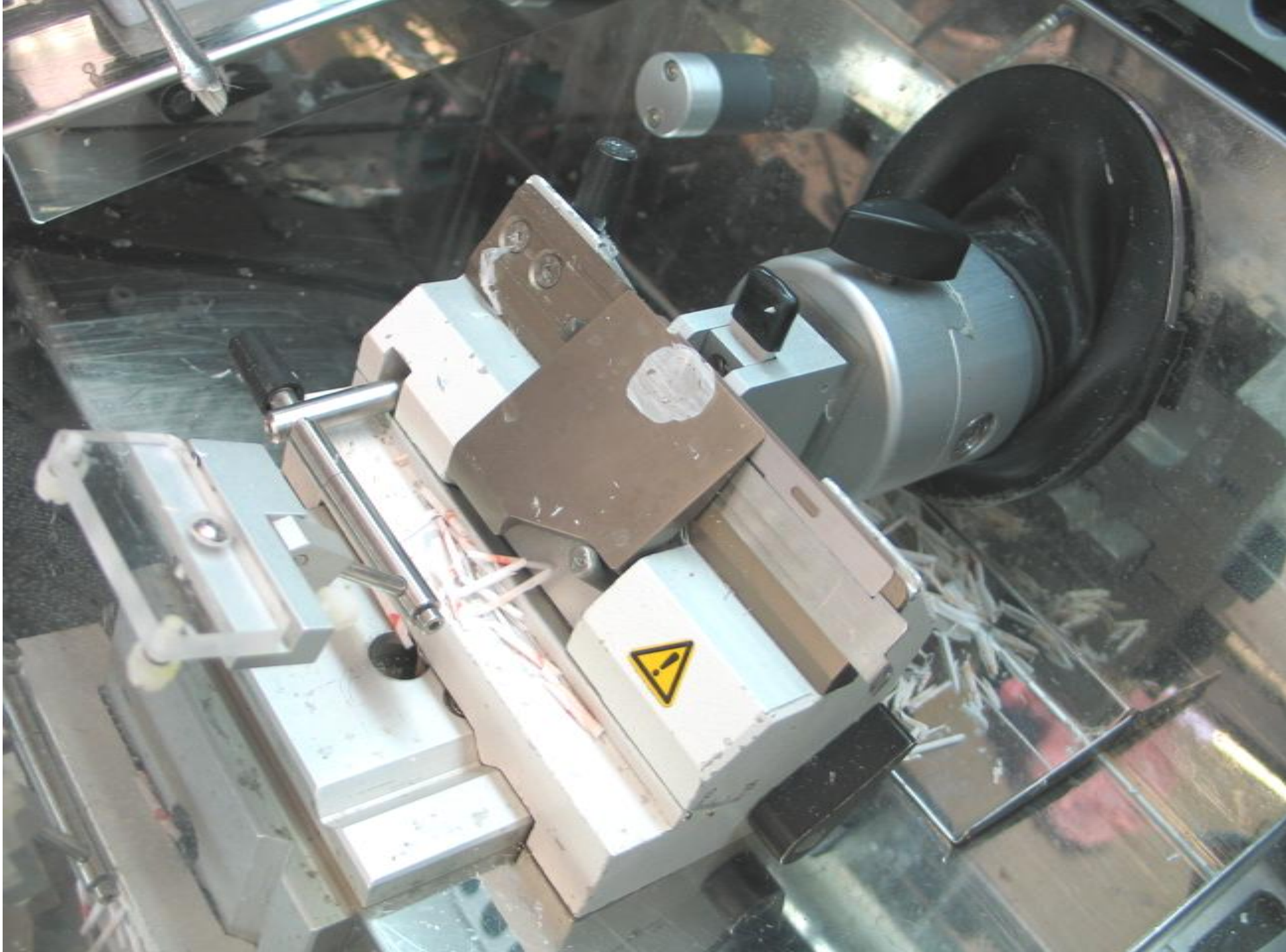
# Principales protéines du cytosquelette



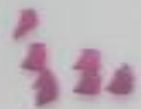








LH07  
478  
HE



LH07  
478  
Methionine



LH07  
478  
FAS



LH07  
478  
N.S



LH07  
478  
NADH



LH07  
478  
4.35



LH07  
478  
9.6



LH07  
478  
Cox/SDH



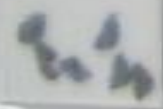
LH07  
478  
ANP



LH07  
478  
Puro Ac



LH 07  
478  
myophosph



+52 a-sarcogly  
LH07 478  
CB  
12/03/2007  
© [LOT] 40057



+38 Dsferline  
LH07 478  
CB  
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+51 caveolin  
LH07 478  
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+46 Or5T 1  
LH07 478  
CB  
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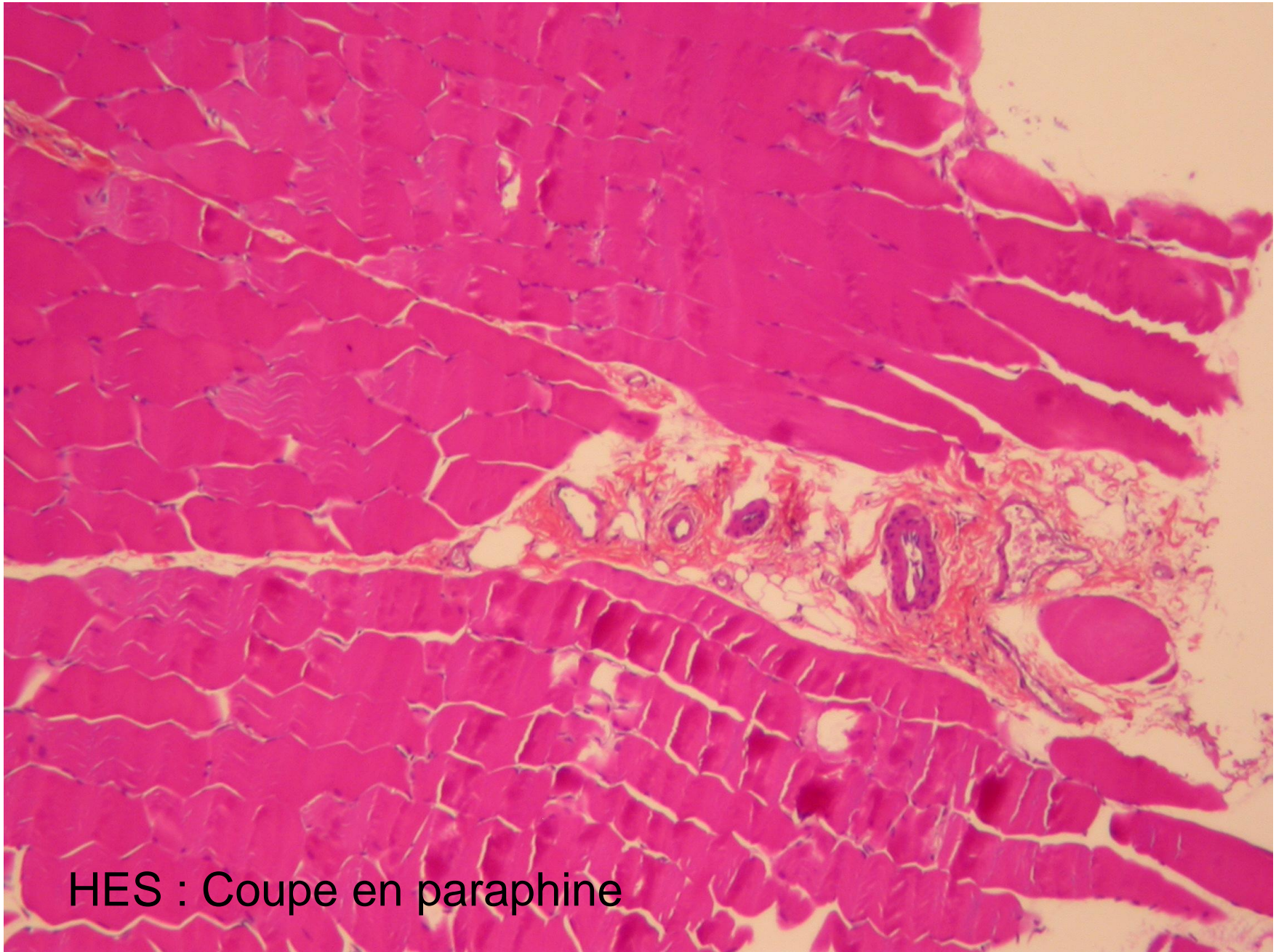


# Divers mécanismes d'atteinte musculaire

- par **lésion du motoneurone** qui innerve la cellule musculaire ou de la transmission neuromusculaire
- par **atteinte primitive** de la cellule musculaire:
  1. soit de son architecture par anomalie d'une des protéines de son cytosquelette : **dystrophie musculaire**
  2. soit de son métabolisme : **myopathie métabolique**
  3. soit par anomalie de son développement : **myopathie congénitale**
- **myopathies inflammatoires**

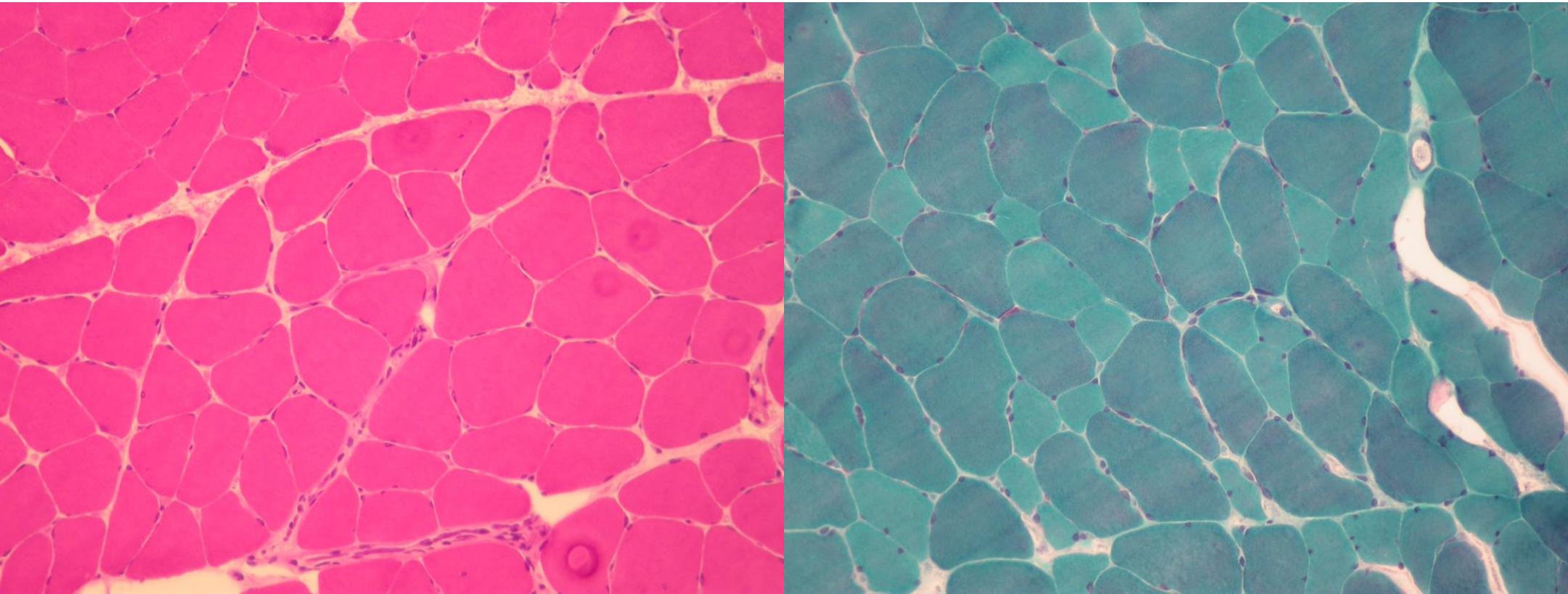
# HISTOCHIMIE

- Hématéine éosine : morphologie des myocytes ,état de l'endomysium...
- Trichrome de Gomori : agrégats tubulaires , amas mitochondriaux
- Noir Soudan : lipides
- PAS: glycogène



HES : Coupe en paraffine

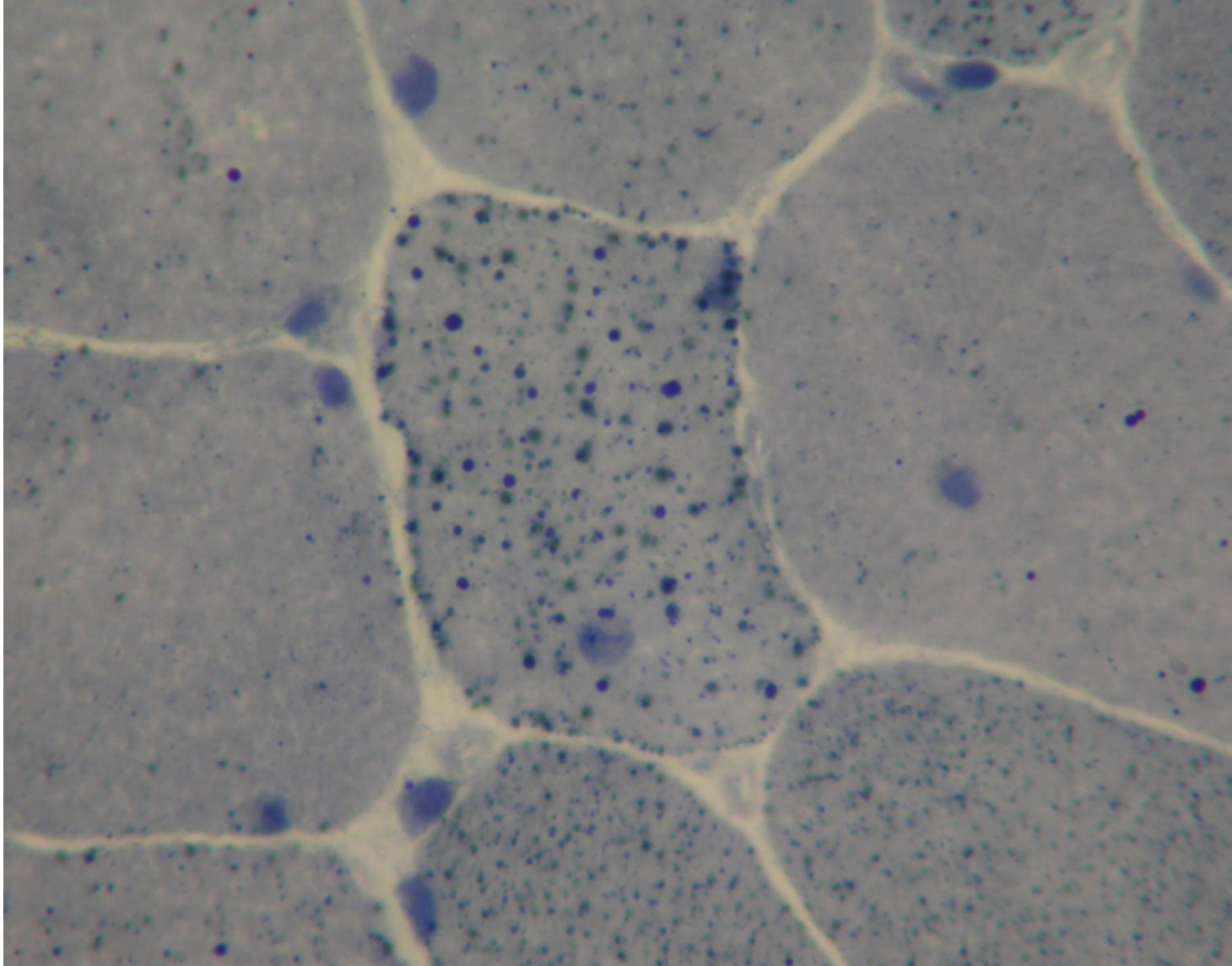
# MUSCLE NORMAL



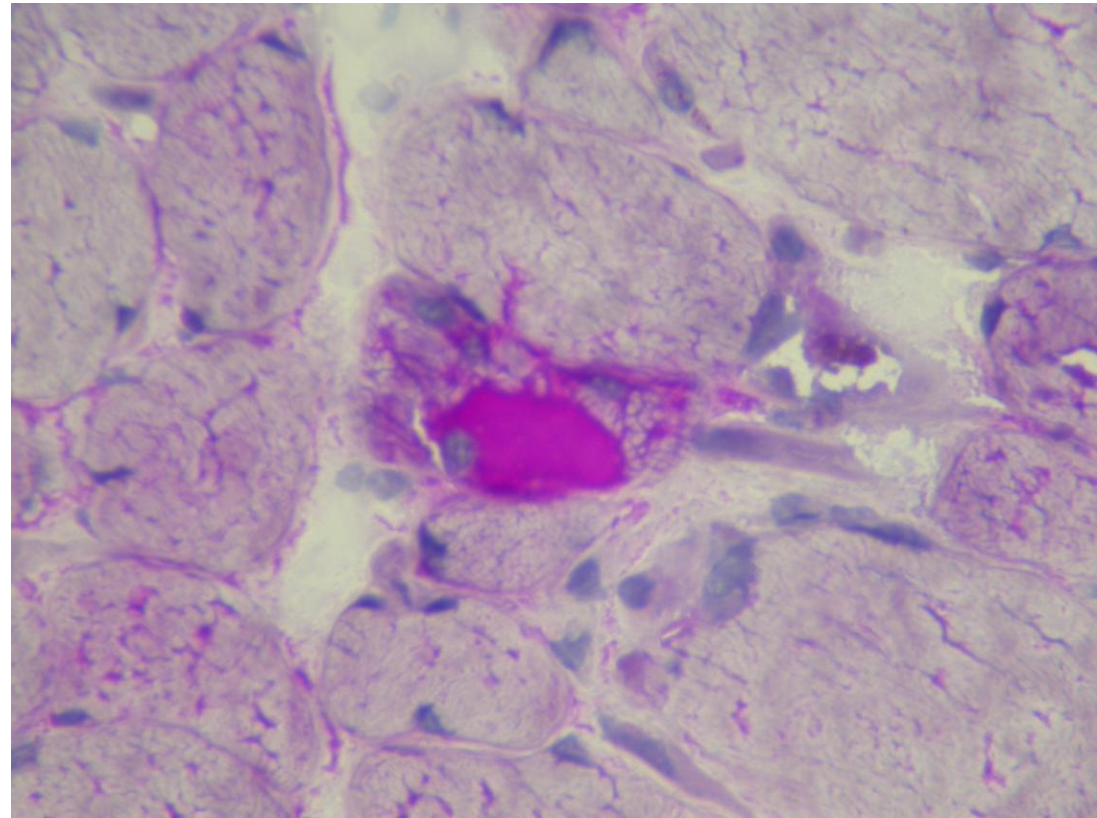
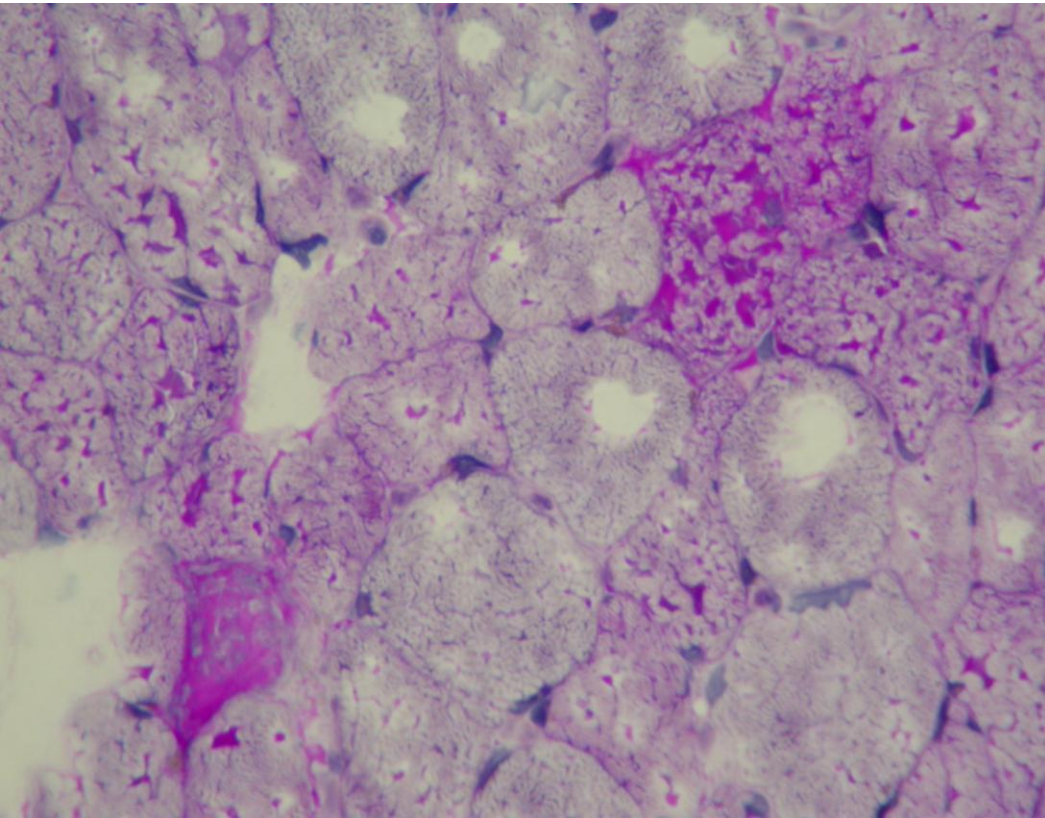
Hématéine éosine

Trichrome de Gomori

# Noir soudan : lipides



# PAS : GLYCOGENE



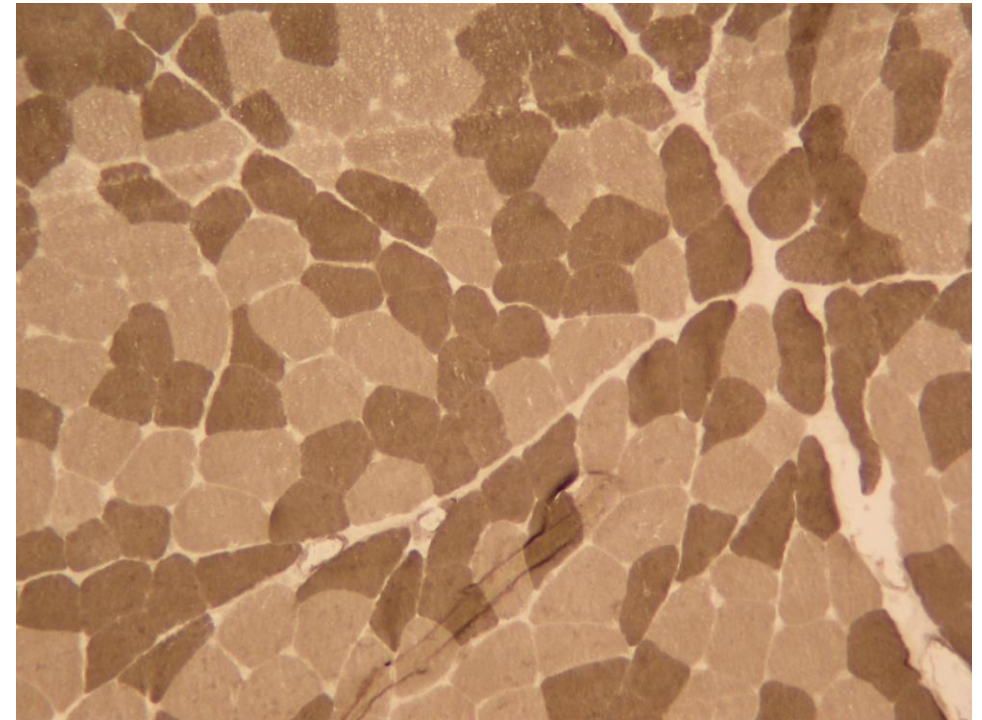
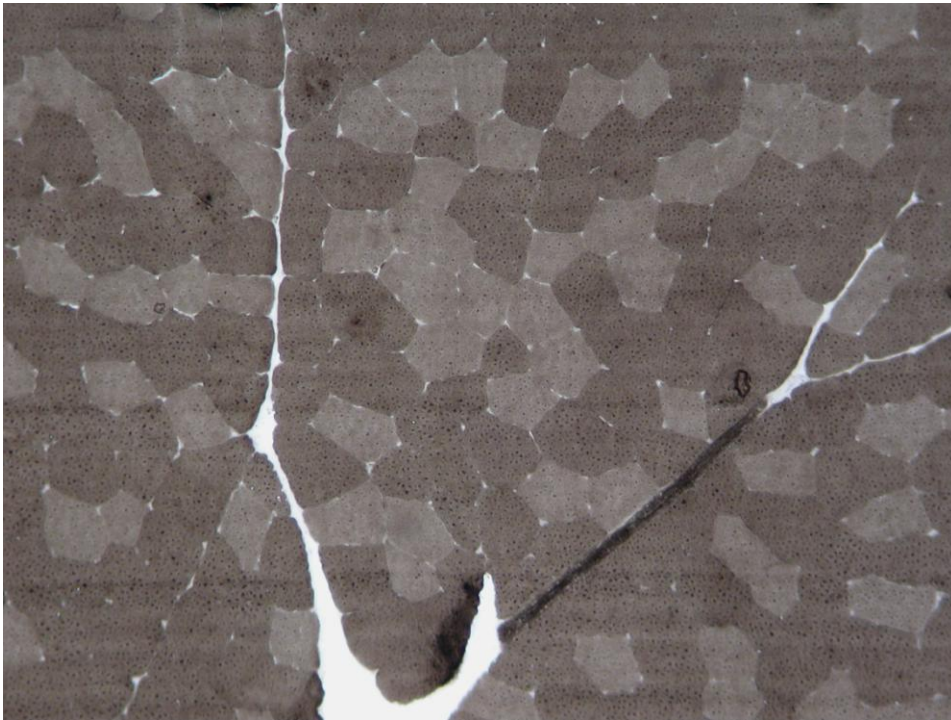
# **HISTOENZYMOLOGIE : lien entre la morphologie et la biochimie des tissus**

- Met en évidence les divers types de fibres
- Donne un aspect fonctionnel de la cellule (éventuel déficit enzymatique , évaluation de l'activité de certains organites intracellulaires :mitochondrie , lysosome , réticulum endoplasmique)
- Peut montrer des variations architecturales non visibles avec les techniques de routine

# ATPases myofibrillaires

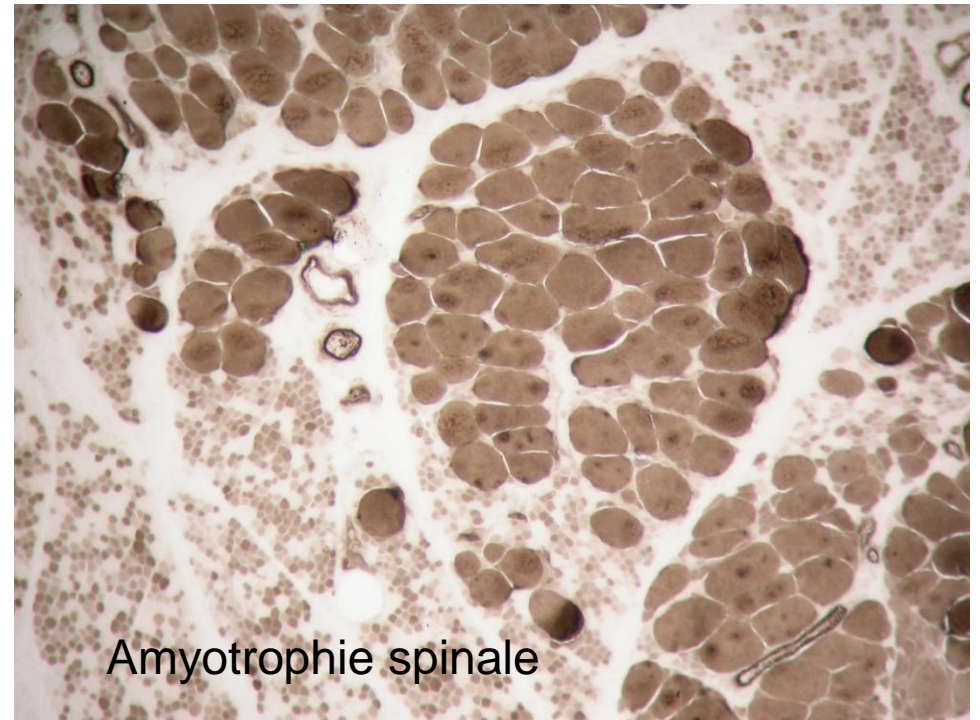
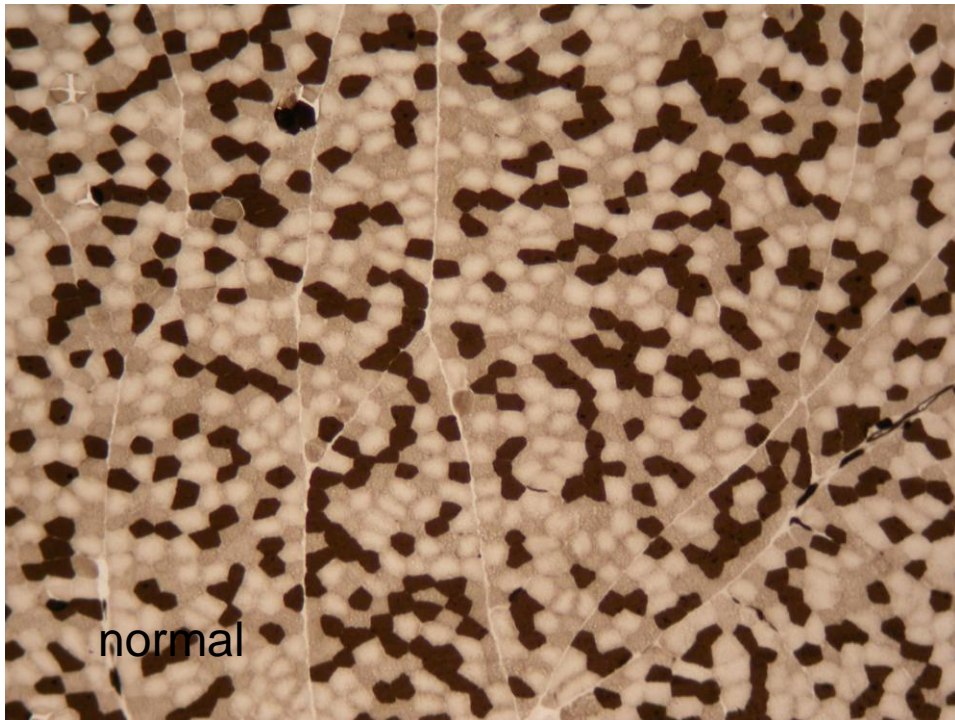
ph 4,35

ph 9,4

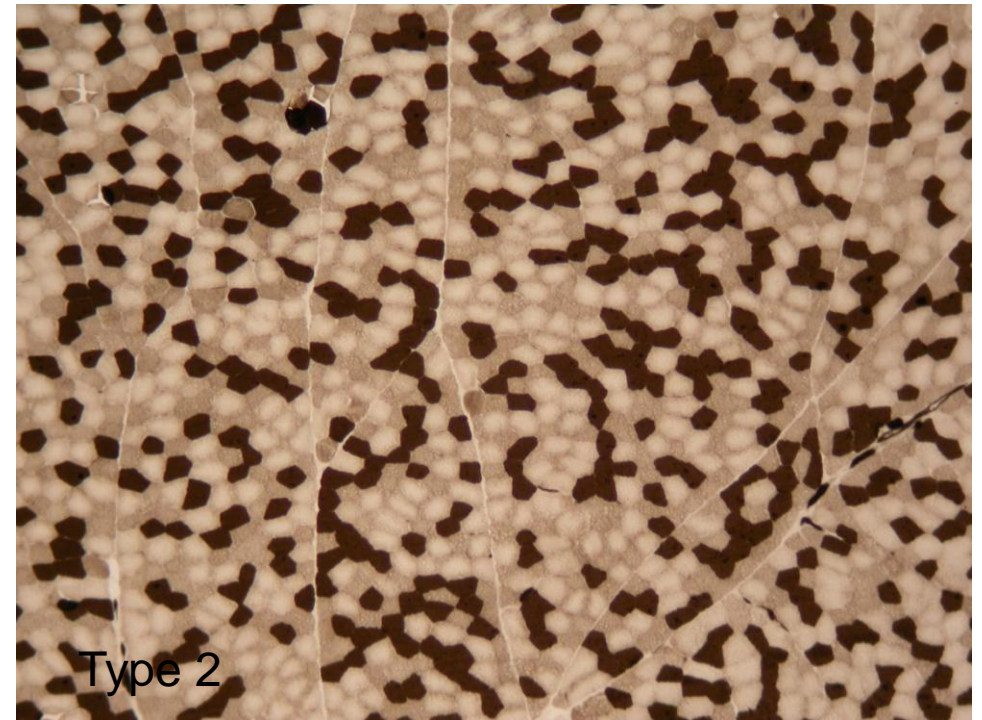
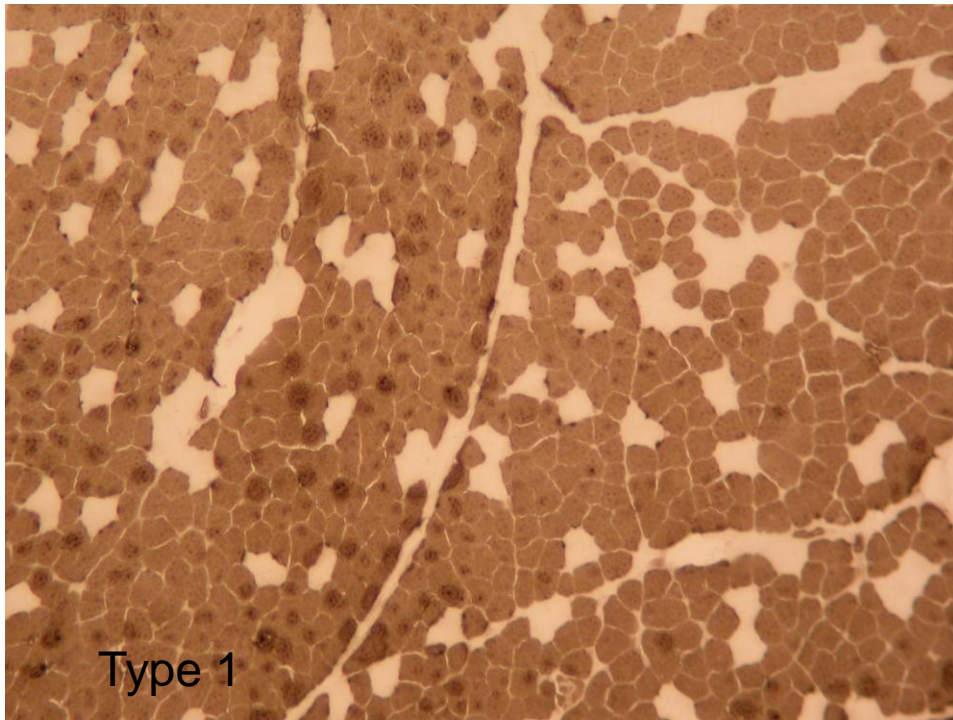


**Substratum : myosine**

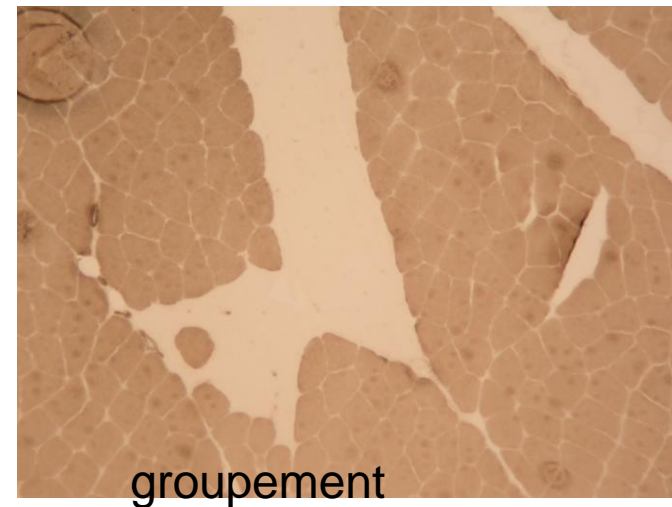
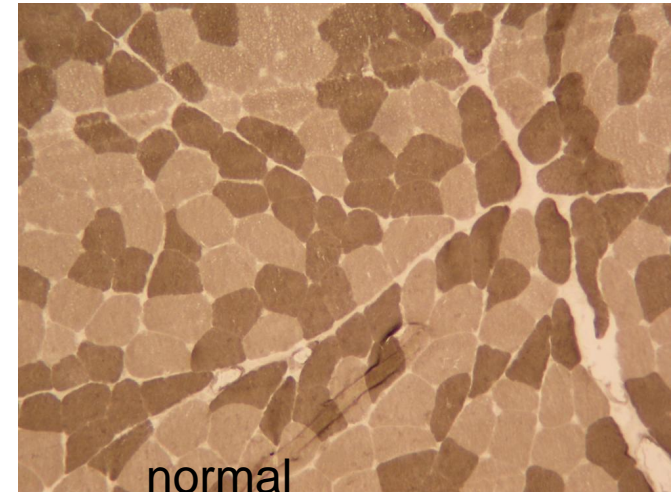
# ATPase 4,35 : processus neuropathique



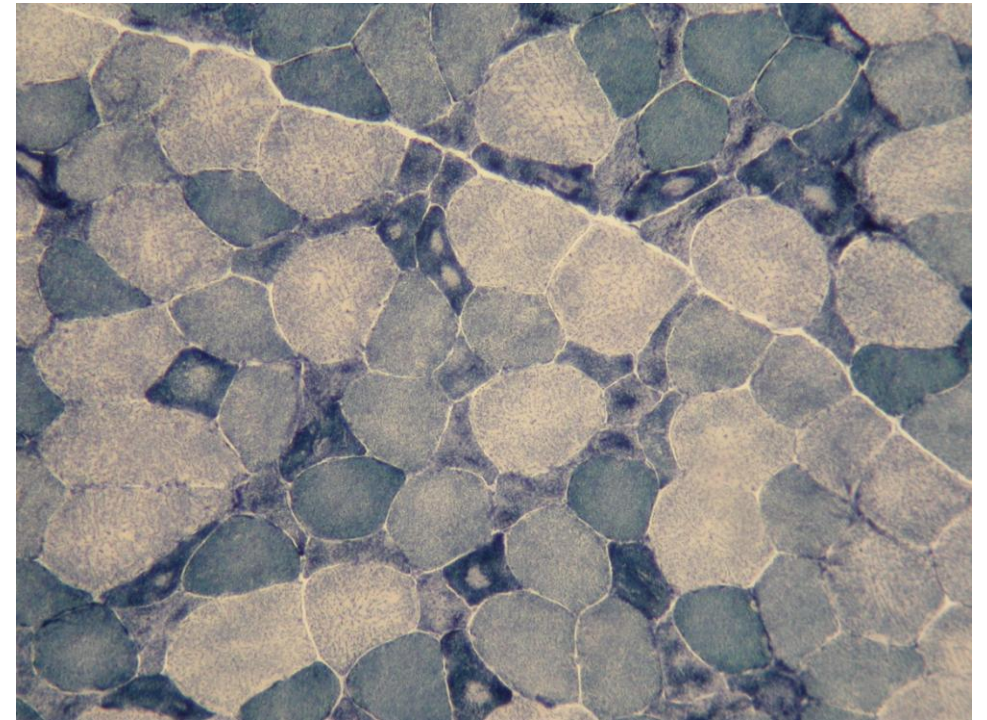
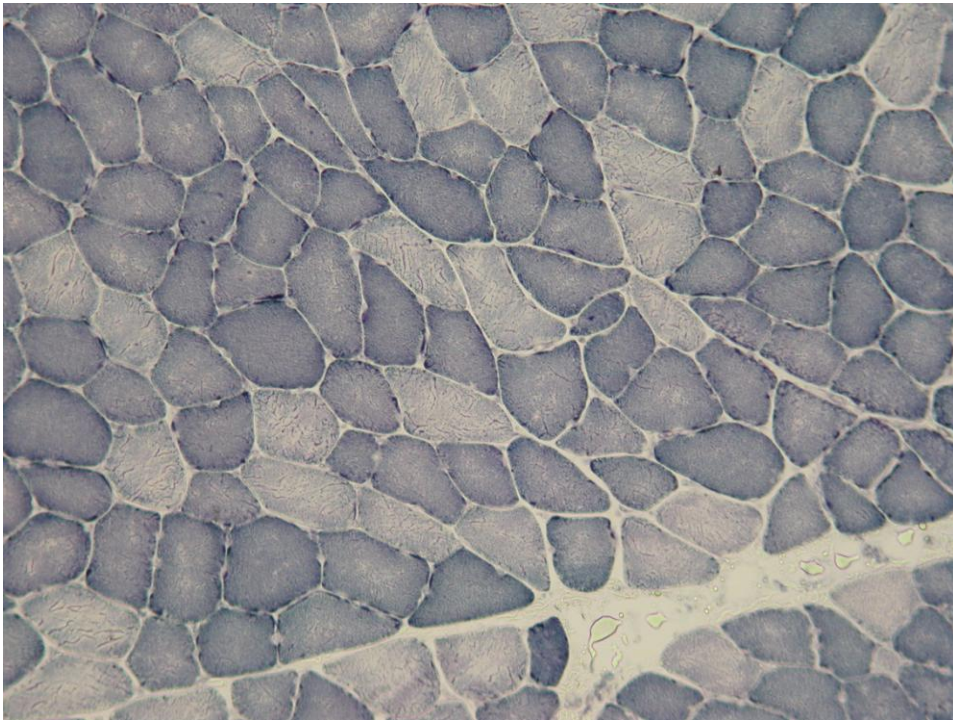
# ATPase 4,35: predominance d'un type de fibres



# Groupement des mêmes types de fibres histochimiques

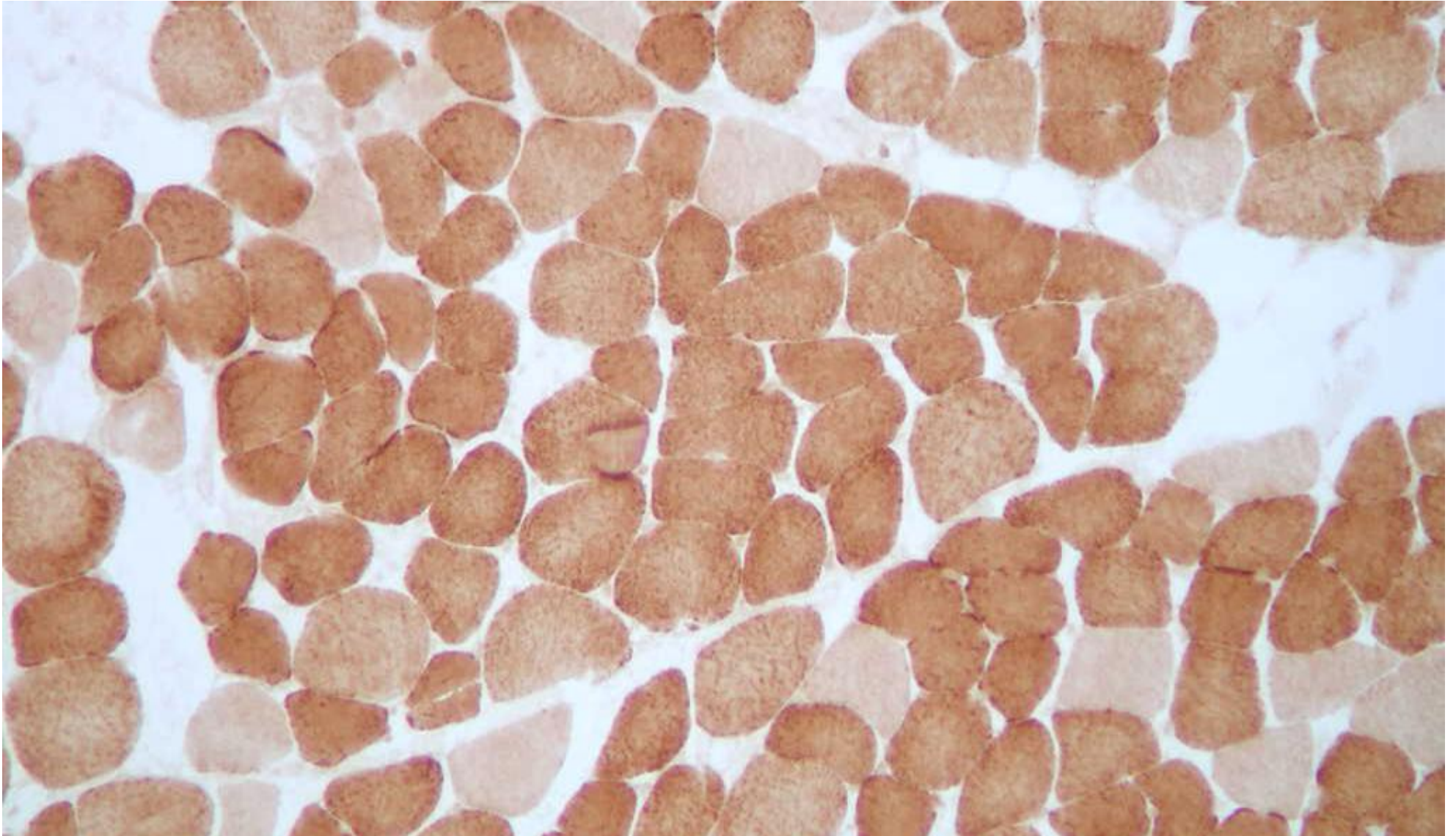


# NADH : ATROPHIE NEUROPATHIQUE



Localisation intermyofibrillaire ,  
Substratum : mitochondries réticulum et système T

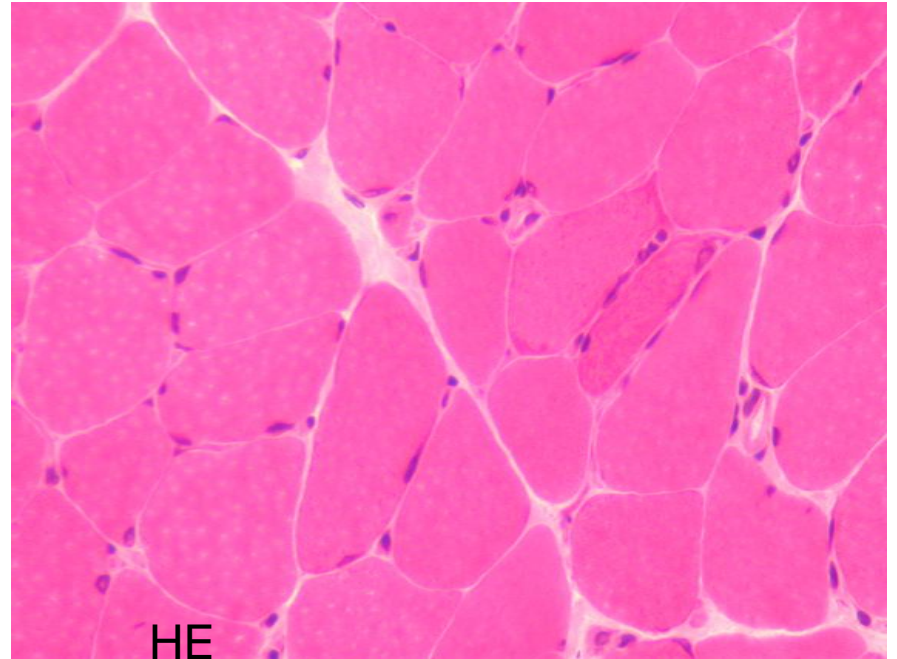
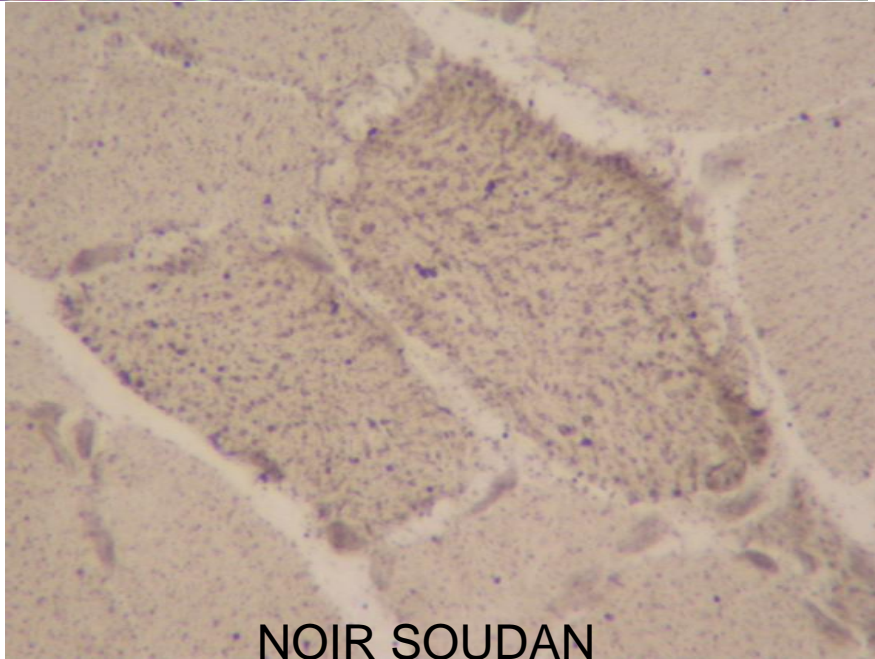
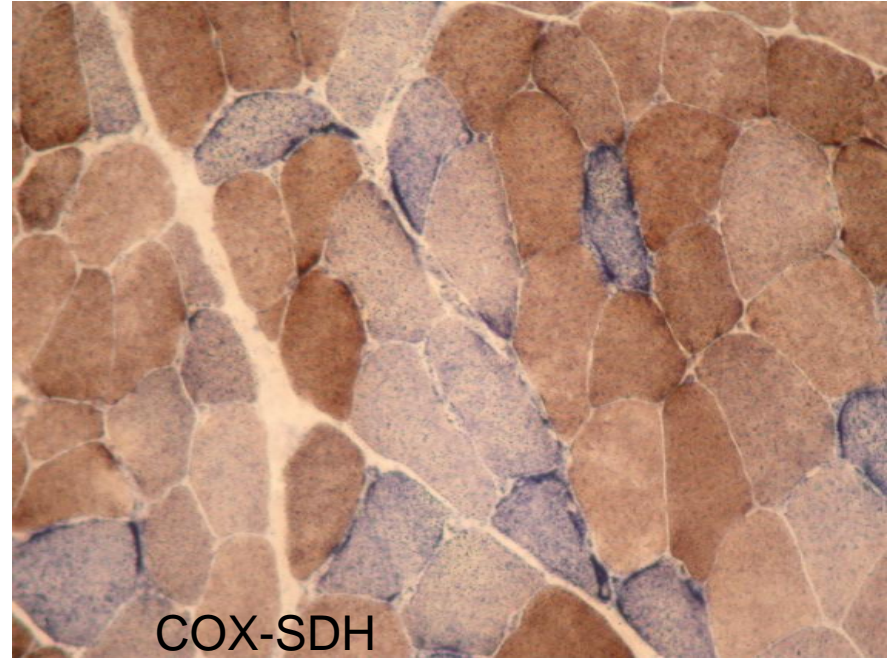
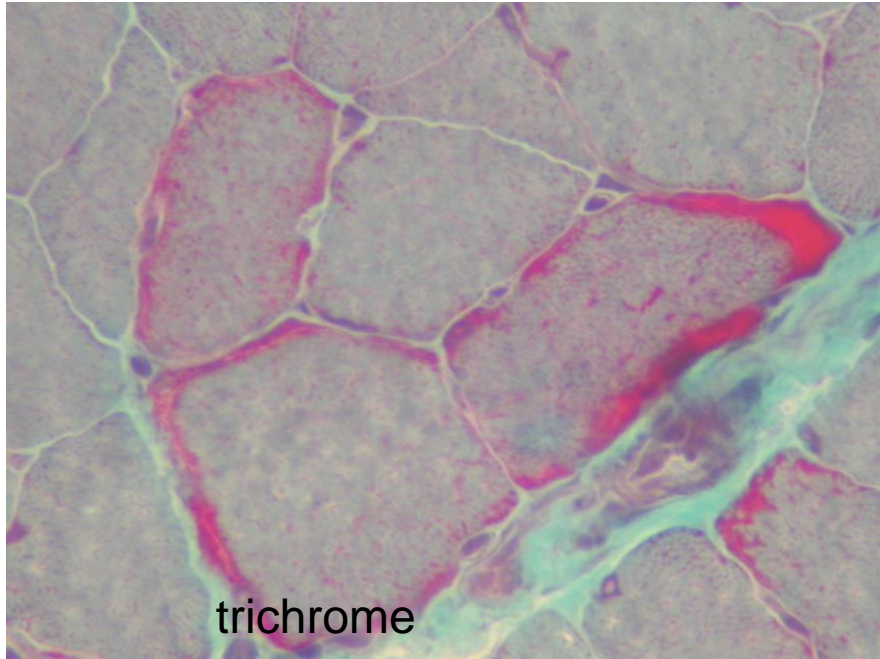
# Cytochrome oxydase-succinodéshydrogénase

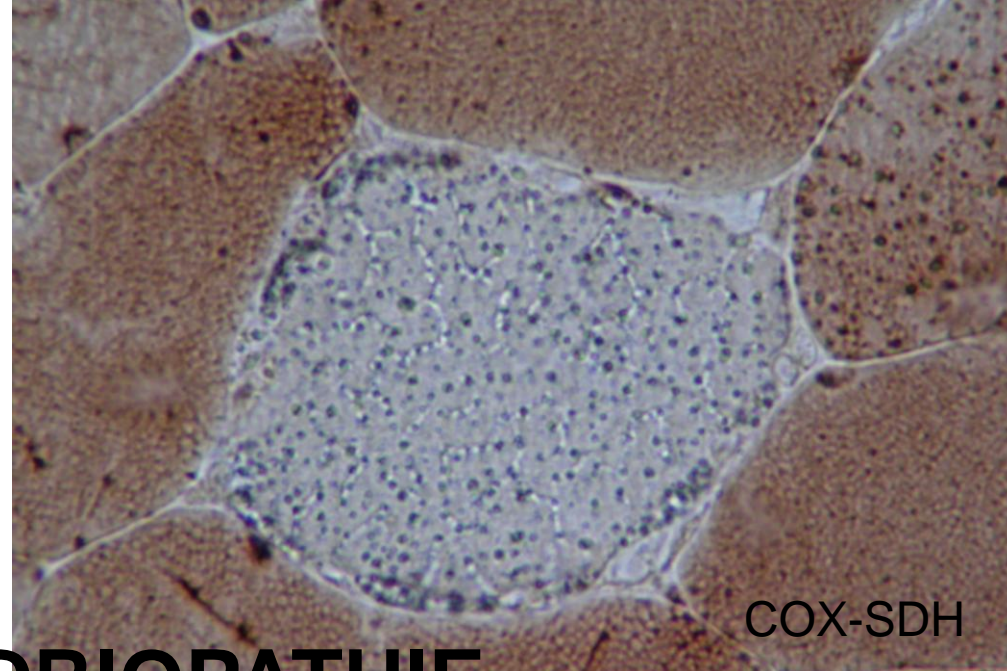
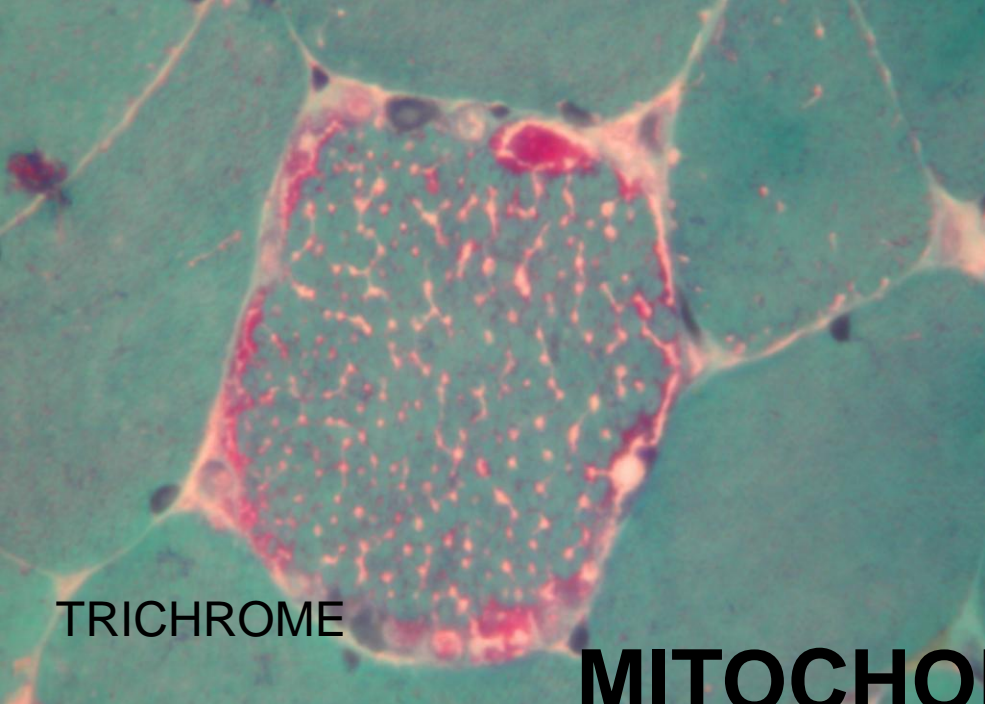


Localisation intermyofibrillaire

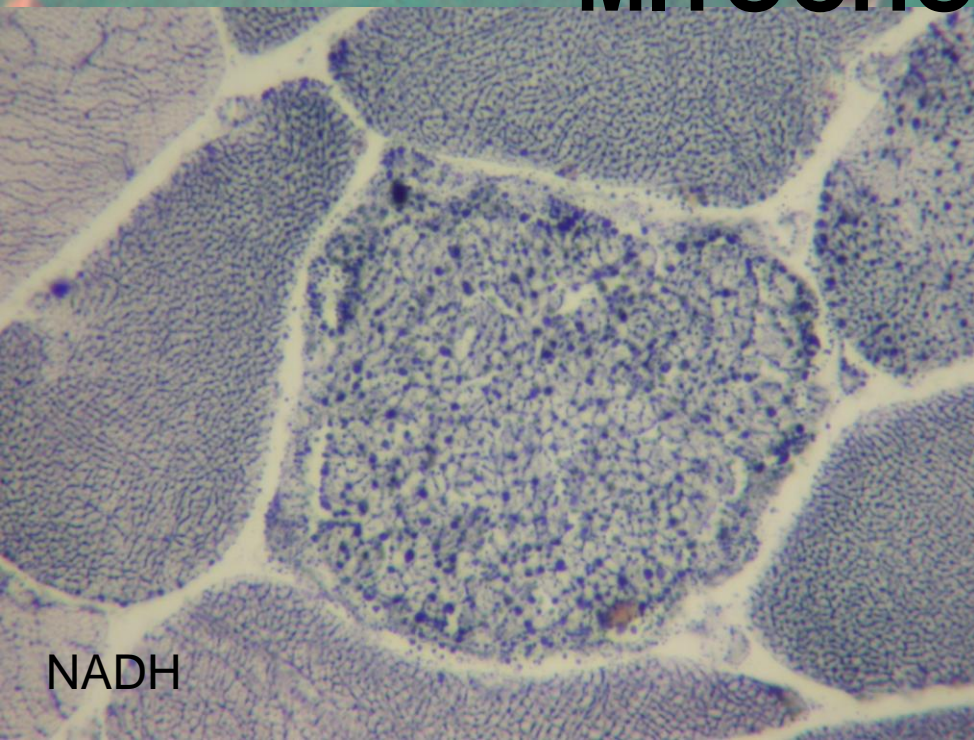
substratum mitochondries

# MITOCHONDRIOPATHIE





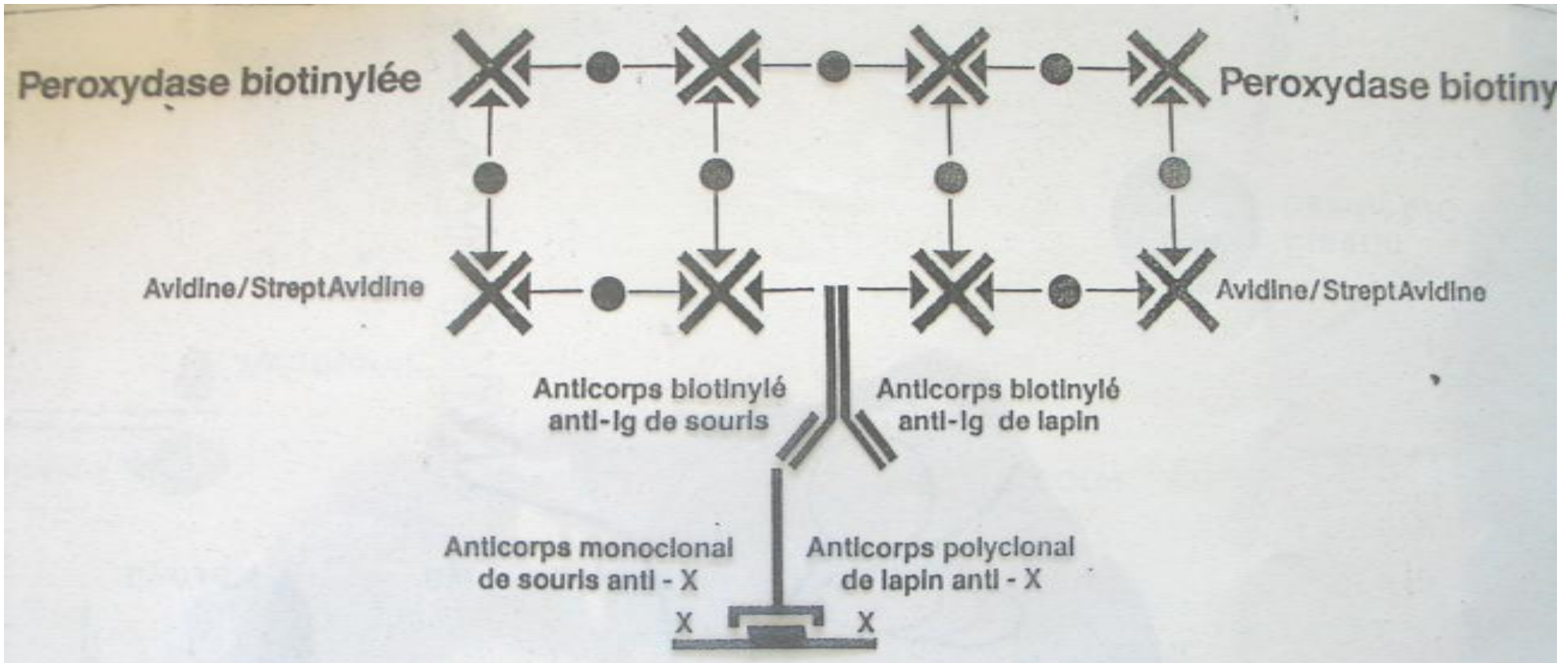
# MITOCHONDRIOPATHIE



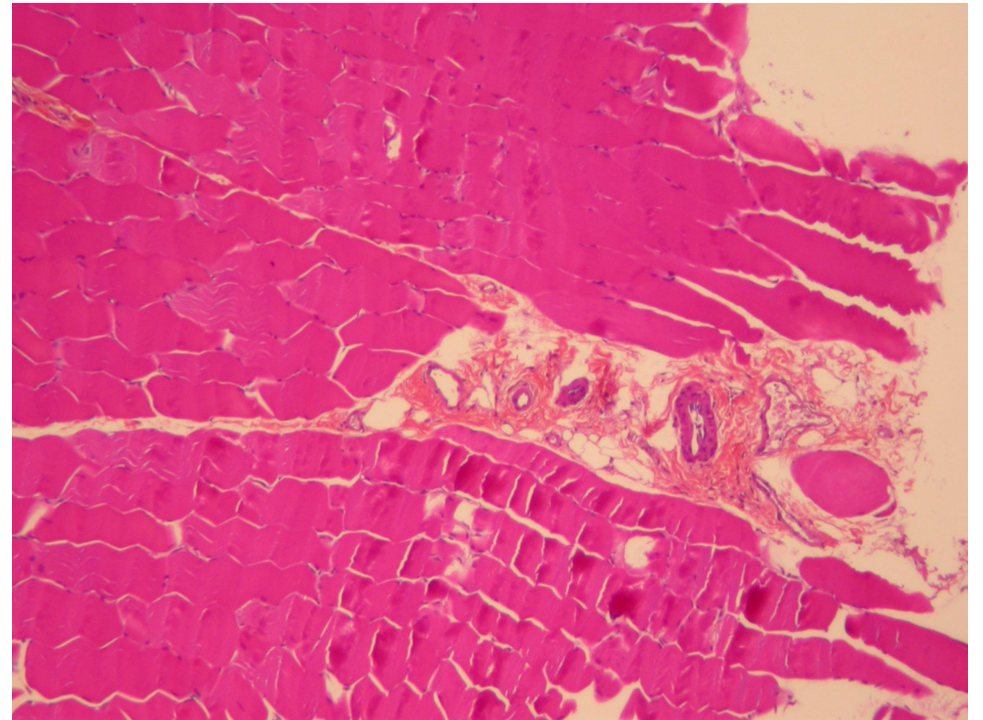
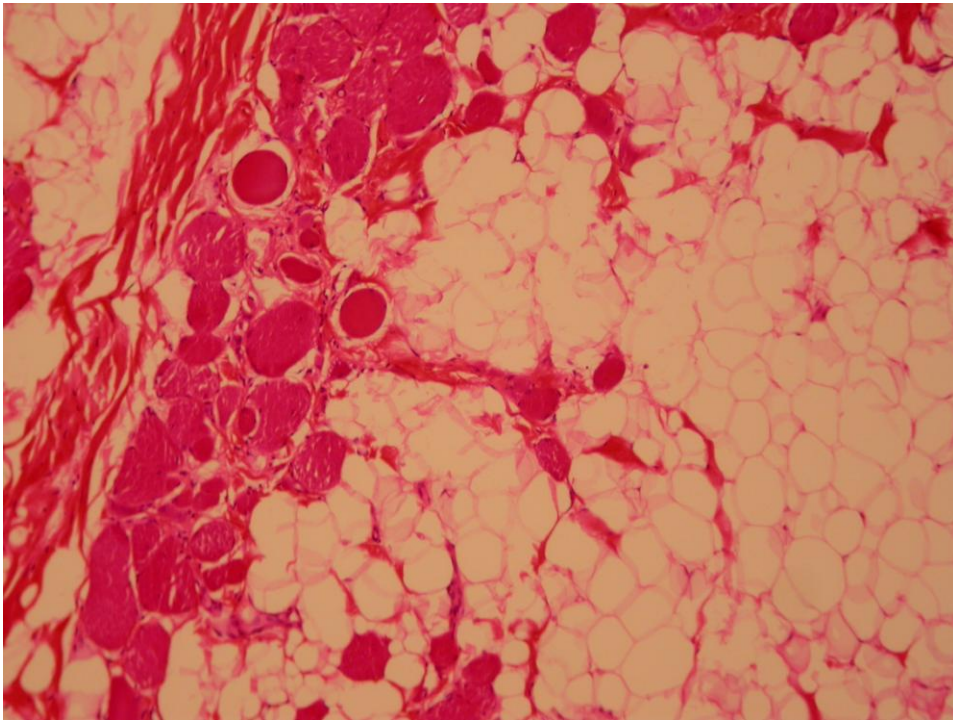
**Immunohistochimie** : mise en évidence d'un antigène dans un tissu

- Étude des protéines du cytosquelette dans les dystrophies musculaires
- Qualification de l'infiltrat leucocytaire dans les myopathies inflammatoires

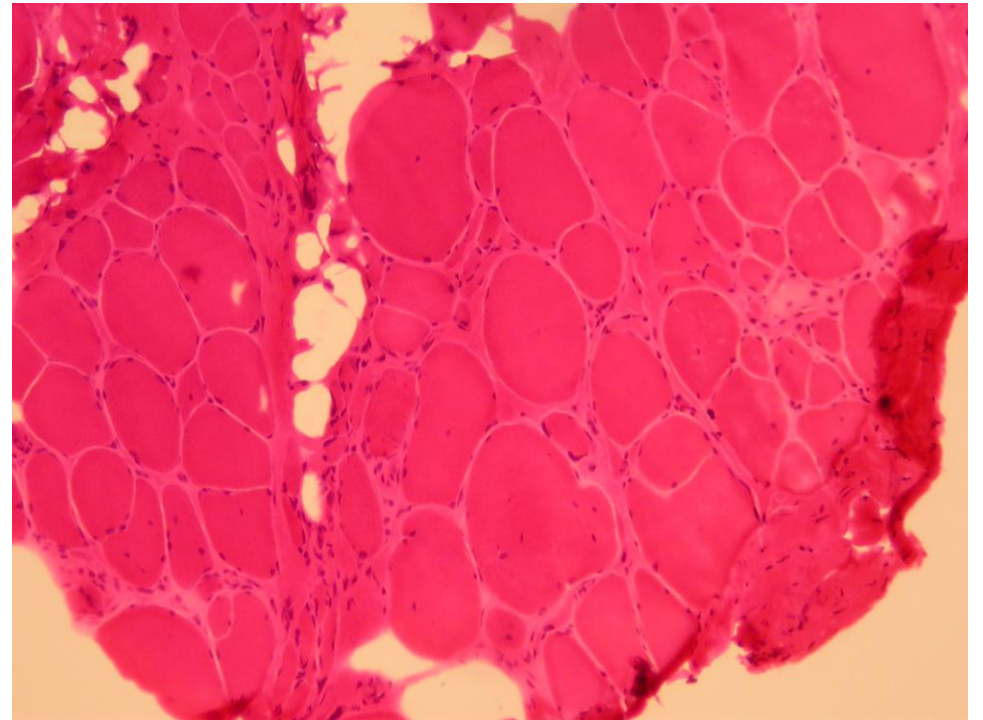
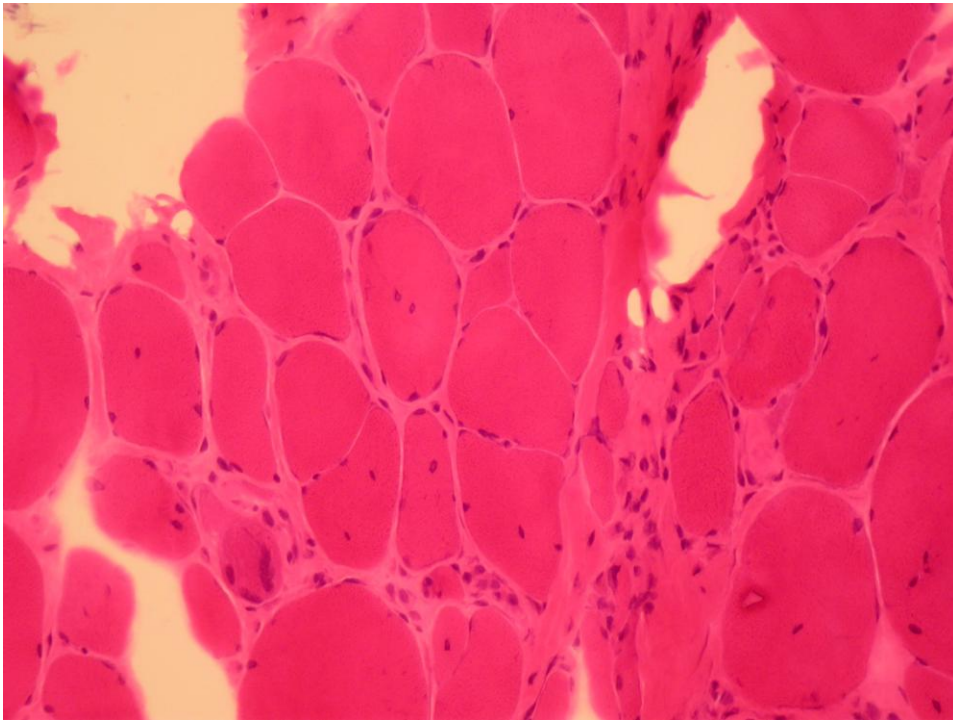
# Technique PAP



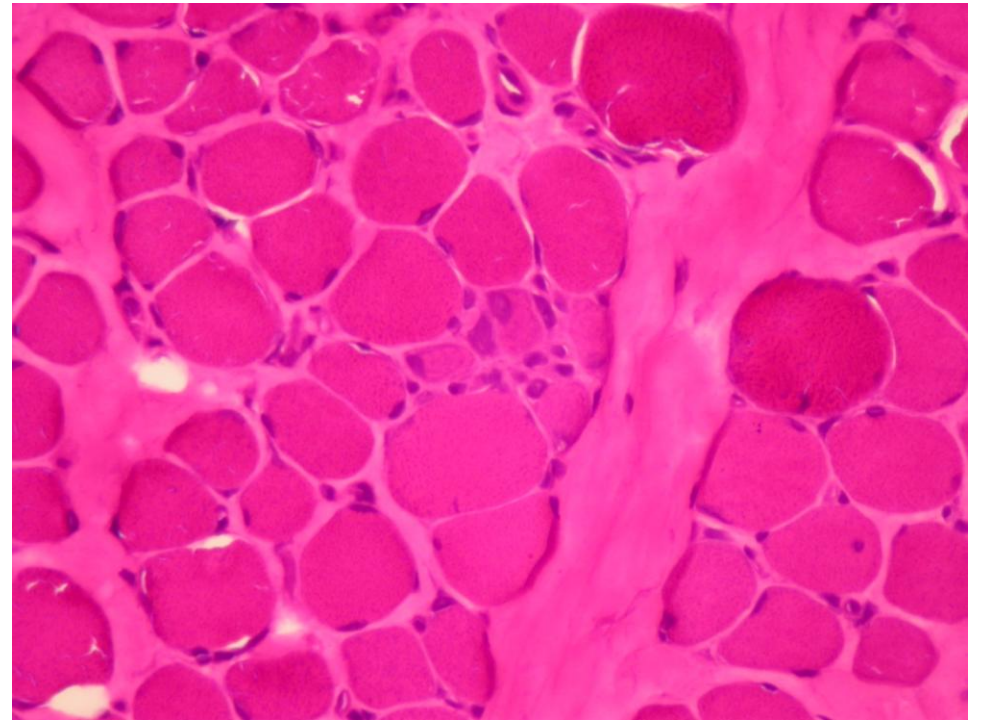
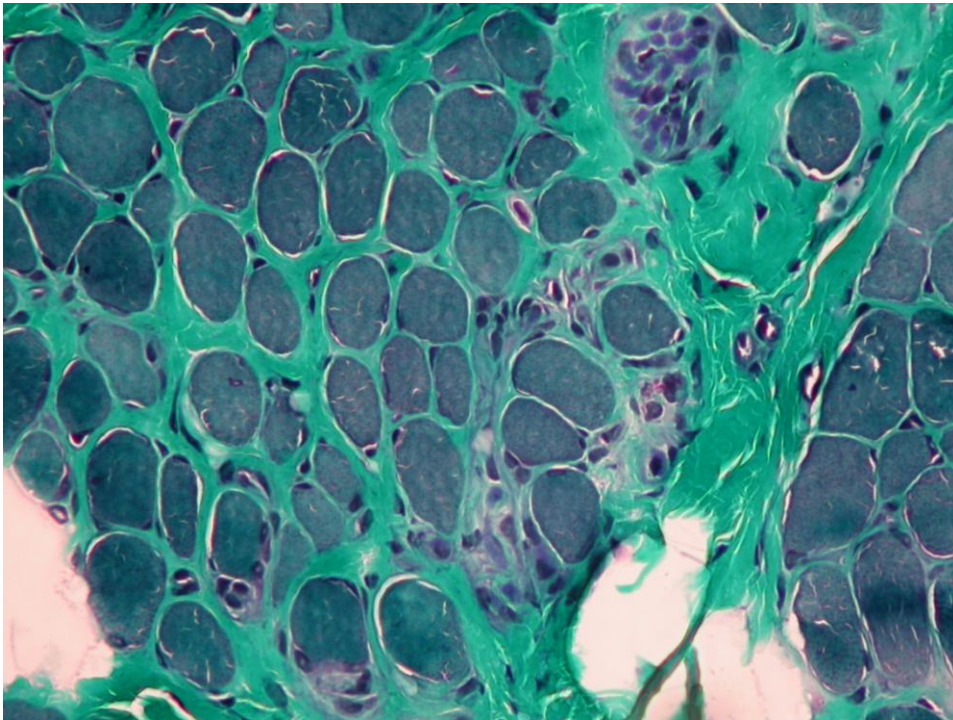
# MYOPATHIE DE DUCHENNE

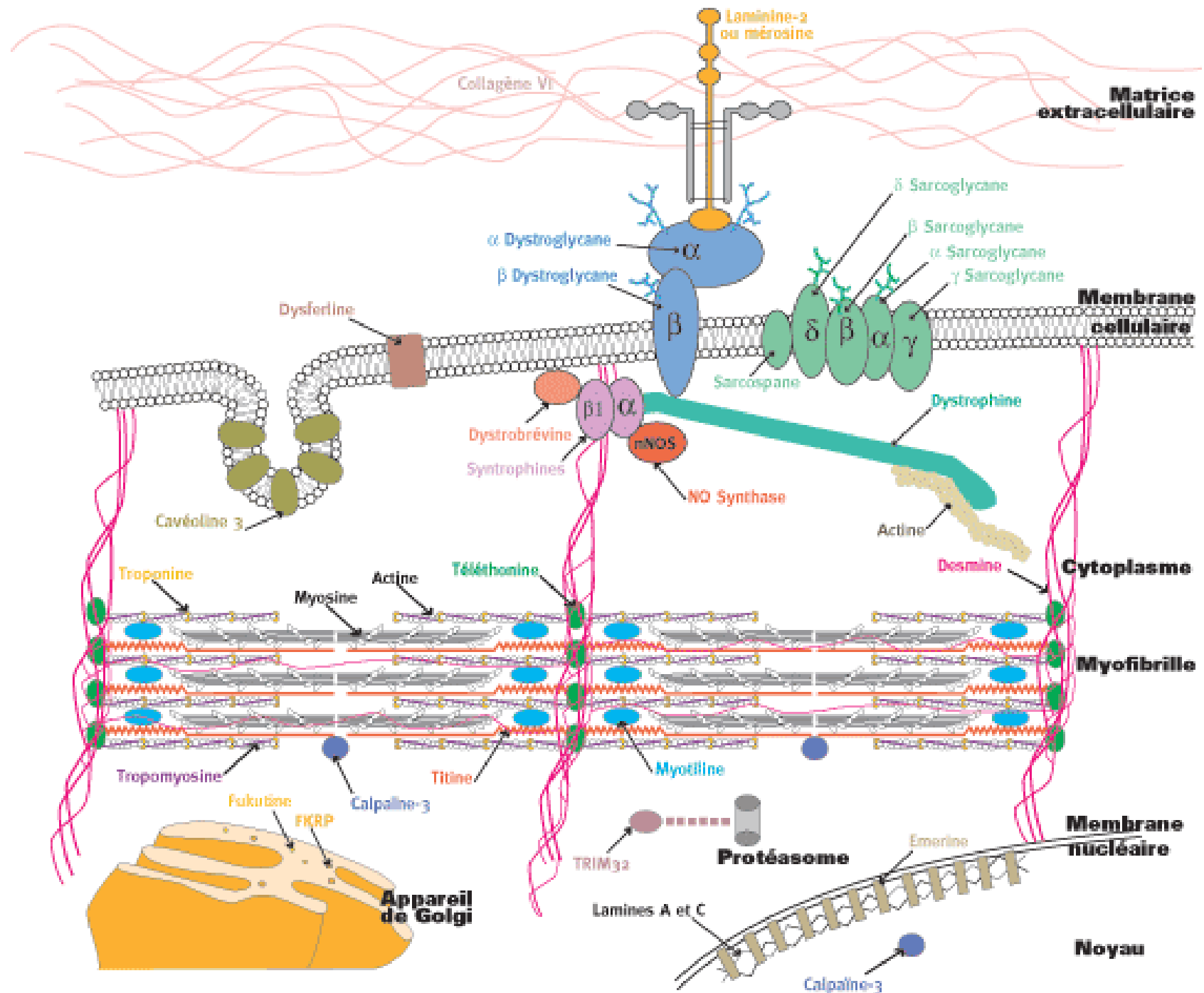


# Myopathie de Duchenne

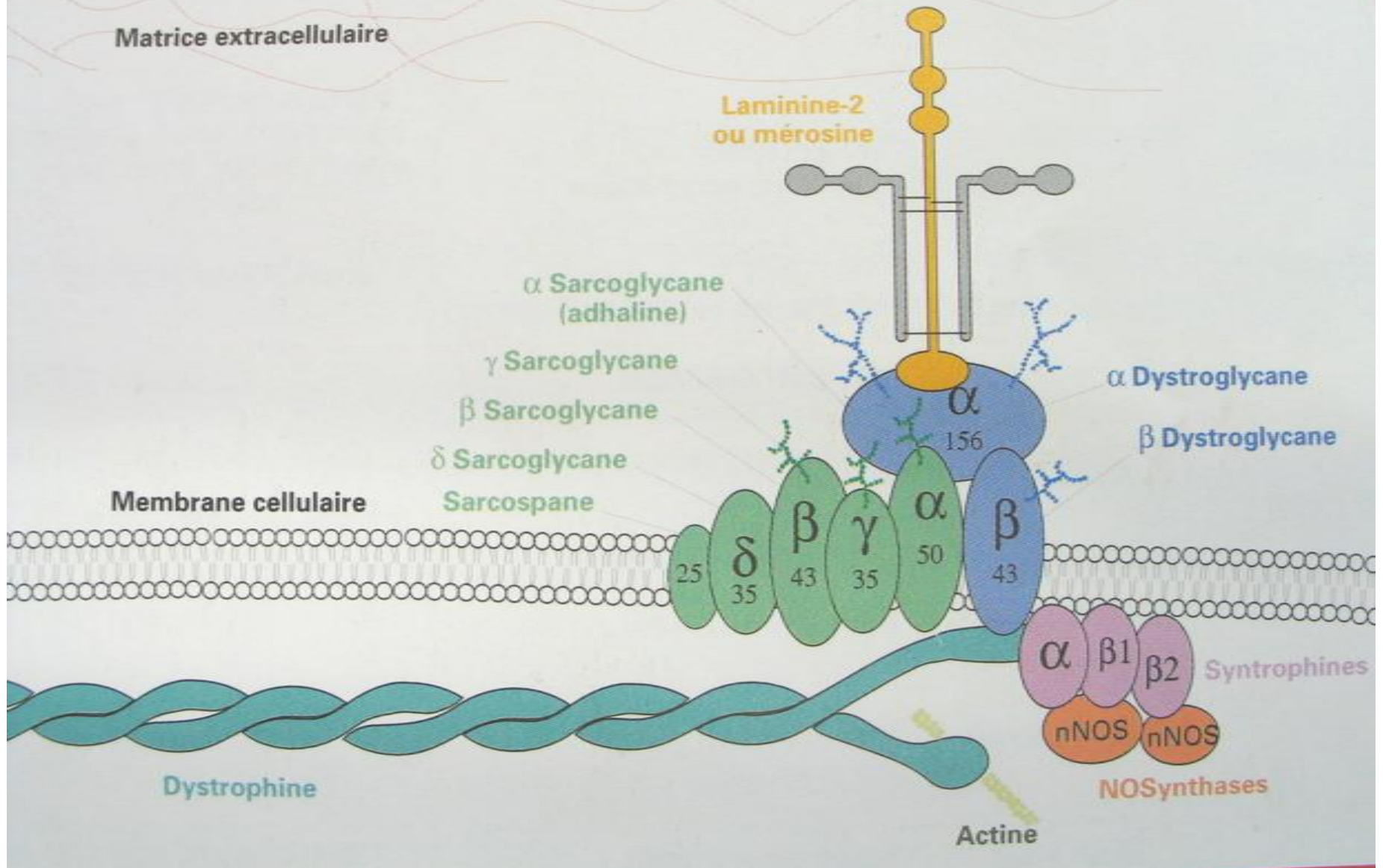


# Myopathie de Duchenne

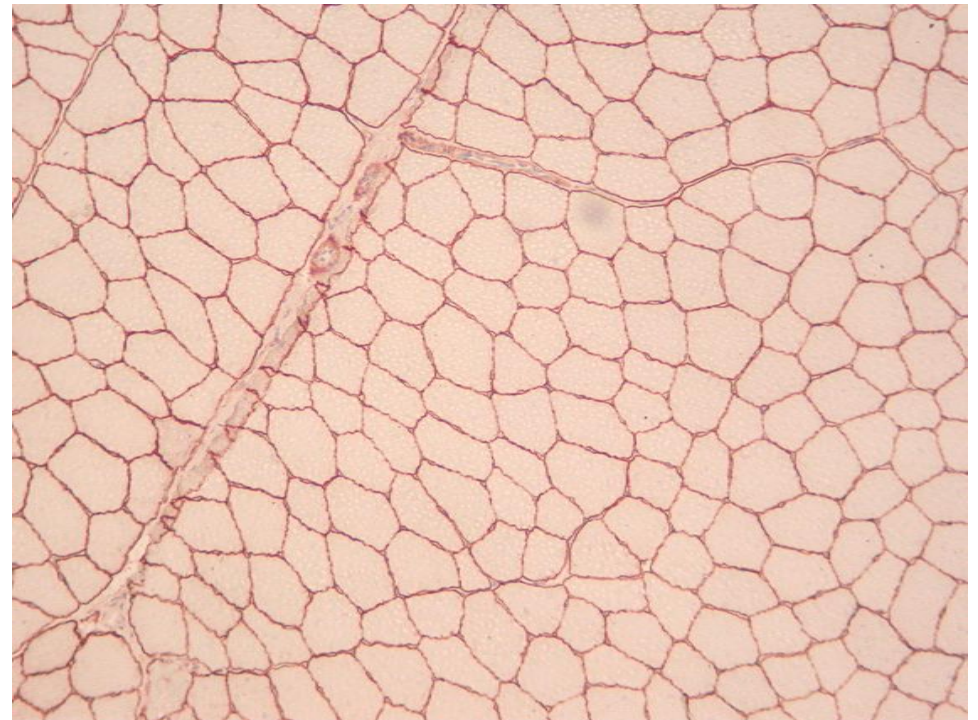
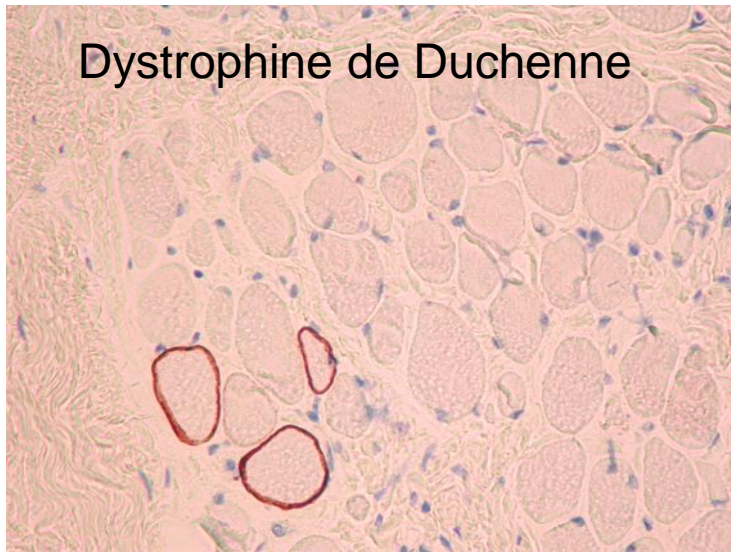
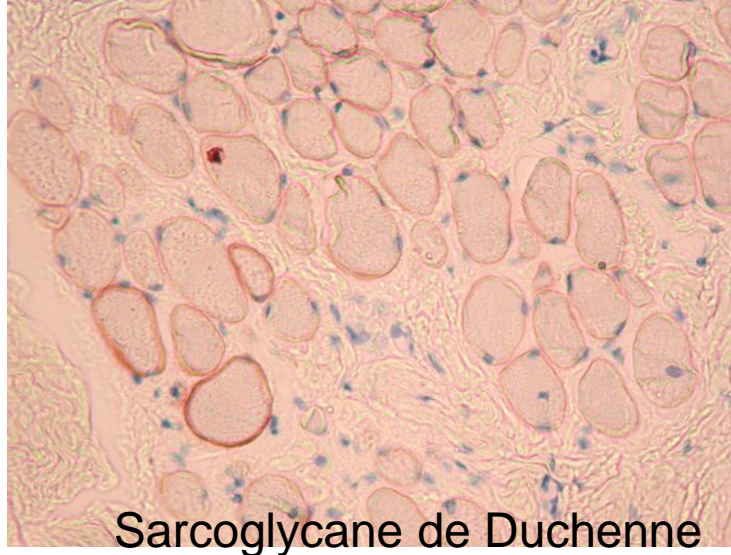




# PROTEINES DU CYTOSQUELETTE

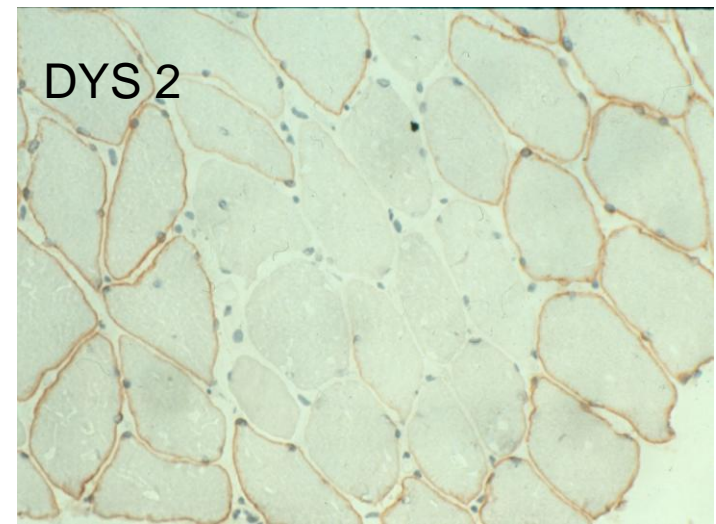
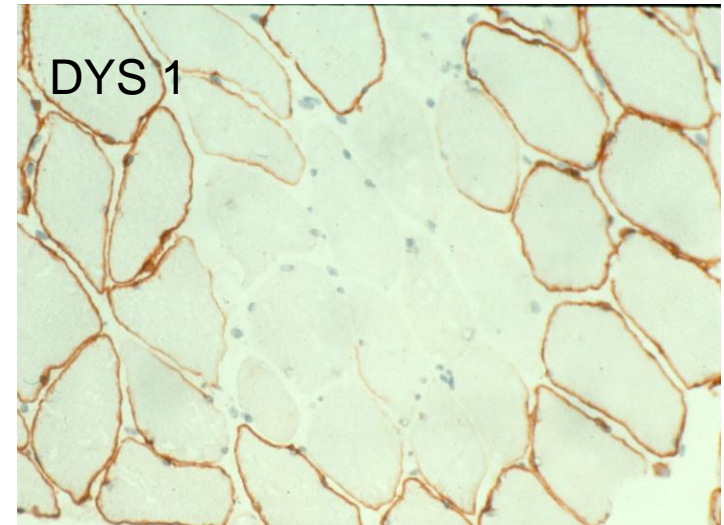
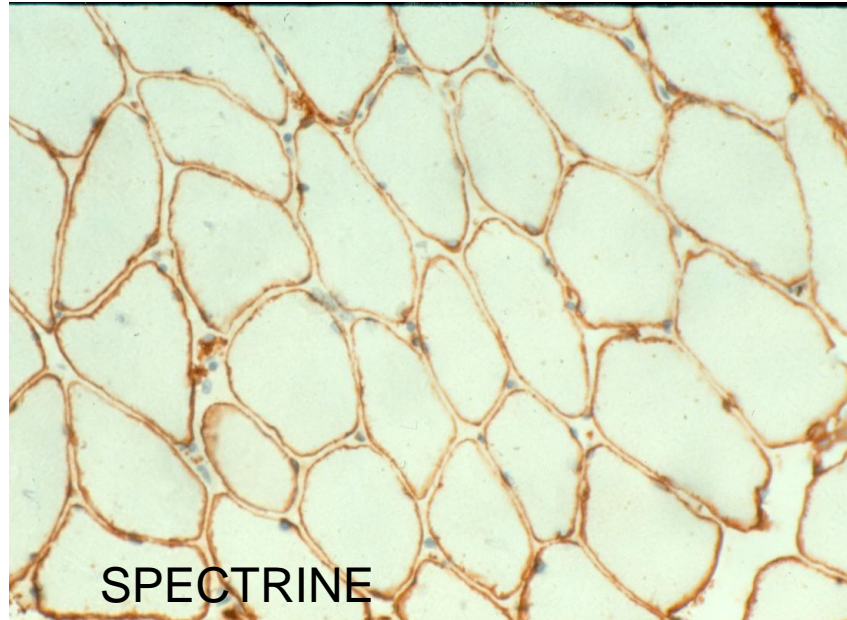


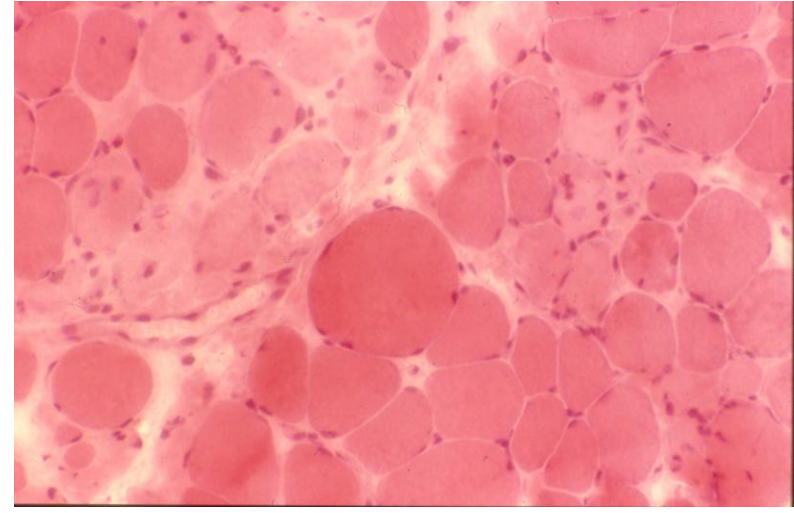
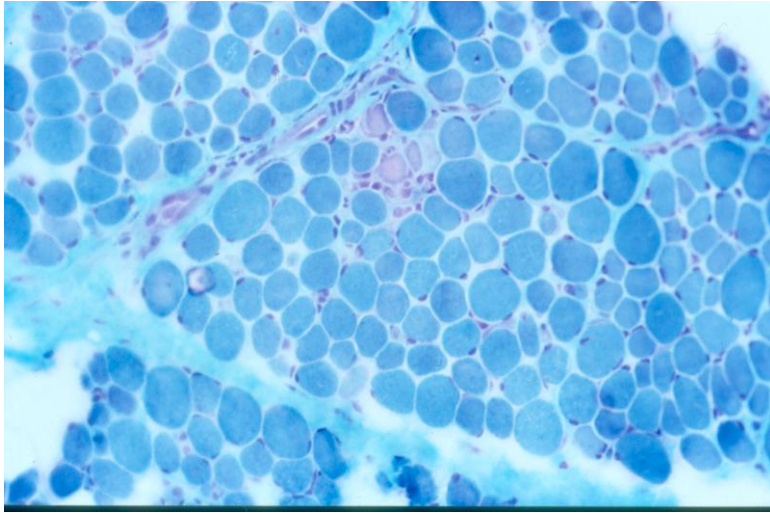
# IMMUNOHISTOCHEMIE



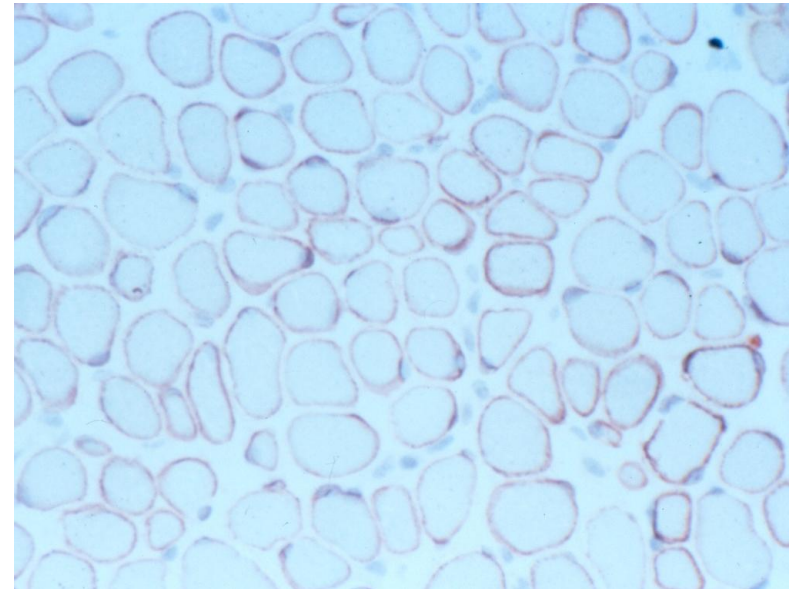
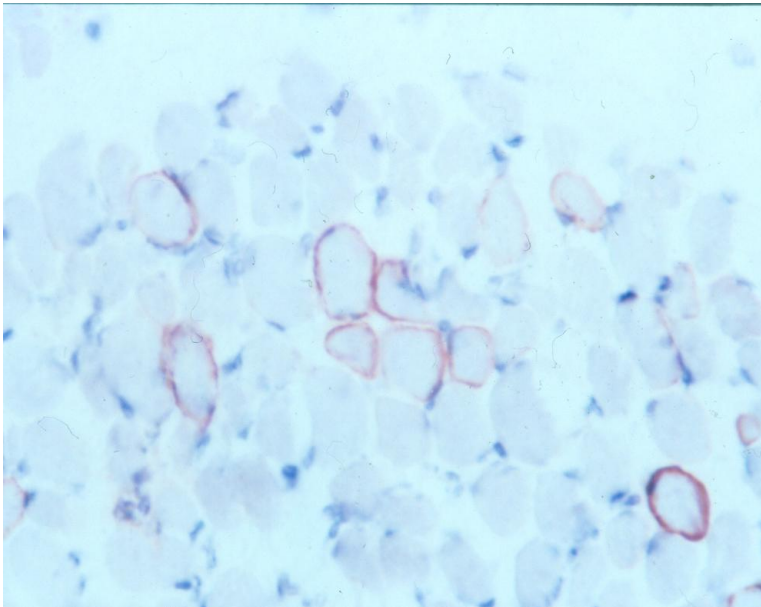
Dystrophine témoin

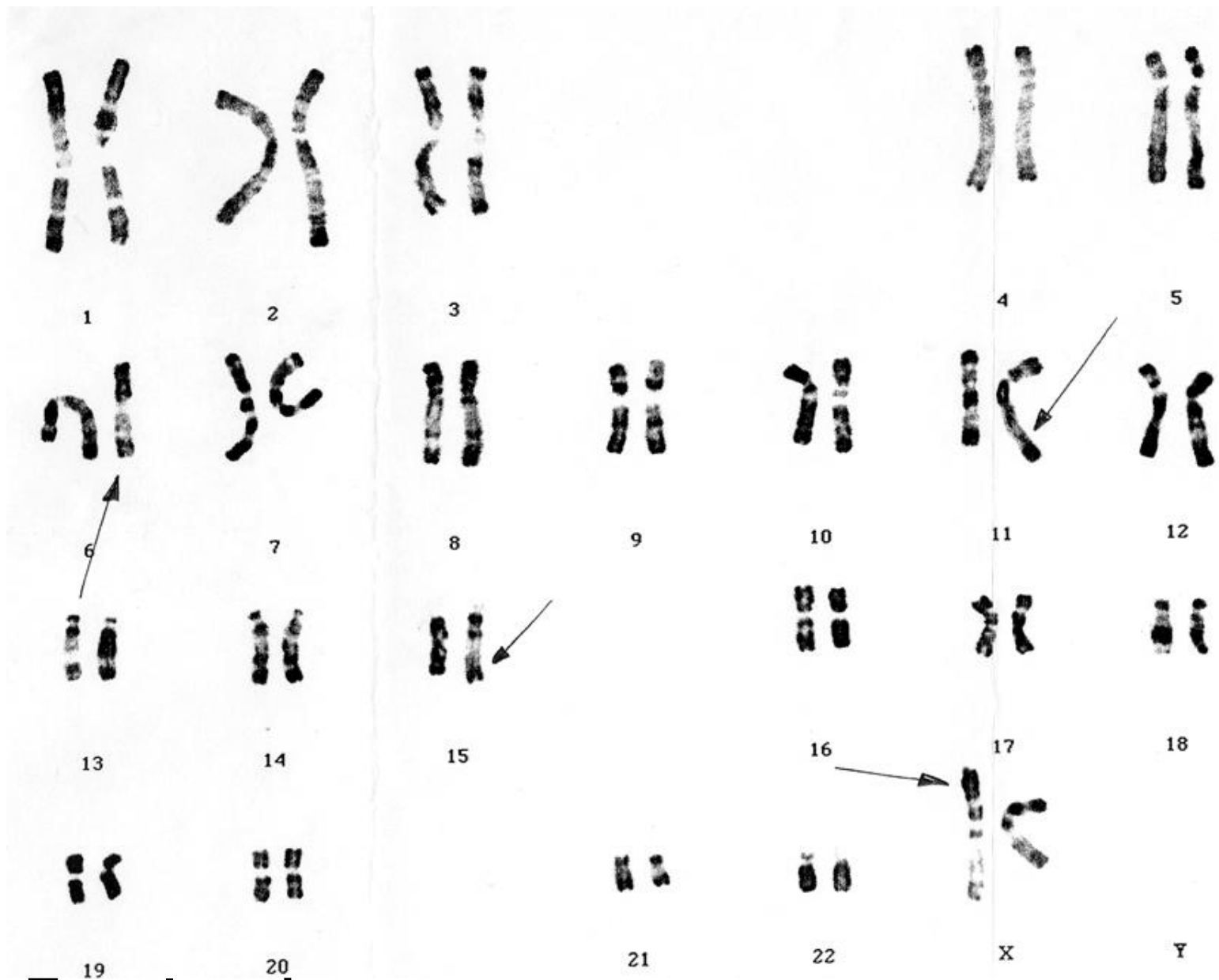
# IHC : porteuse de Duchenne



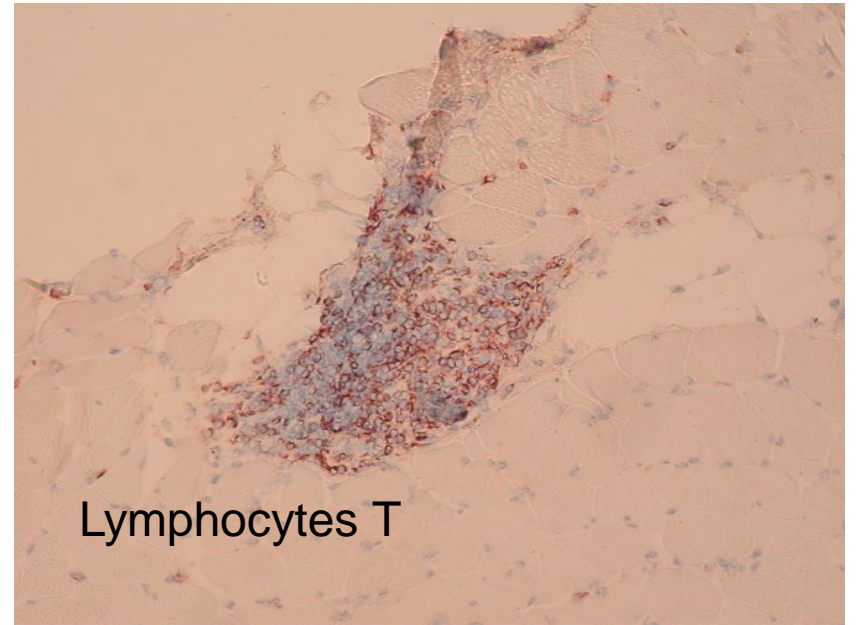


## **DUCHENNE FEMININ**

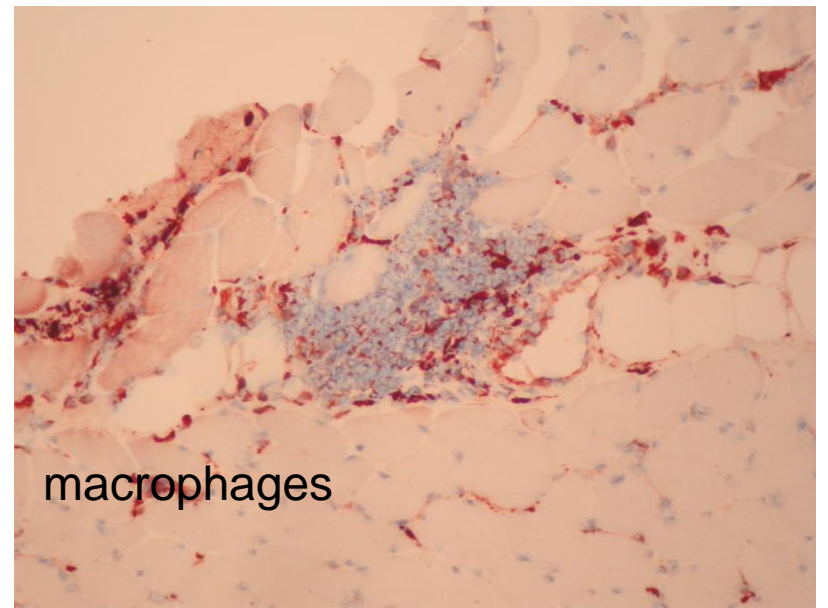
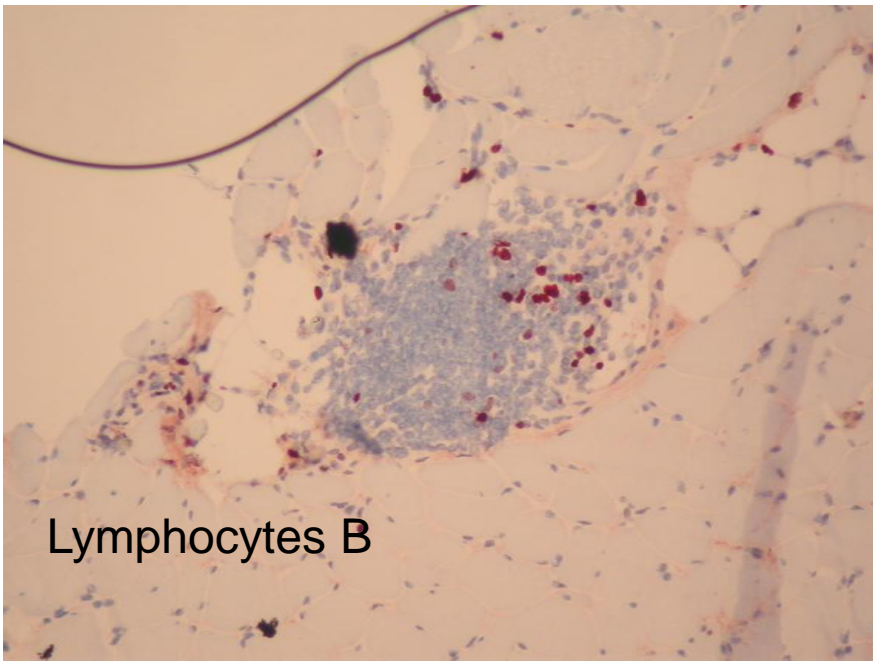




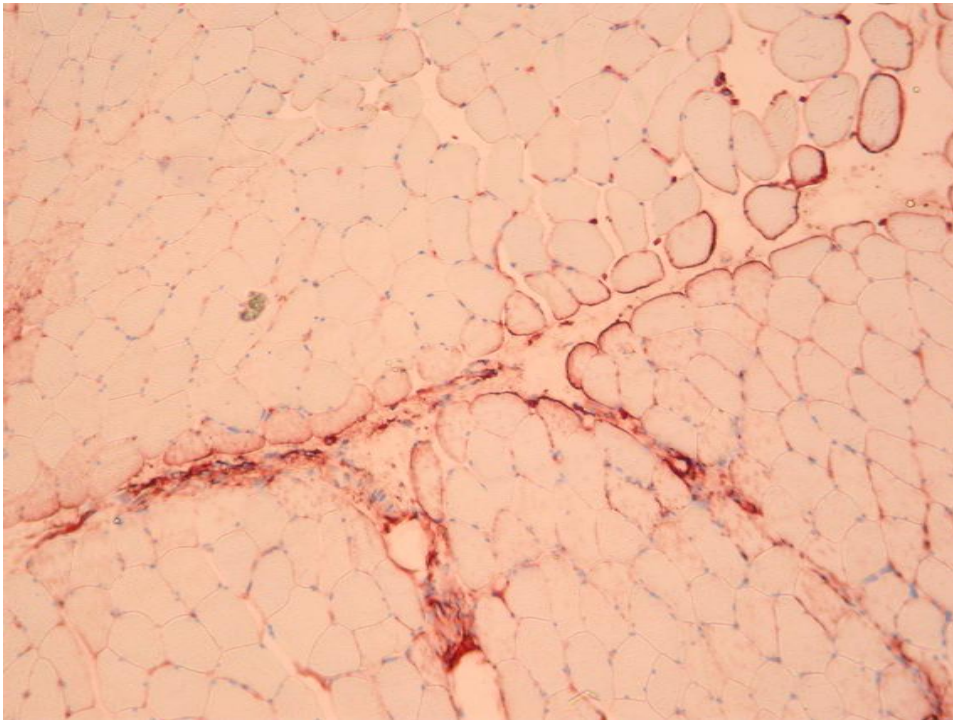
**Translocation x autosome**



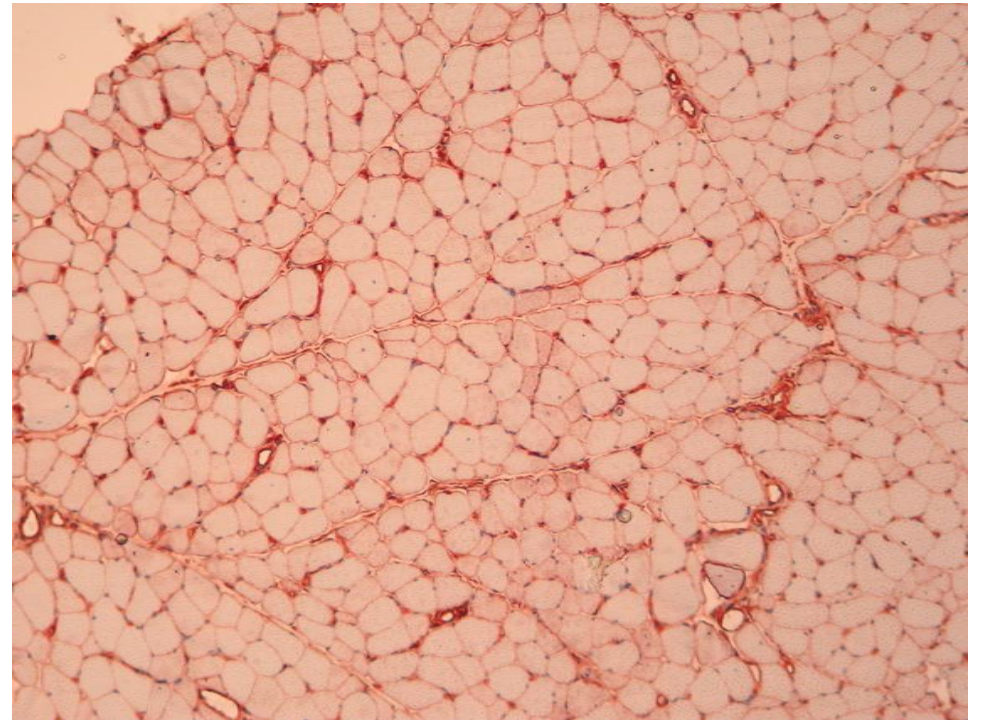
## MYOSITE



# MYOSITE : HLA de classe 1



dermatomyosite



polymyosite