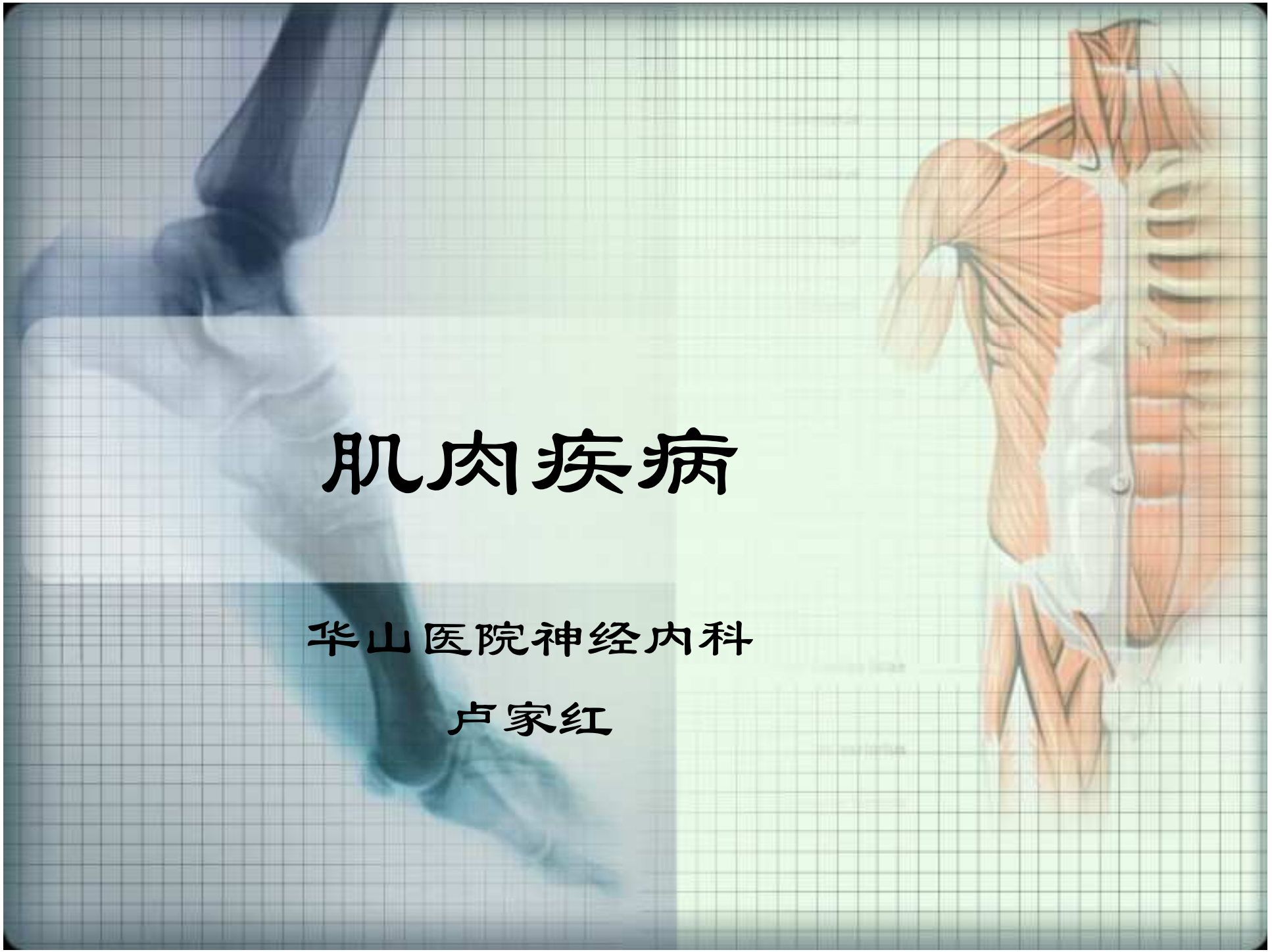
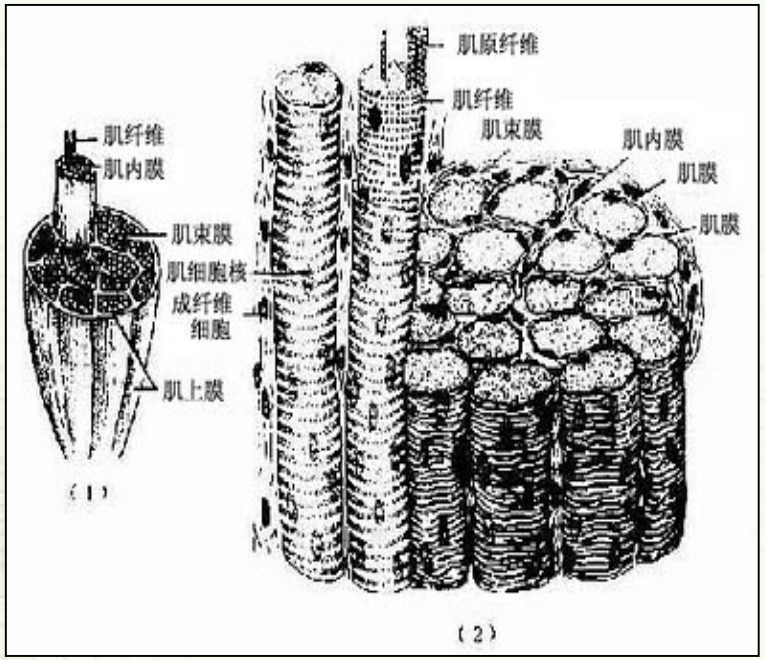


# 肌肉疾病

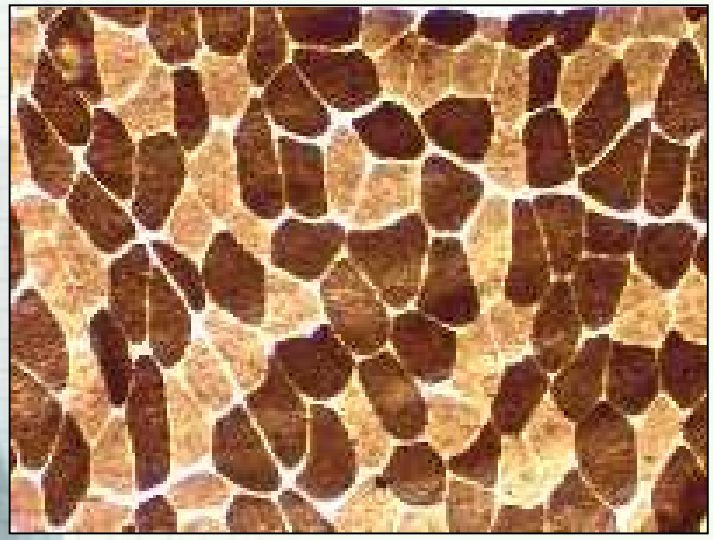
华山医院神经内科

卢家红

# 概述




骨骼肌组成



ATPase pH 9.4 stain

"Checkerboard" pattern of
type I （Light）and II （Dark）fibers

## 肌纤维类型

I型纤维

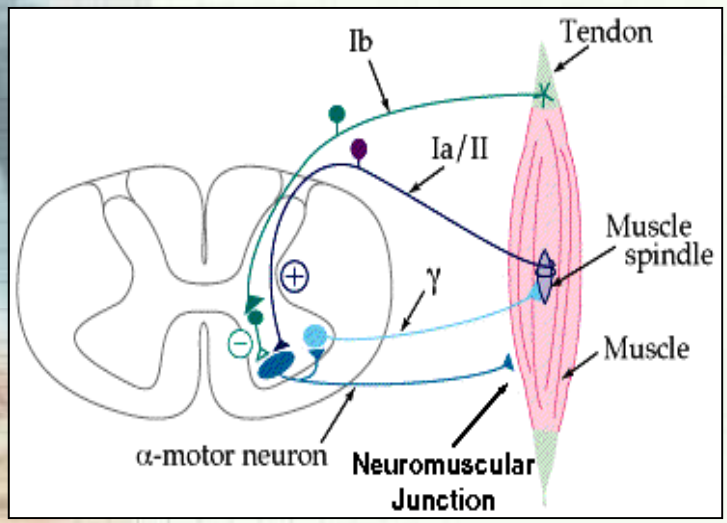
慢收缩纤维

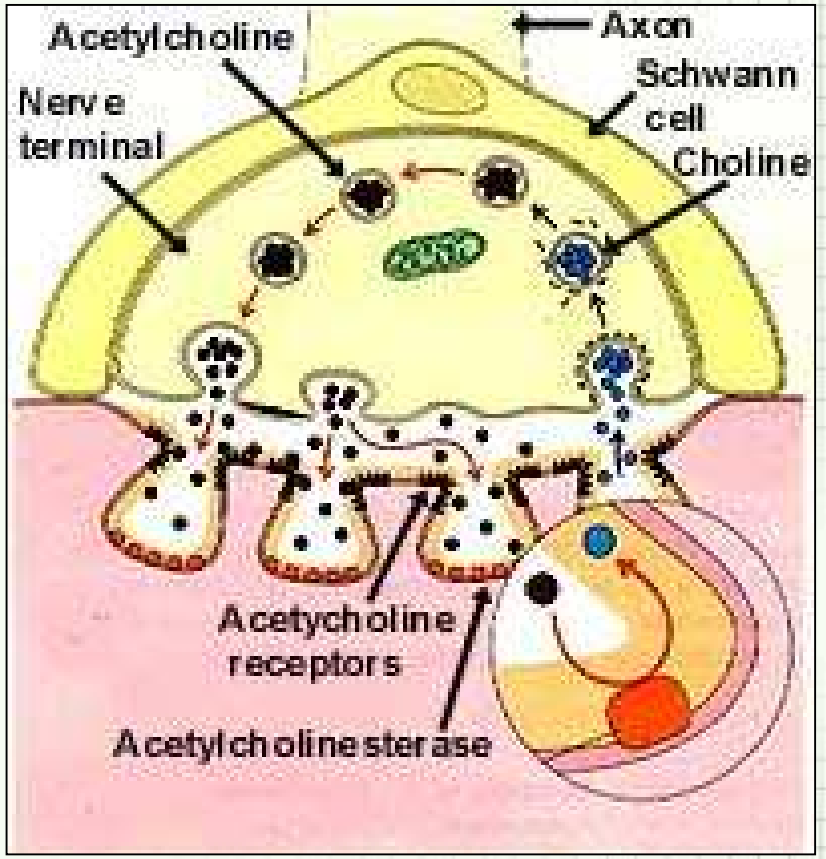
II型纤维

快收缩纤维

## 神经肌肉接头（NMJ）

## 运动单位



# 肌肉疾病分类

- 肌肉本身
  - 通道病
  - 肌营养不良
  - 炎症性肌病
  - 代谢性肌病
  - 先天性肌病
- 神经肌肉接头
  - 突触前膜病变（LES）
  - 突触间隙病变（有机磷中毒）
  - 突触后膜病变（MG）

# 肌肉疾病特点

## 临床特点

无力：近端、对称

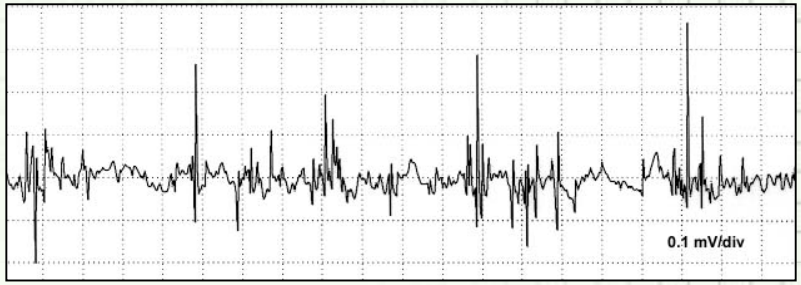
无力＞萎缩

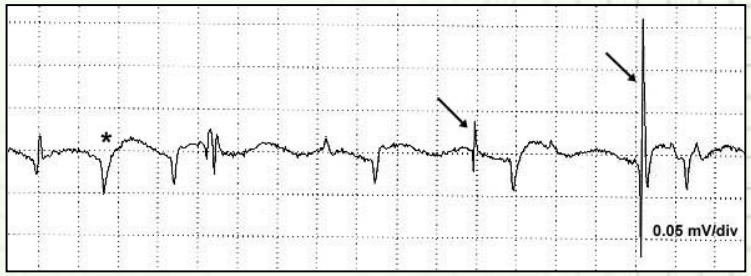
感觉：正常

腱反射：明显无力区域反射减退

# 肌肉疾病特点

## 肌电图



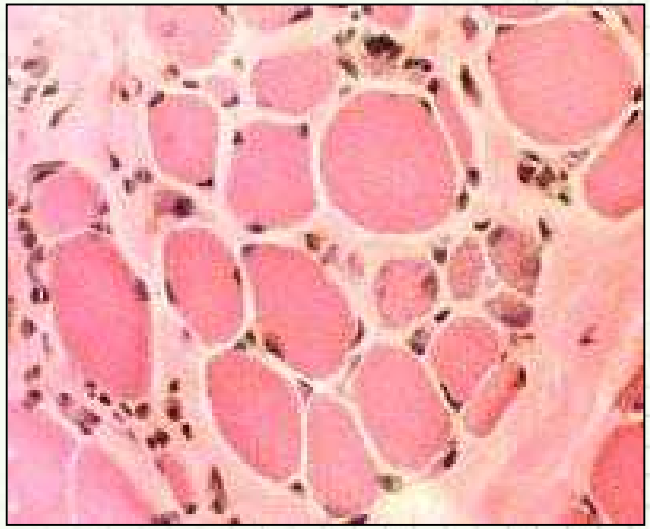

波幅低、多相短棘波

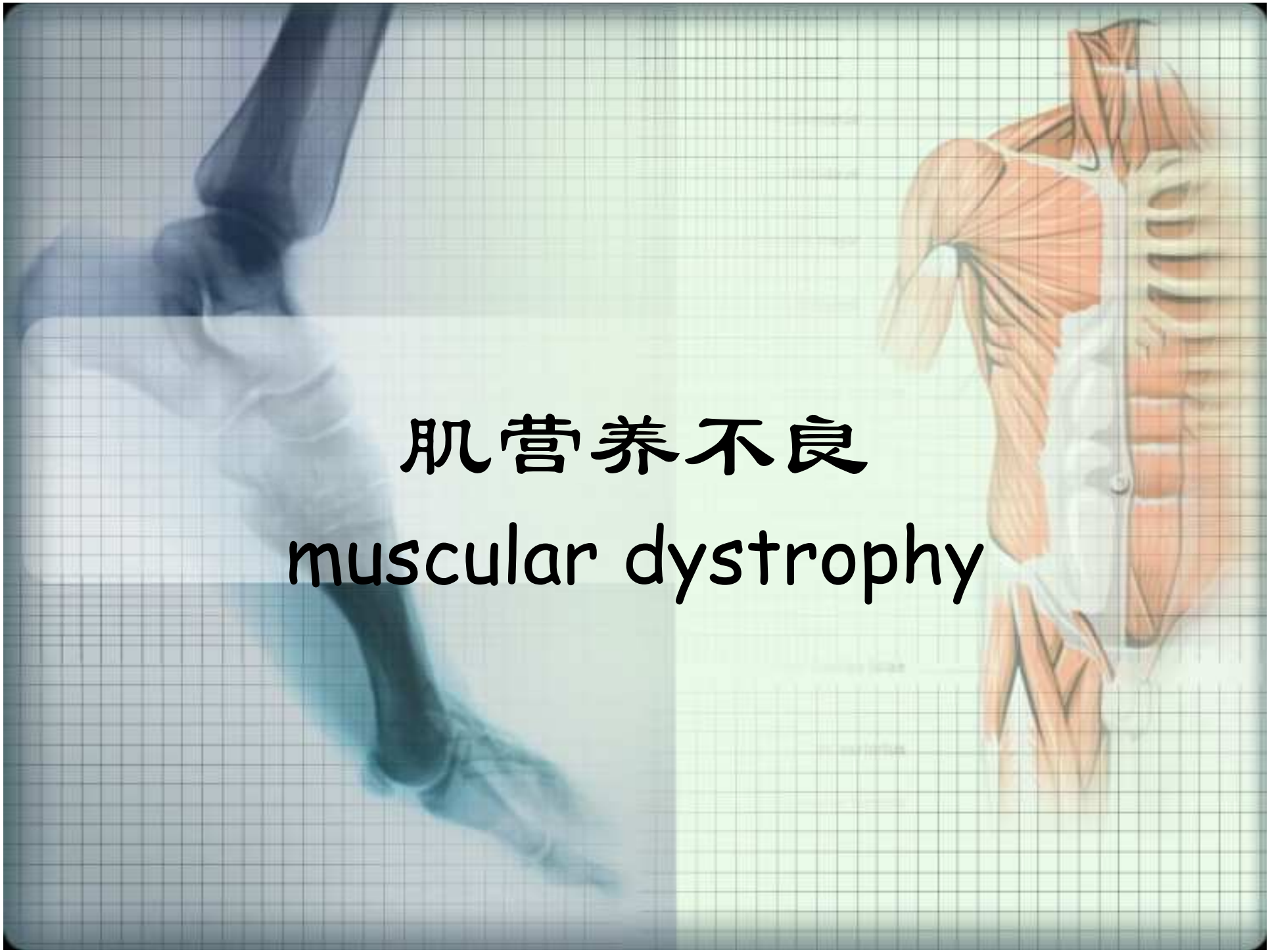
纤颤波、正尖波

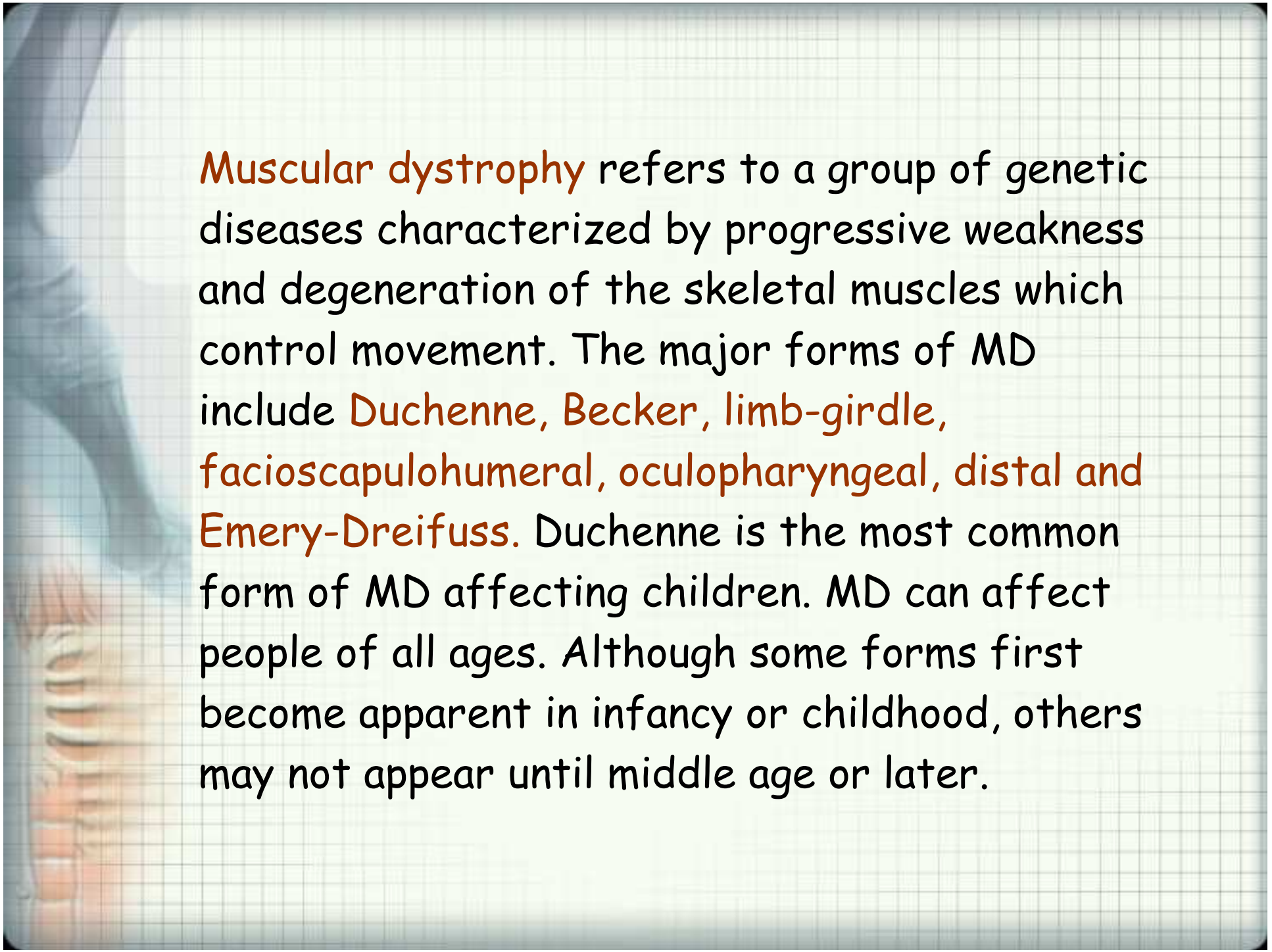
## 其他实验室检查

血清CPK（CK）高

肌肉活检（fiber size、connective tissue）



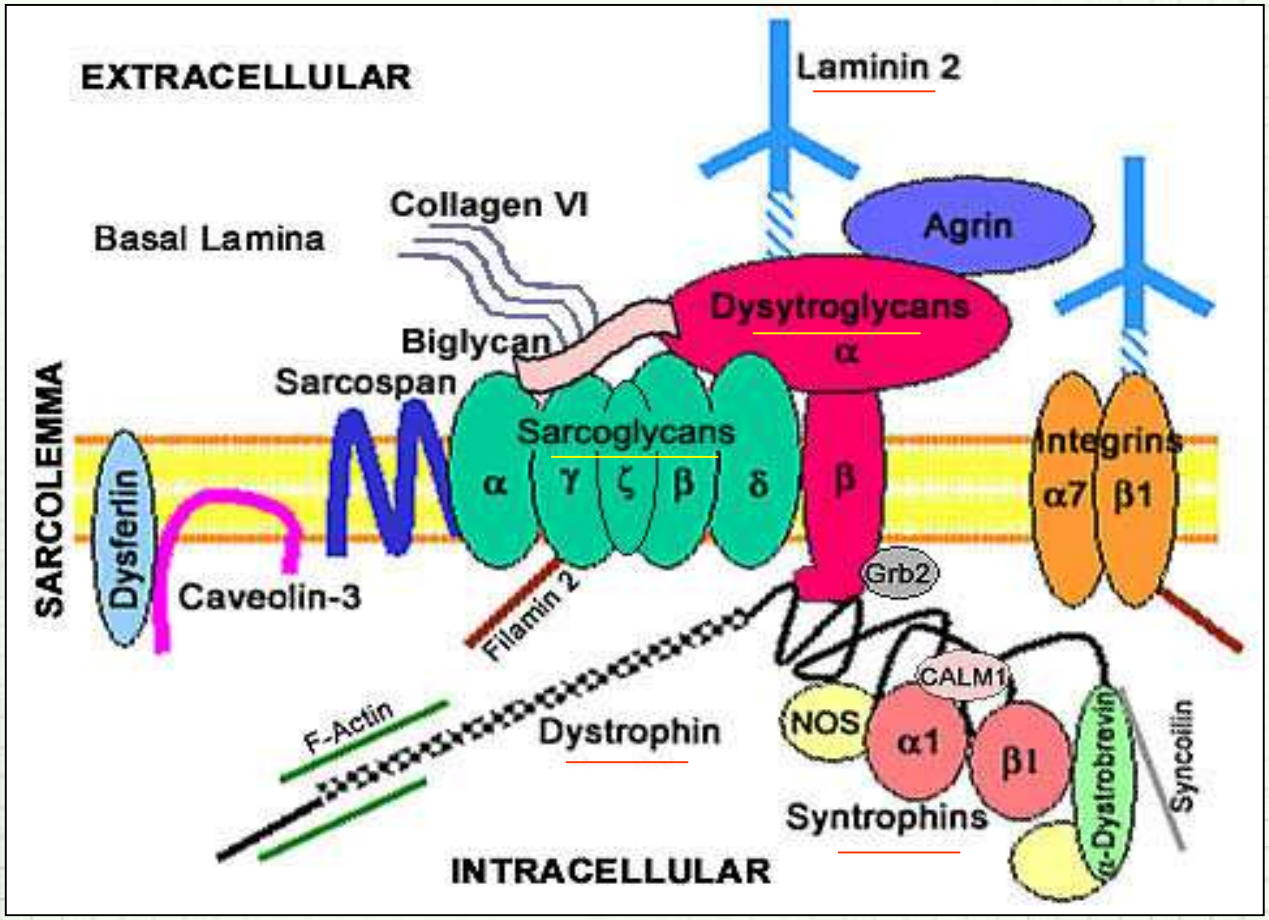

肌营养不良
muscular dystrophy



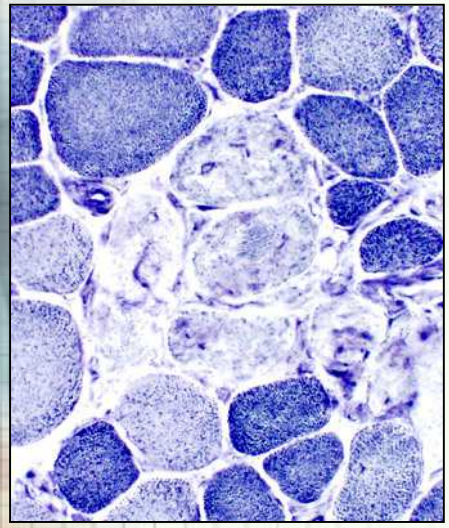
Muscular dystrophy refers to a group of genetic diseases characterized by progressive weakness and degeneration of the skeletal muscles which control movement. The major forms of MD include Duchenne, Becker, limb-girdle, facioscapulohumeral, oculopharyngeal, distal and Emery-Dreifuss. Duchenne is the most common form of MD affecting children. MD can affect people of all ages. Although some forms first become apparent in infancy or childhood, others may not appear until middle age or later.

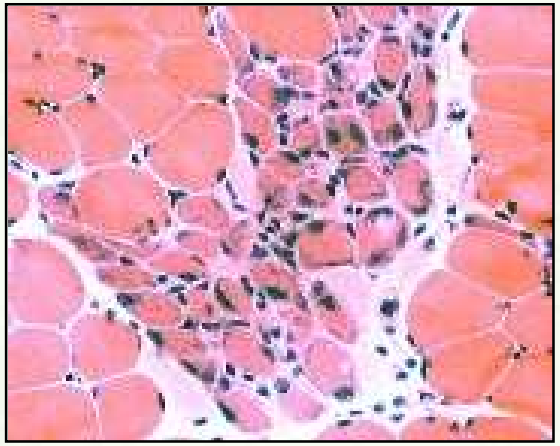
# 病因

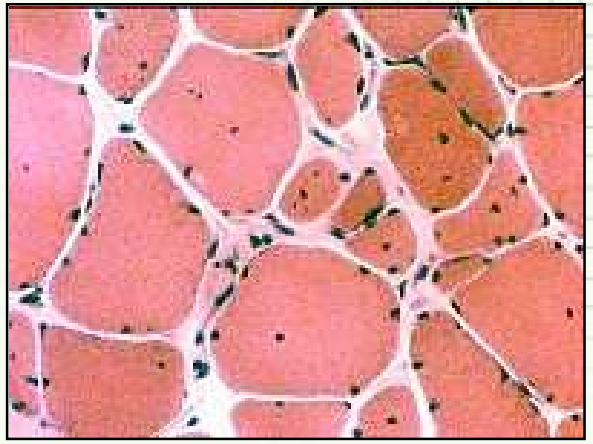
遗传致骨架蛋白异常



# 病理

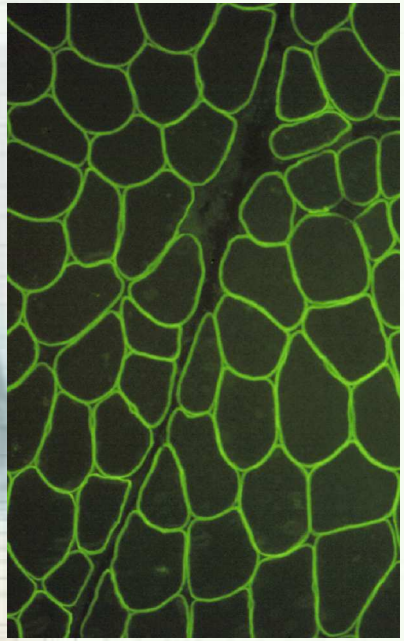




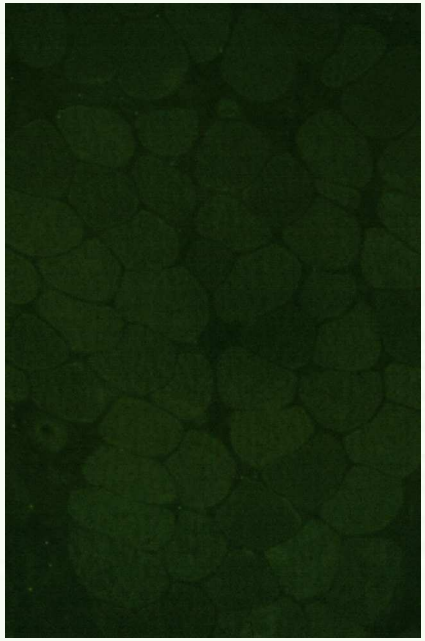


坏死、再生、大小不均、核内移等（NADH、HE）

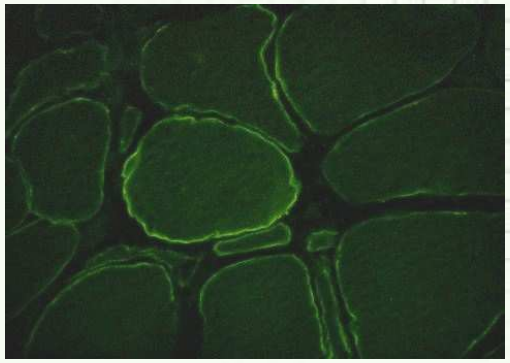
# Dystrophin免疫荧光染色



normal



DMD



BMD

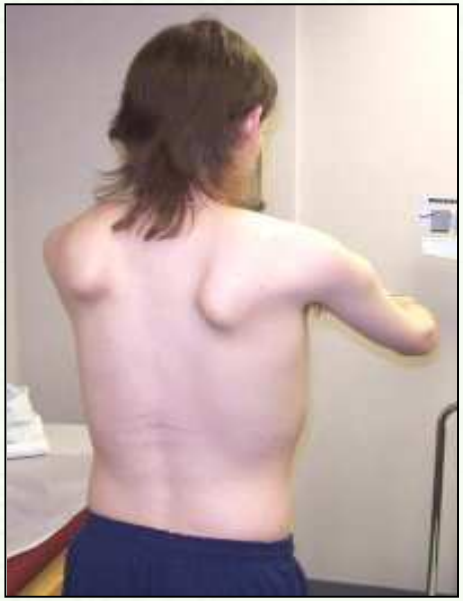
# 临床表现

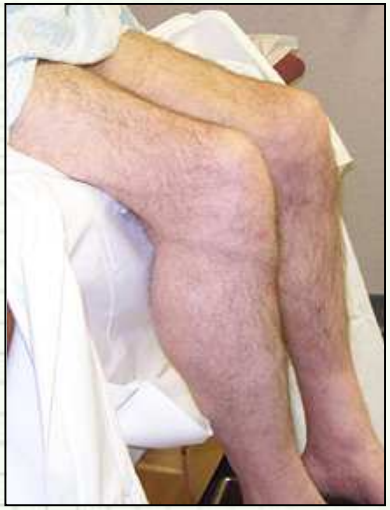
## 分型

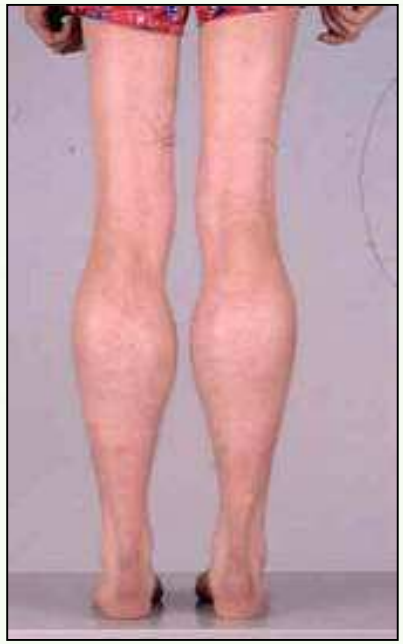
- 假肥大型（X-link隐性遗传）
  - DMD（Duchenne Muscular Dystrophy）
  - BMD（Becker Muscular Dystrophy）
- 肢带型肌营养不良（常显、常隐）
- 面肩肱型肌营养不良（常显）
- 艾德肌营养不良
- 其他肌营养不良（眼肌、远端）

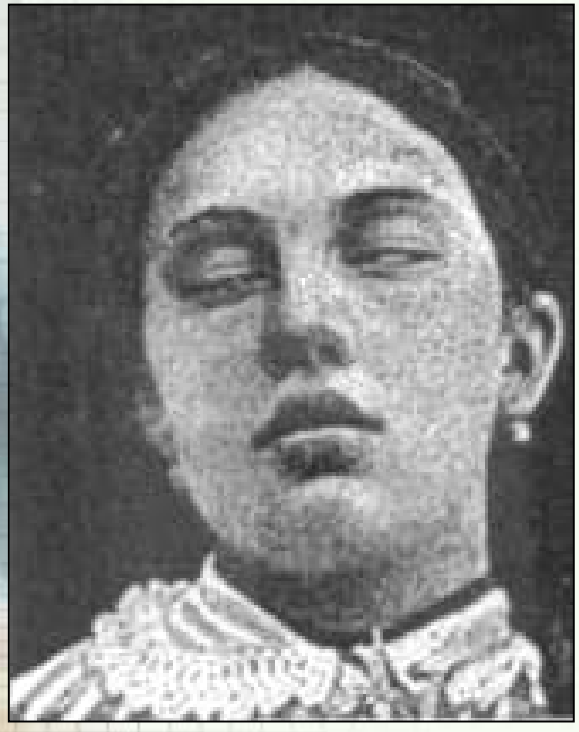
## 特征性症状和体征

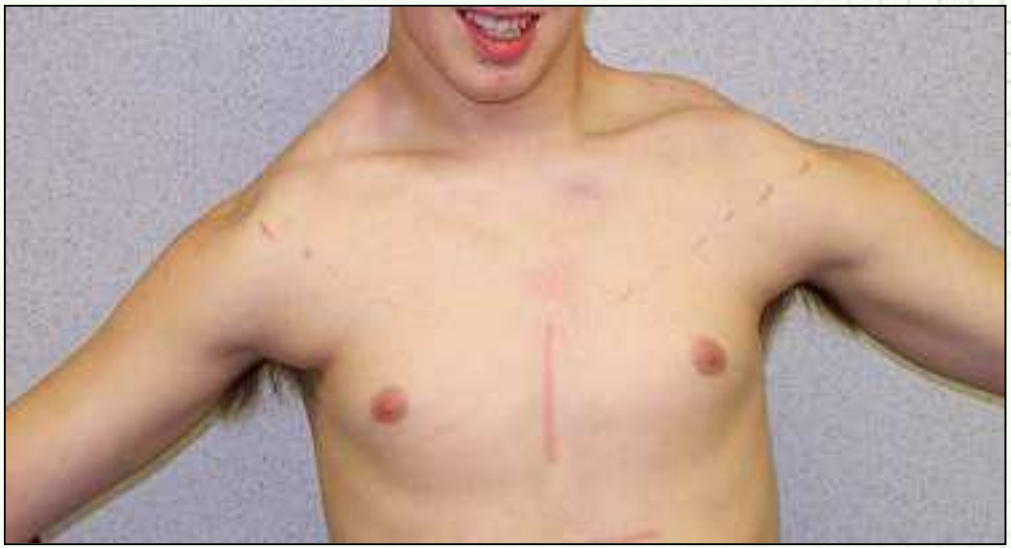
- 假肥大型
  - 鸭步
  - Gowers现象
  - 翼状肩
  - 假肥大
  - 足跟着地困难
- 肢带型
- 面肩肱型
  - 蝠翼、衣架





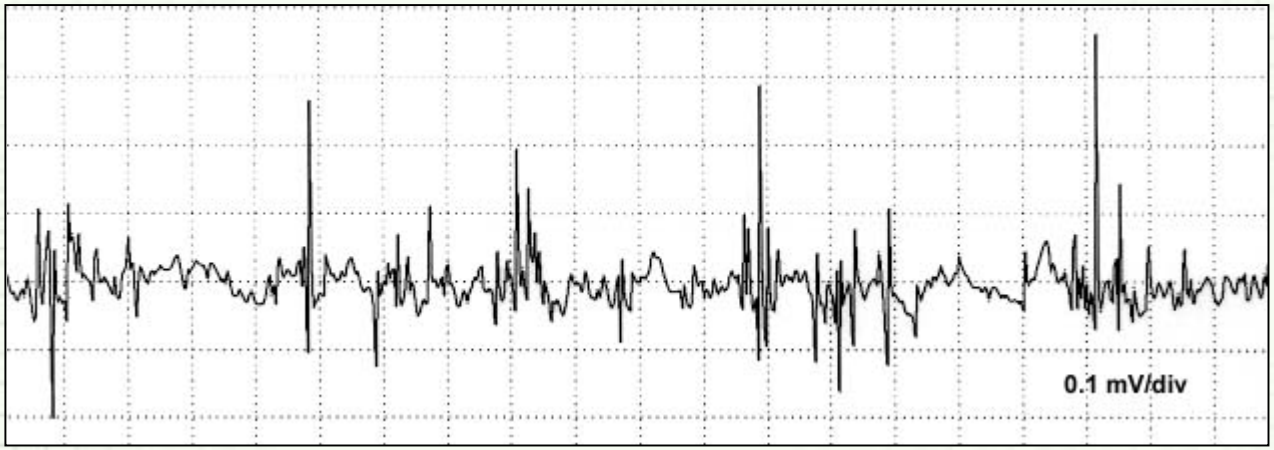


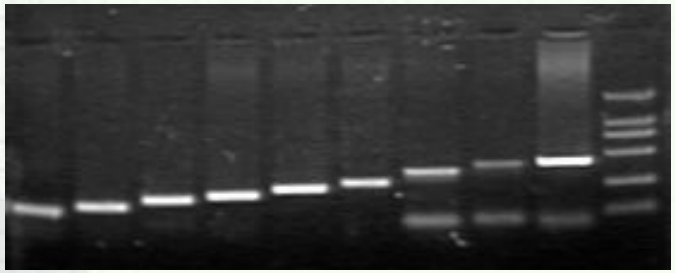


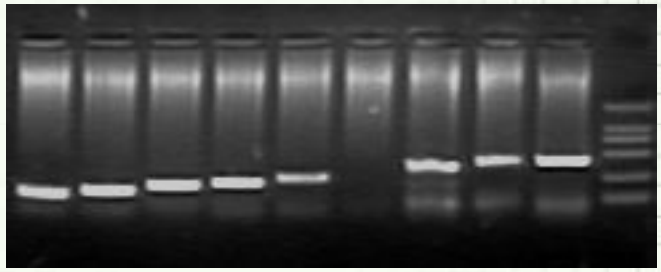


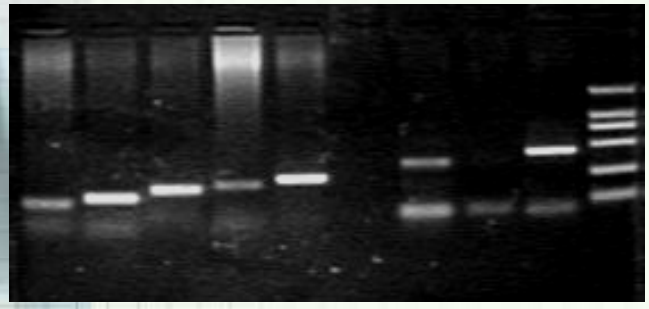
# 实验室检查

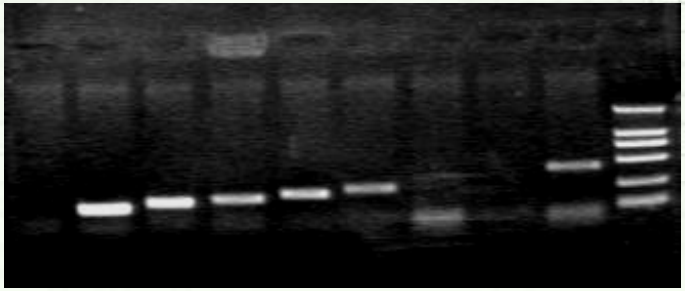
- CPK、LDH、ALT、AST升高（DMD、BMD）
- 肌电图示肌源性损害









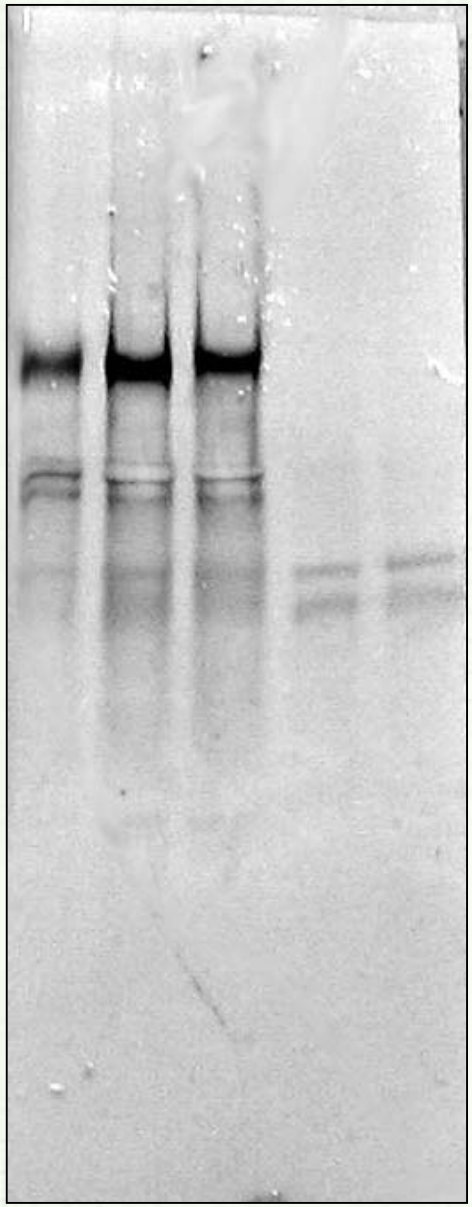


基因检测（Chamberlian多重PCR，9对引物分别）

只能检测基因缺失（2/3，另1/3点突变多为研究检测）

1 2 3 4 5

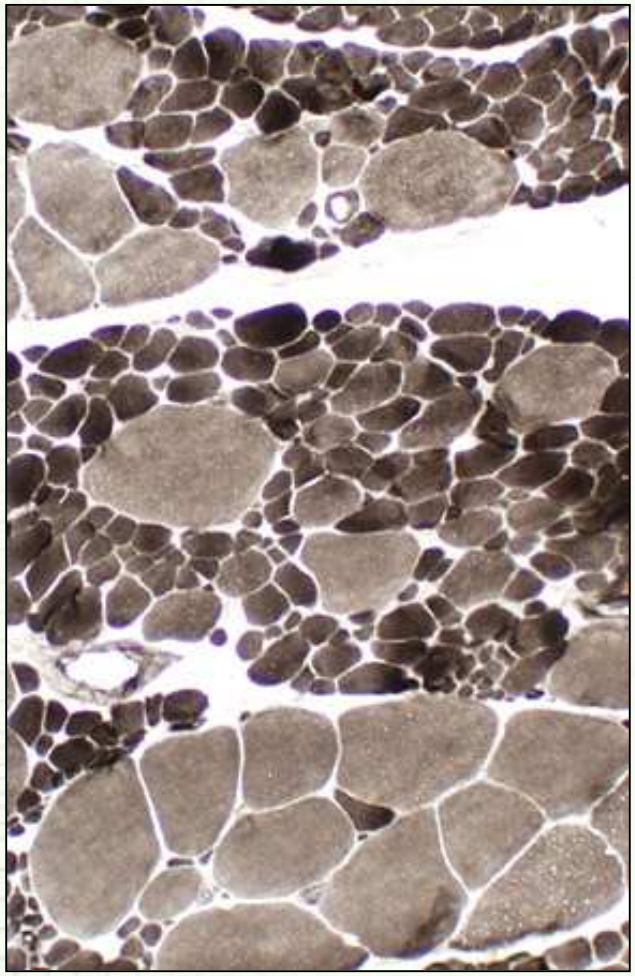
dystrophin



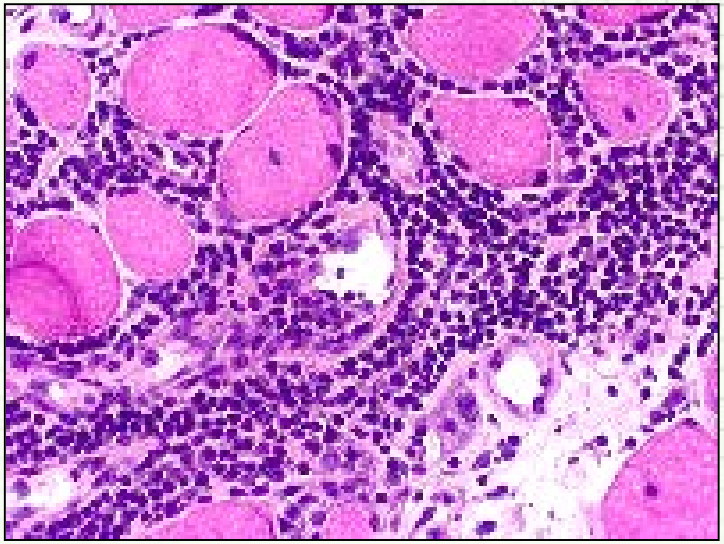
1 myositis
2 LGMD
3 normal
4 DMD
5 DMD

# 诊断和鉴别诊断

- 年龄、性别
- 病程
- 无力分布
- 肌酶和肌电图
- 基因、病理（组织病理、免疫组化）
  - 运动神经元病
  - 多发性肌炎



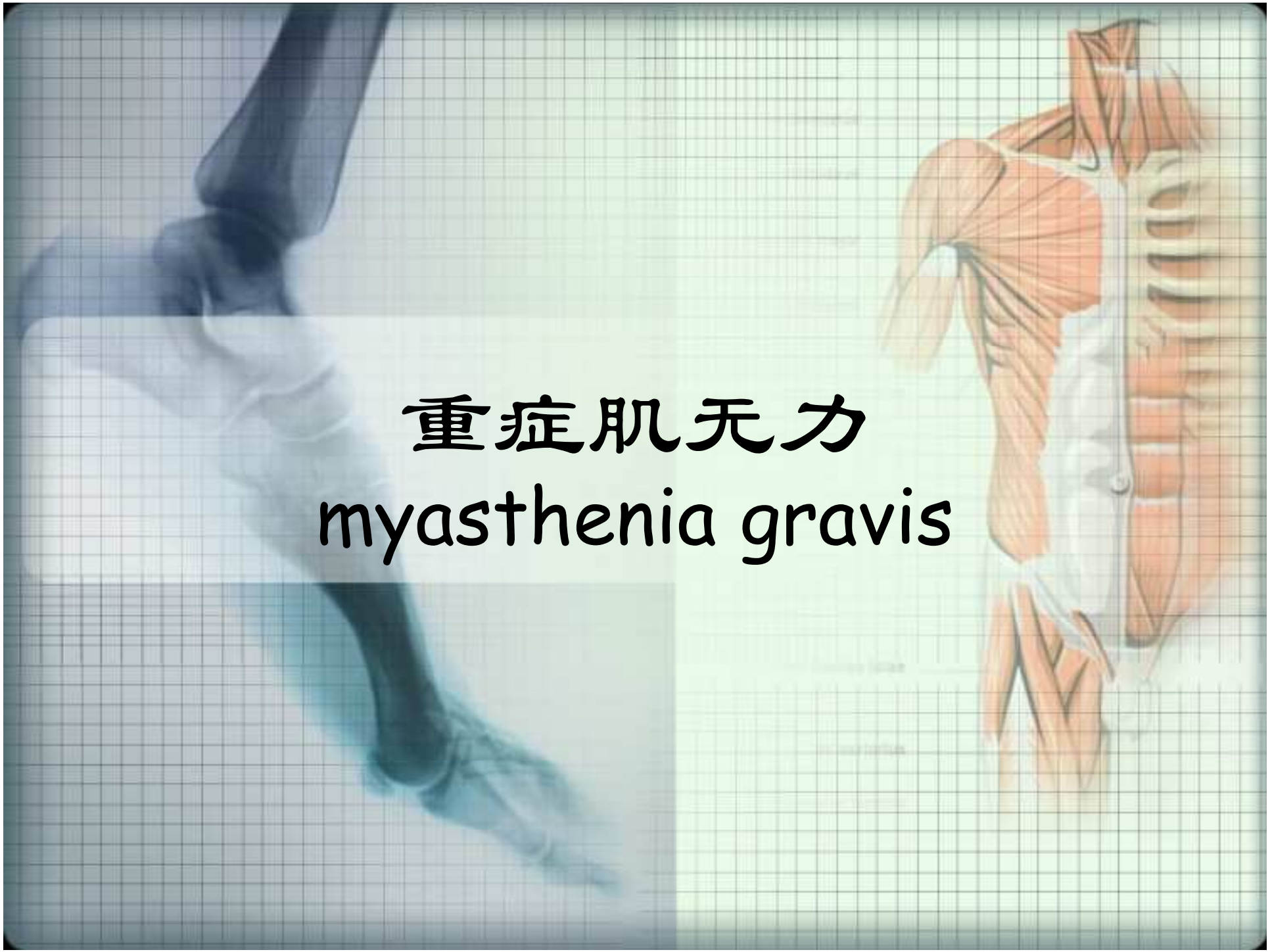
运动神经元病SMA



多肌炎

# 治疗

- 以支持治疗为主
- 治疗进展和展望
  - 强的松（DMD临床研究小组1.5mg/kg/d、Brooke 0.75mg/kg/d、Dubowitz假日疗法）
  - 成肌细胞移植（Dys缺陷鼠）
  - 基因治疗（基因大）
  - 氨基糖甙类药物治疗（影响终止密码子）
  - 干细胞治疗
  - 上调Utrophin（同源蛋白）


重症肌无力
myasthenia gravis

Myasthenia gravis is a chronic autoimmune disease characterized by weakness of the skeletal muscles. The hallmark of myasthenia gravis is muscle weakness that increases during periods of activity and improves after periods of rest. Muscles that control eye and eyelid movements, facial expression, chewing, talking, and swallowing are often, but not always, involved. The muscles that control breathing and neck and limb movements may also be affected. Myasthenia gravis is caused by a defect in the transmission of nerve impulses to muscles. In myasthenia gravis, antibodies produced by the body's own immune system block, alter, or destroy the receptors for acetylcholine.

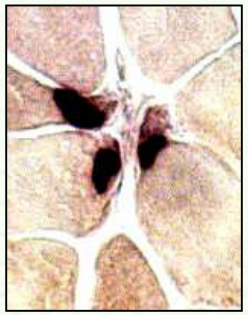
# 病因与发病机制

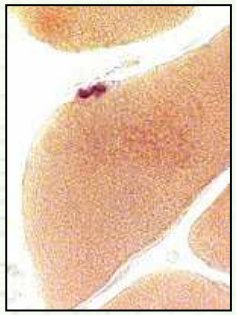
## 自身免疫性疾病

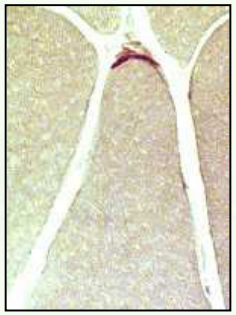
- anti-AChR antibody is found in approximately 80-90% of patients with MG
- presence of associated autoimmune disorders in patients suffering from MG
- infants born to myasthenic mothers can develop a transient myasthenia-like syndrome.
- Patients with MG will have a therapeutic response to various immunomodulating therapies
- active transfer experiments
- passive transfer experiments

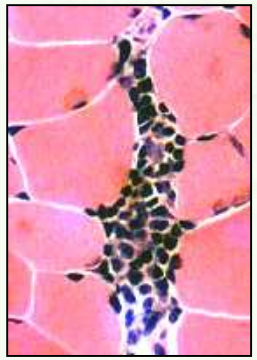
MG can be considered a B cell mediated disease, as antibodies (a B cell product) against AChR are responsible for the disease. However, the importance of T cells in pathogenesis of MG is becoming increasingly apparent. The thymus is the central organ in T cell mediated immunity, and thymic abnormalities such as thymic hyperplasia or thymoma are well recognized in myasthenic patients (90%)

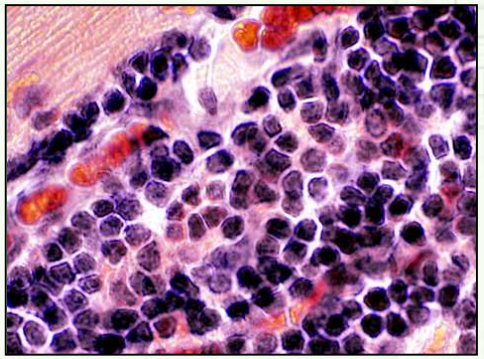
# 病理











# 临床表现

- 发病年龄（女20-30岁、男40-50岁、儿童）
- 波动（晨轻暮重、疲劳加重、休息缓解）
- 累及骨骼肌（颅神经支配肌、脊神经支配肌）

## 临床分型

- 单纯眼肌型
- 延髓肌型
- 全身肌无力型
- 脊髓肌无力型（对称、近端）
- 肌萎缩型

儿童重症肌无力、新生儿重症肌无力

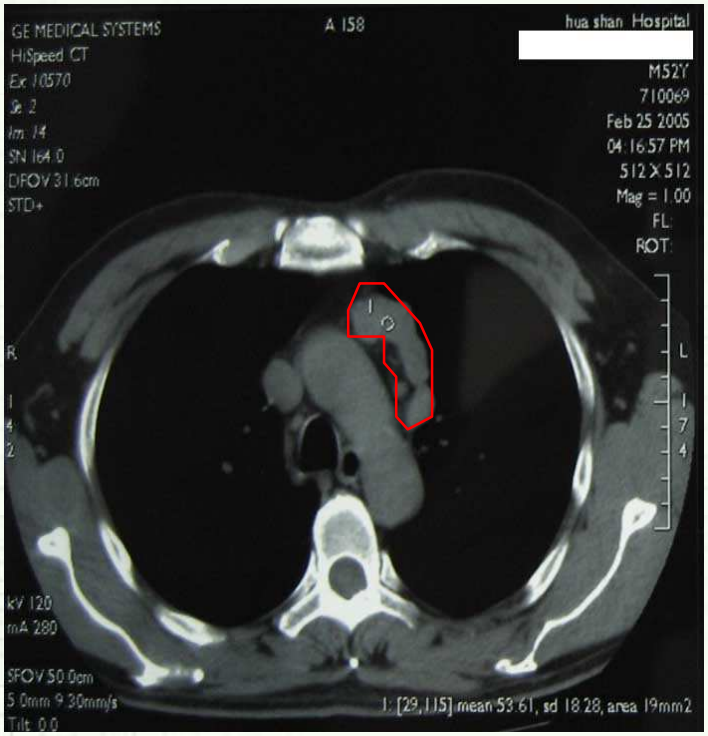
- 改良Osserman分型
  - Ⅰ型、Ⅱa型、Ⅱb型、Ⅲ型、Ⅳ型
- 美国重症肌无力基金会（MGFA）分型
  - Ⅰ、Ⅱa、Ⅱb、Ⅲa、Ⅲb、Ⅳa、Ⅳb、Ⅴ
- 国内常用分型
  - 单纯眼肌、延髓肌型、脊髓肌型、全身型、肌萎缩型

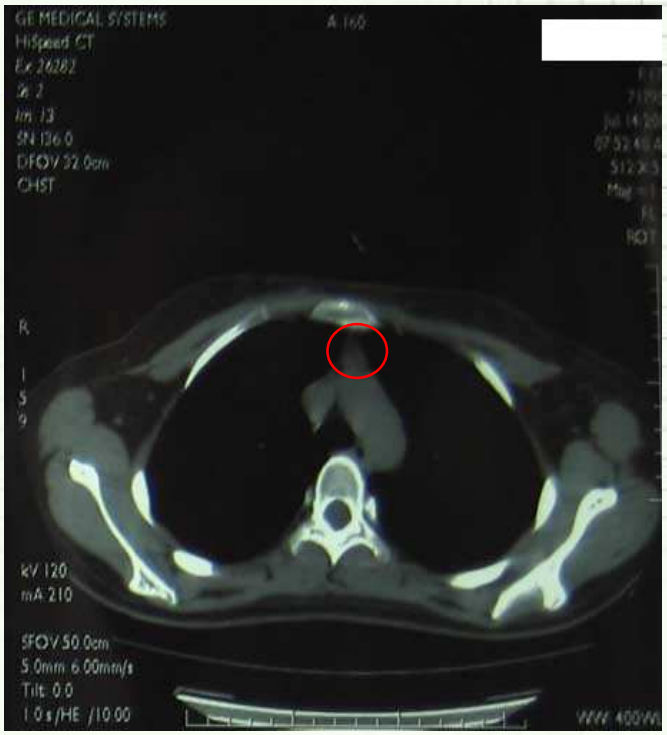
危象（呼吸肌无力、麻痹不能维持正常通气功能的危急状况）

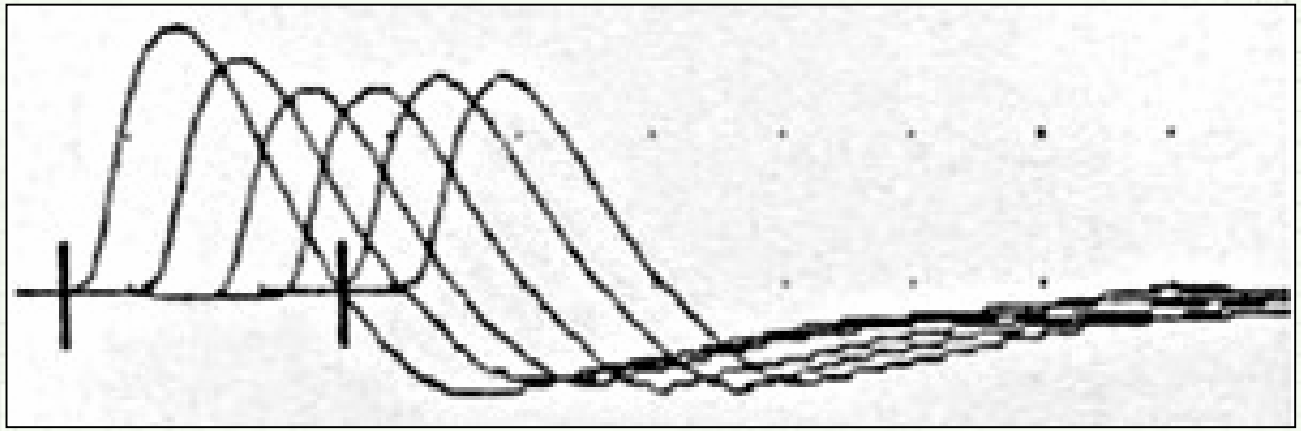
- 肌无力危象（疾病发展、感染、药物使用不当）
- 胆碱能危象(目前以免疫治疗为主，少见)
- 反拗性危象

# 实验室检查

- 70%-80%病人AChR抗体阳性，眼肌型阳性率低
- 部分SNMG中可测到MuSK抗体
- 合并甲亢者可有T3、T4增高
- 胸腺CT示胸腺增生（60%-75%）或胸腺瘤(10%-15% )
- RNS低频重复电刺激（3Hz/秒），电位衰减10%以上


GE MEDICAL SYSTEMS
A 158
hua shan Hospital
HiSpeed CT
Ex 10570
Se 2
Im 14
SN 164 0
DFOV 31 6cm
STD+
M52Y
710069
Feb 25 2005
04 16 57 PM
512 X 512
Mag = 1 00
FL
ROT
R
L
kV 120
mA 280
SFOV 50 0cm
5 0mm 9 30mm/s
Tilt 0 0
1: [28, 115] mean 53 61, sd 18 28, area 19mm2


GE MEDICAL SYSTEMS
A 160
HiSpeed CT
Ex 26282
Se 2
Im 13
SN 136 0
DFOV 32 0cm
CHST
R
kV 120
mA 210
SFOV 50 0cm
5 0mm 6 00mm/s
Tilt 0 0
1 0 s/HE /10 00



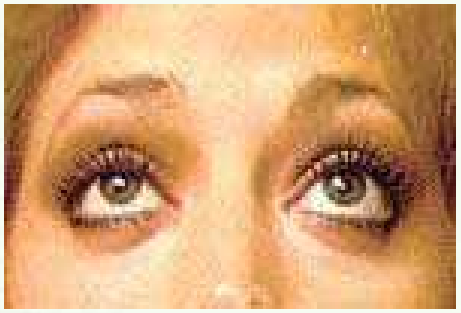
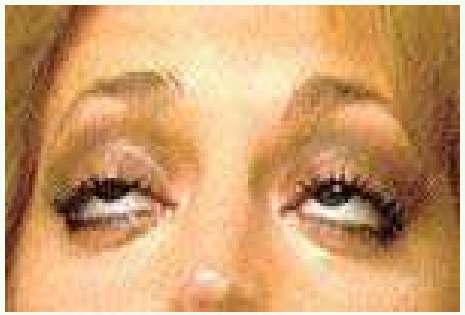
低频（3Hz）

CMAP 波幅递减达10%以上

全身型75%眼肌型50%

# 诊断

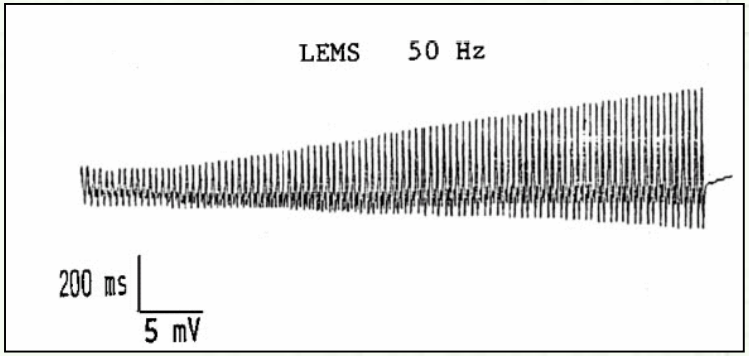
- 疲劳试验

- 新斯的明试验
- 重复电刺激

# 鉴别诊断

- 慢性进行性眼外肌麻痹
- 运动神经元病（延髓肌麻痹）
- 急性感染性多发性神经病
- 肌无力综合征




高频刺激波幅增加100%以上

# 治疗

- 抗胆碱酯酶药物
- 免疫抑制或调节
  - 激素
  - 硫唑嘌呤
  - 环磷酰胺
  - 血浆交换
  - 静脉丙球
- 胸腺切除
- 危象处理