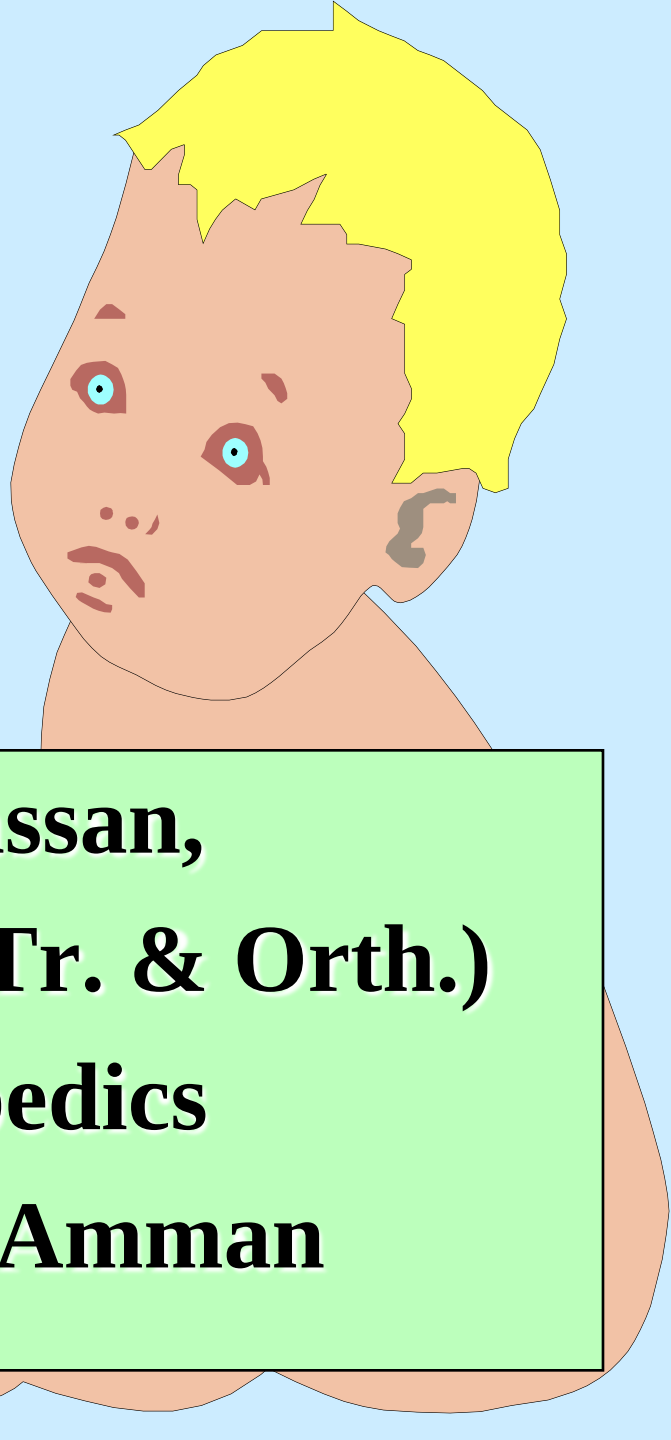
Muscle Dystrophy



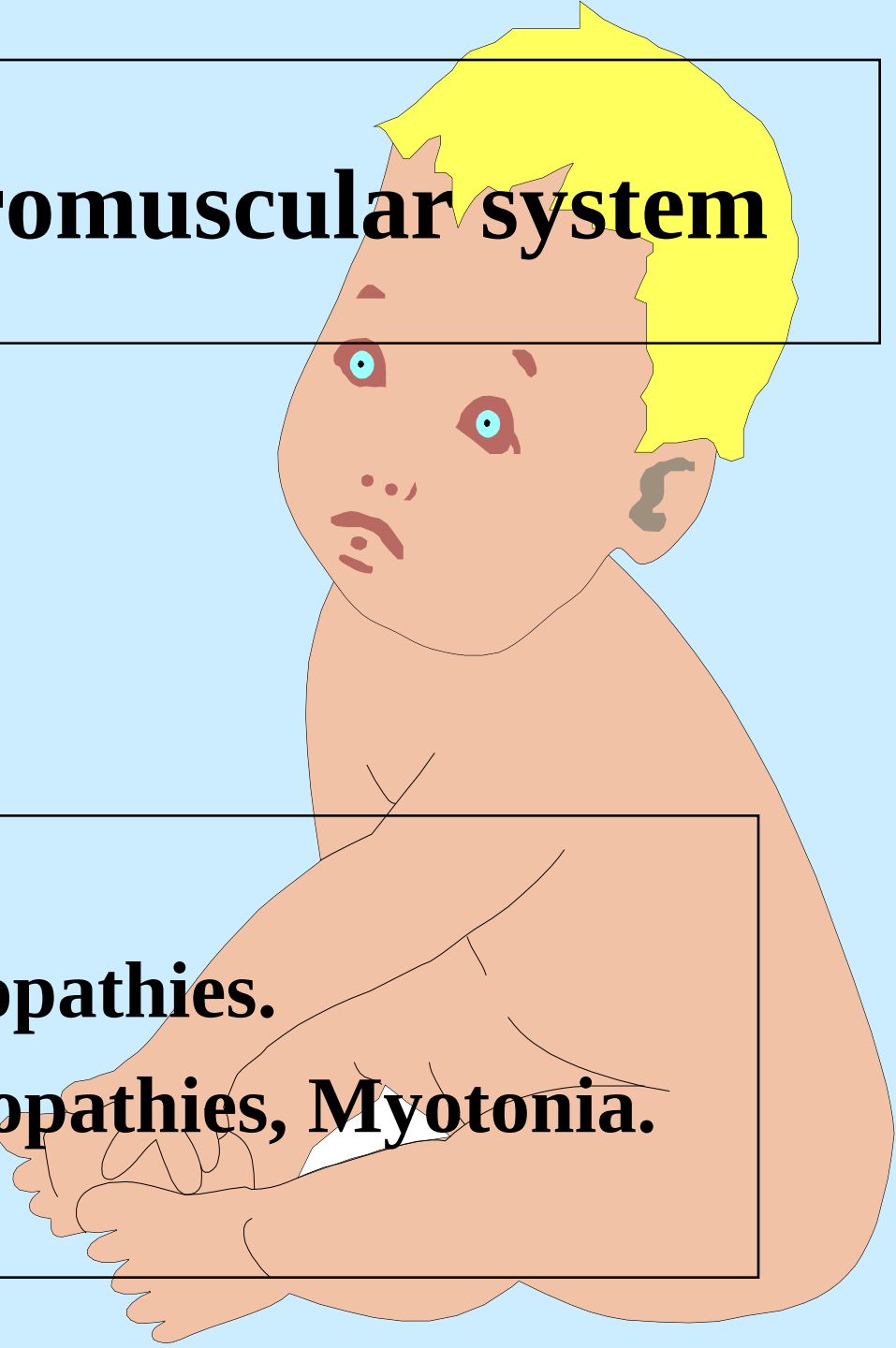
Freih Odeh Abu Hassan,

F.R.C.S. (Eng.), F.R.C.S. (Tr. & Orth.)

Professor of Orthopedics

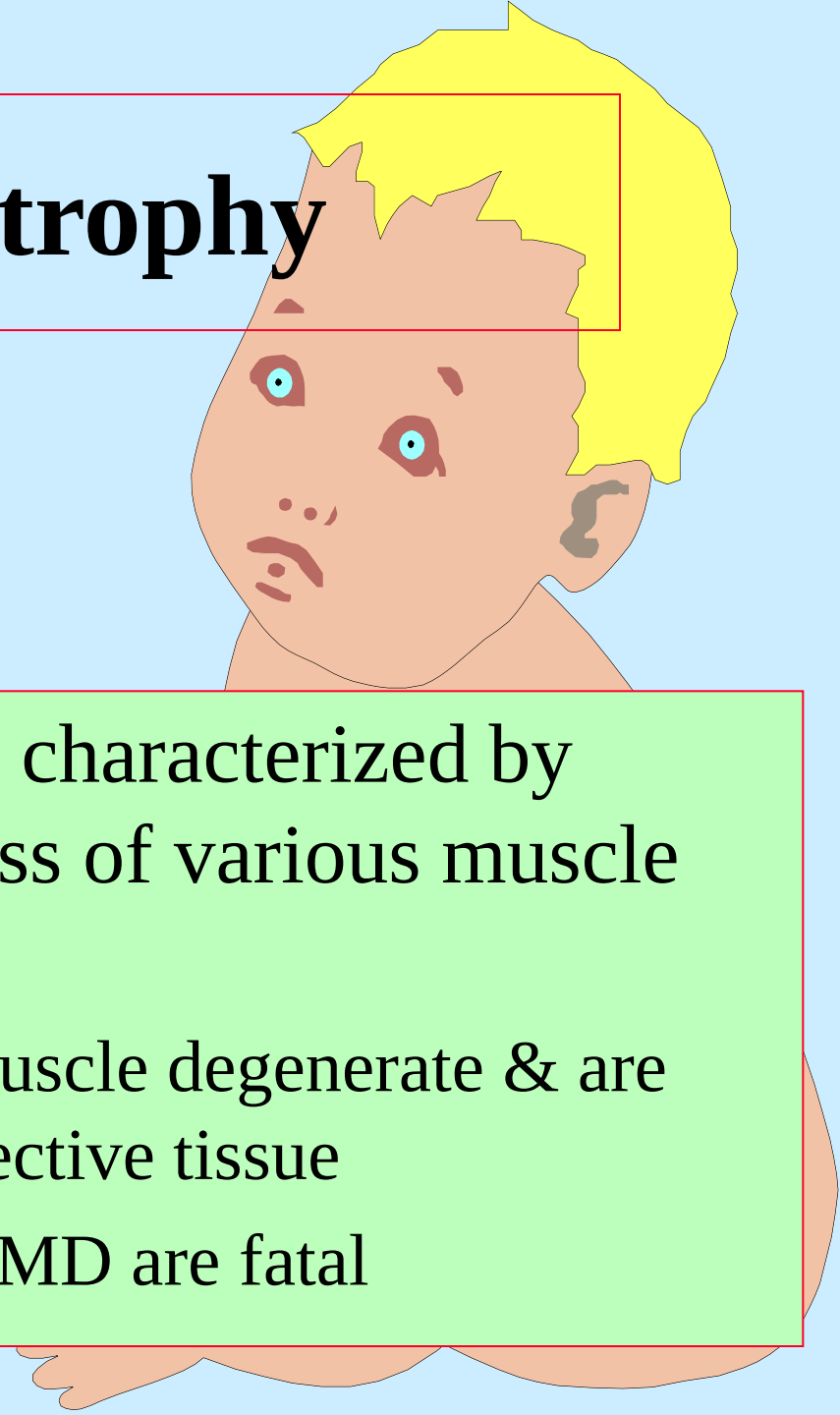
University of Jordan - Amman

Disorders of Neuromuscular system



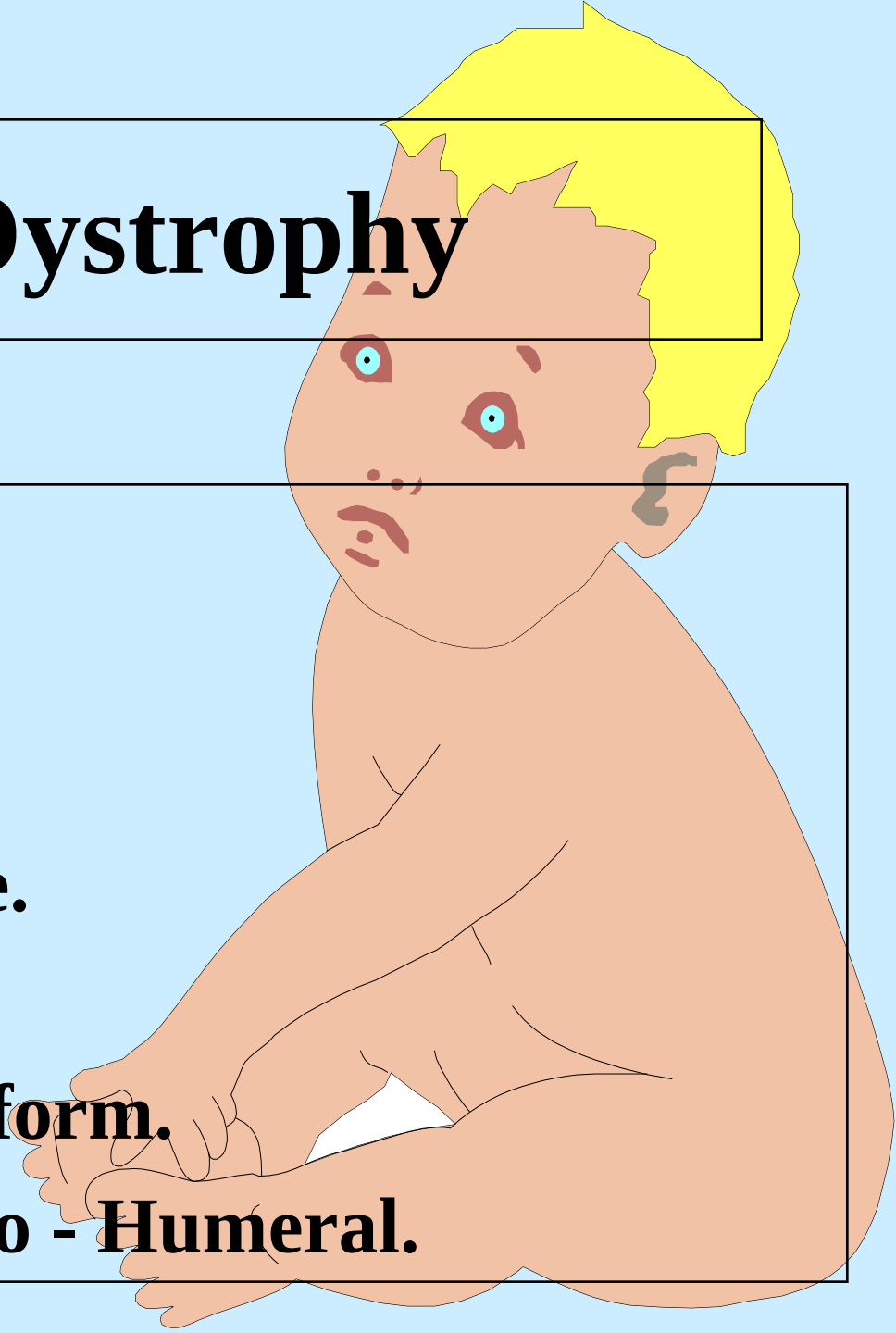
- AHC : SMA, Polio.
- Nerve fiber : Neuropathies.
- Muscle : M.D., Myopathies, Myotonia.

Muscular Dystrophy



- Group of inherited diseases characterized by progressive,diffuse weakness of various muscle groups
  - Muscle cells w/in belly of muscle degenerate & are replaced by adipose & connective tissue
  - Secondary complications to MD are fatal

Muscle Dystrophy



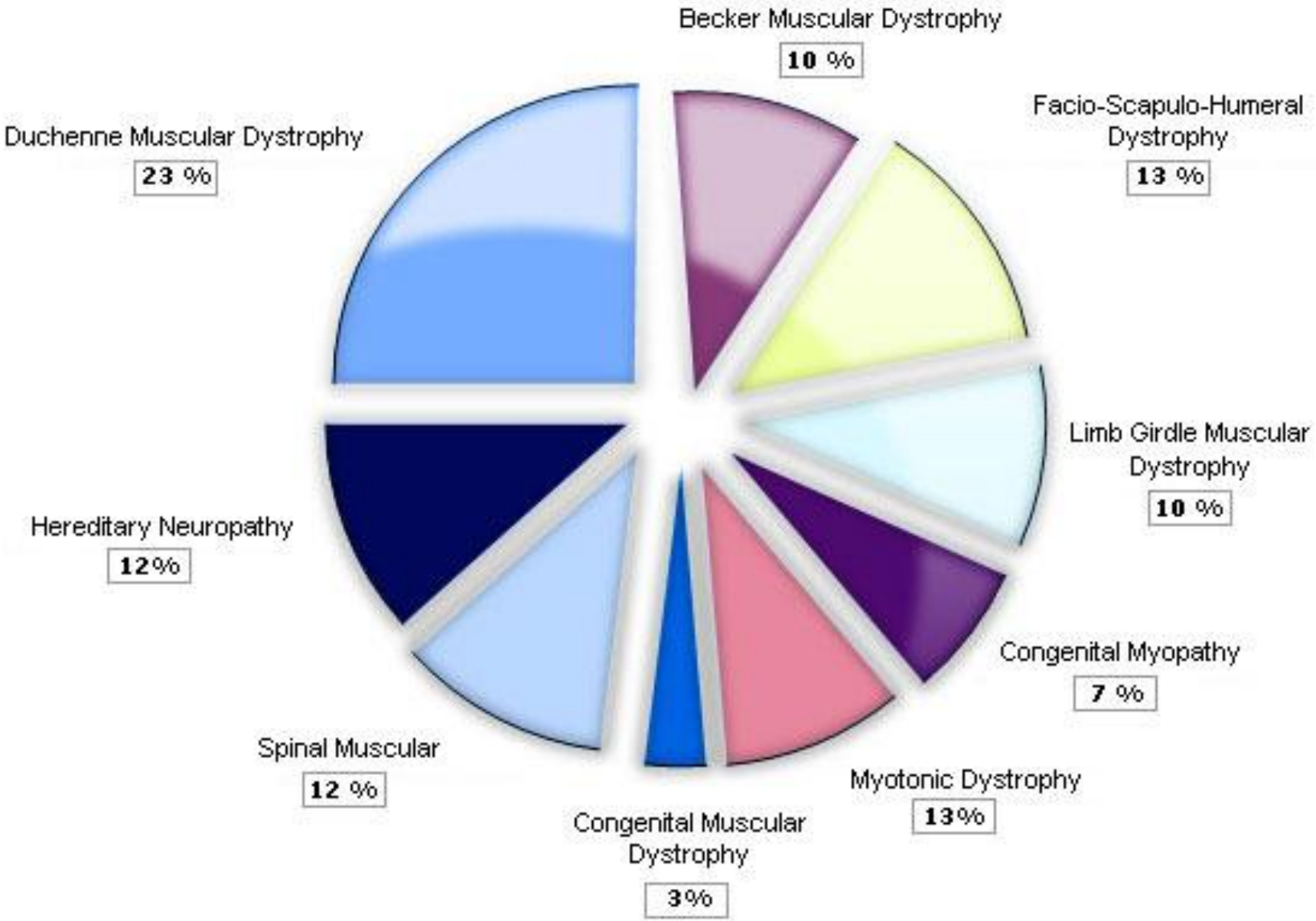
X-Linked Recessive

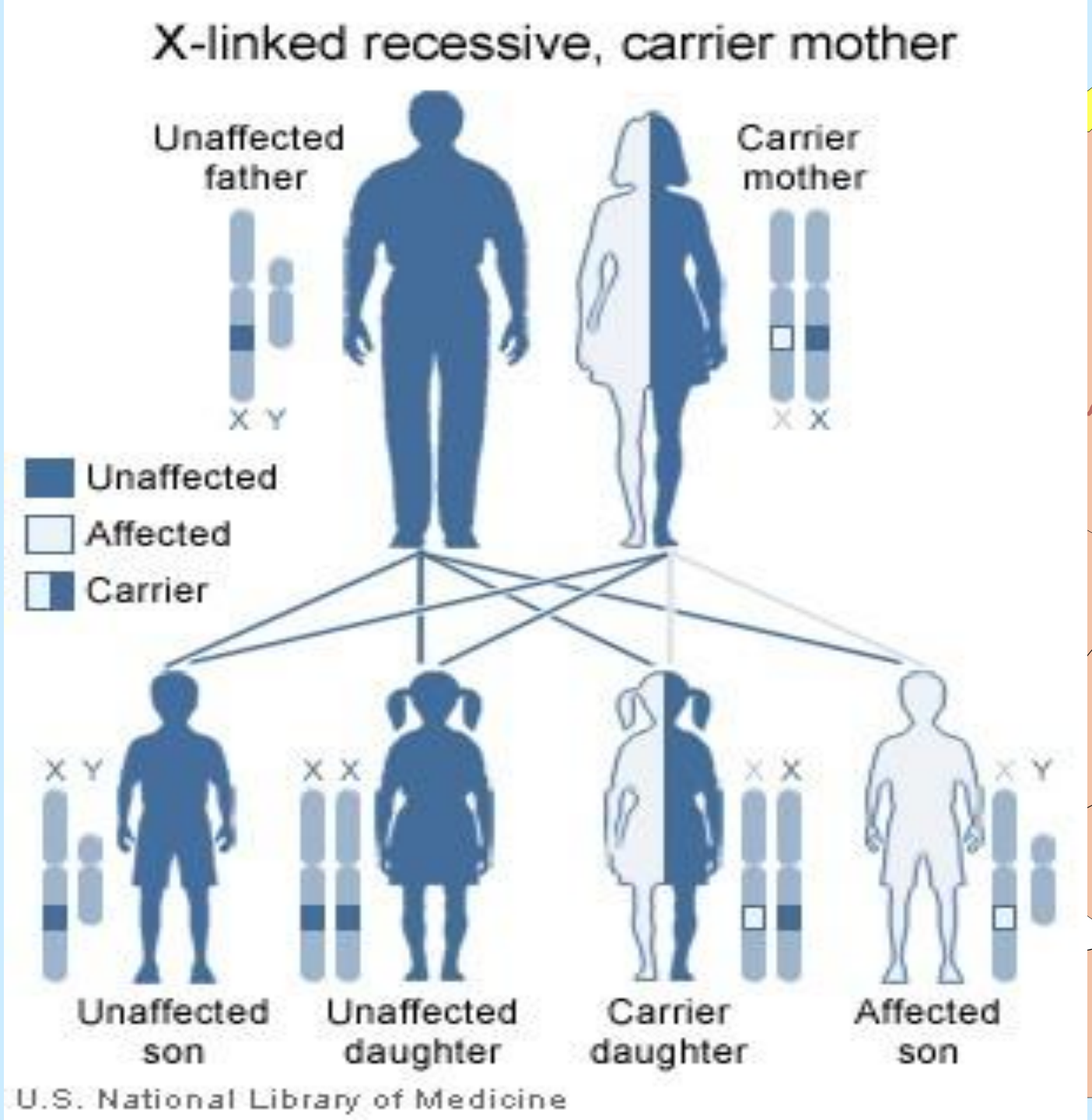
- **Duchenne,**
- **Becker.**

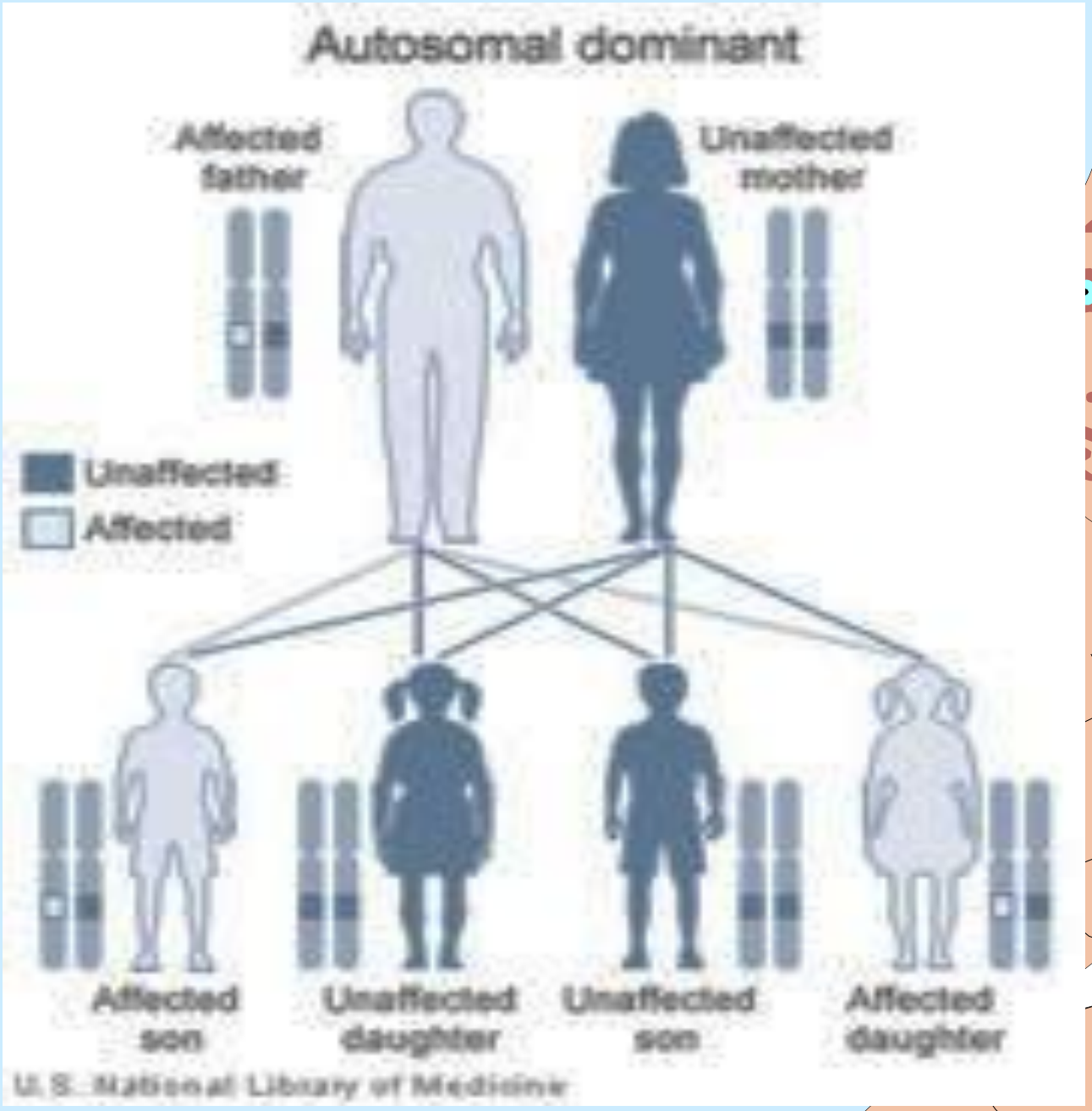
A.R.

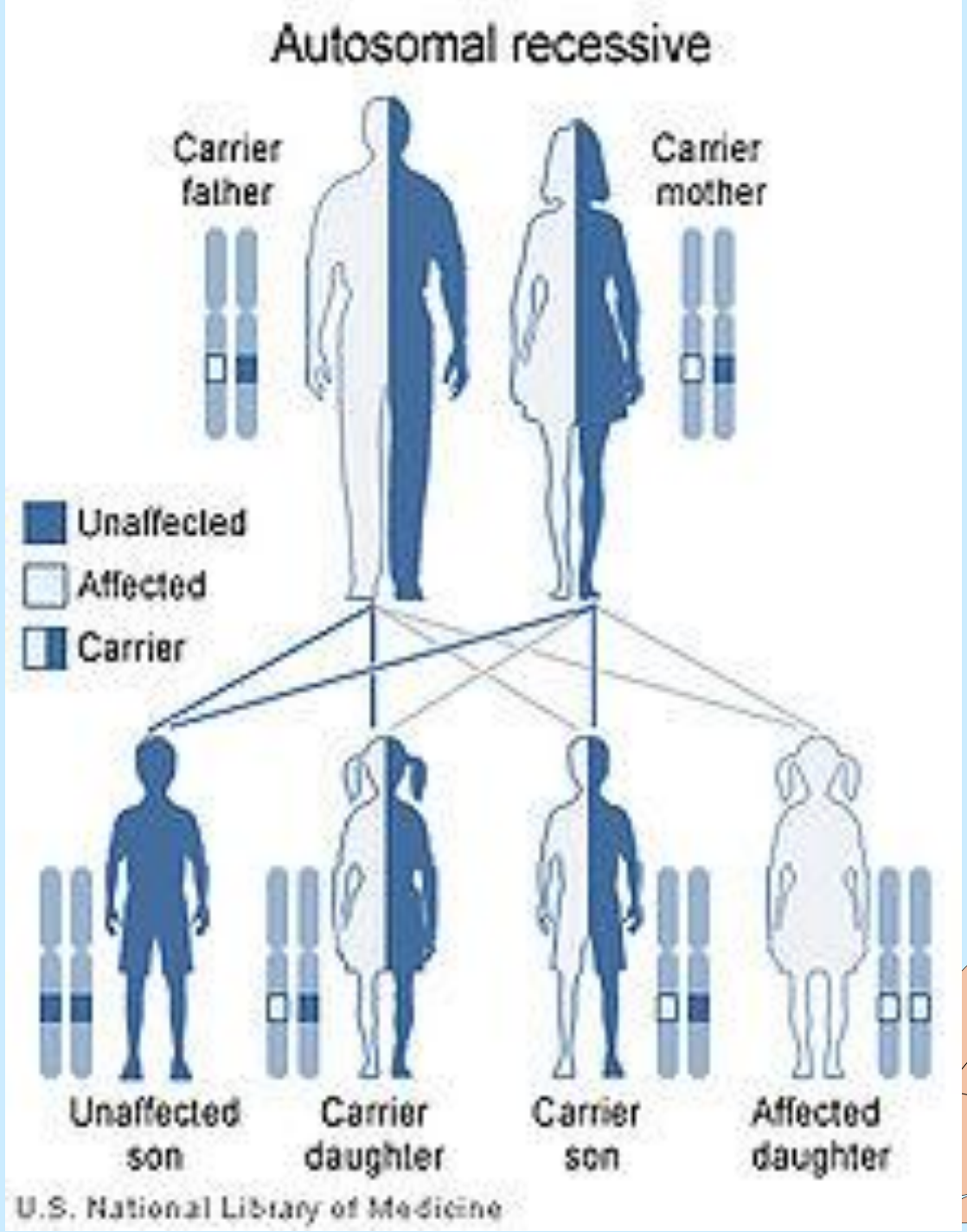
- **Limb girdle.**
- **Congenital.**
- **Childhood form.**

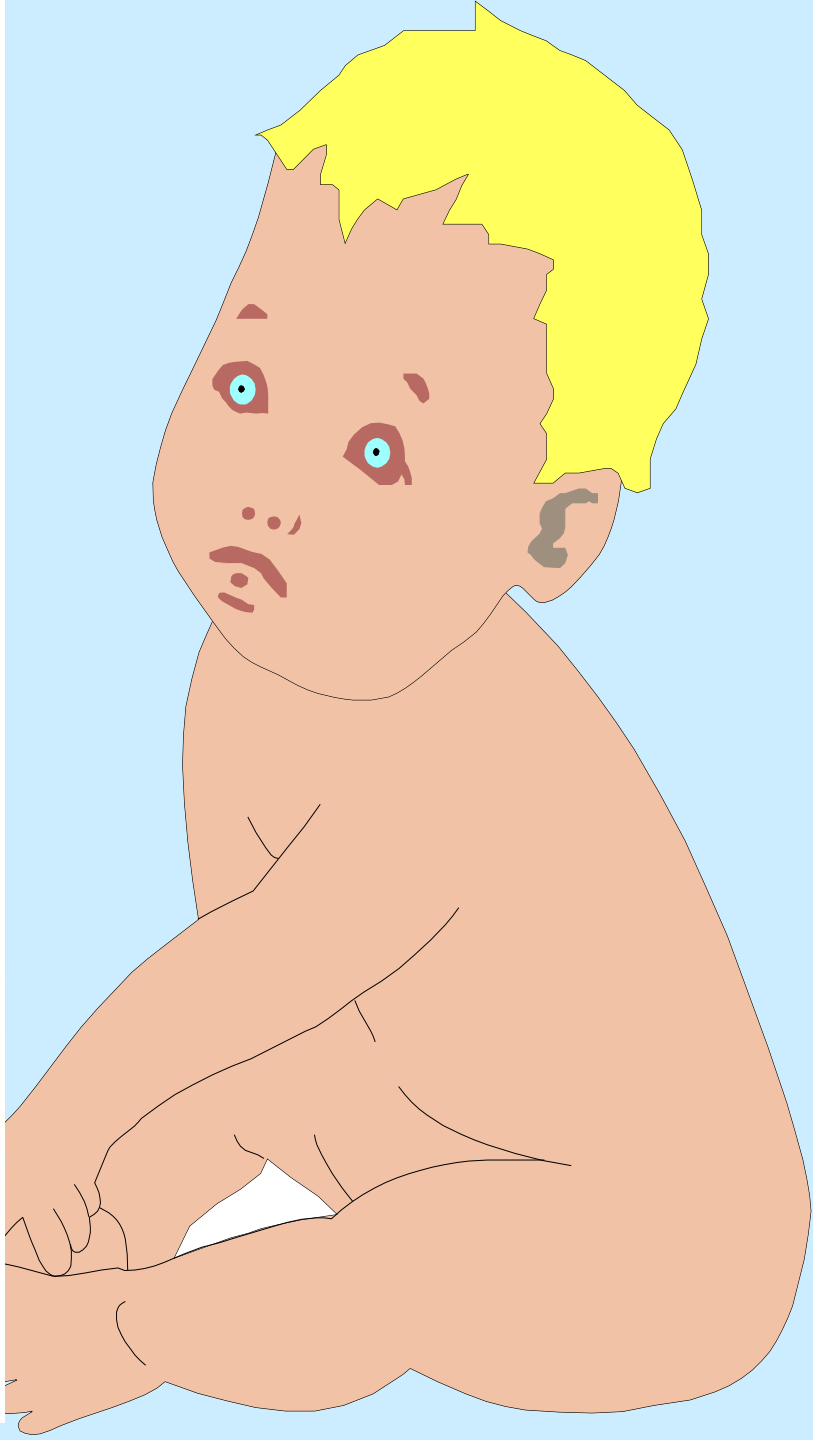
A.D. **Fascio - Scapulo - Humeral.**

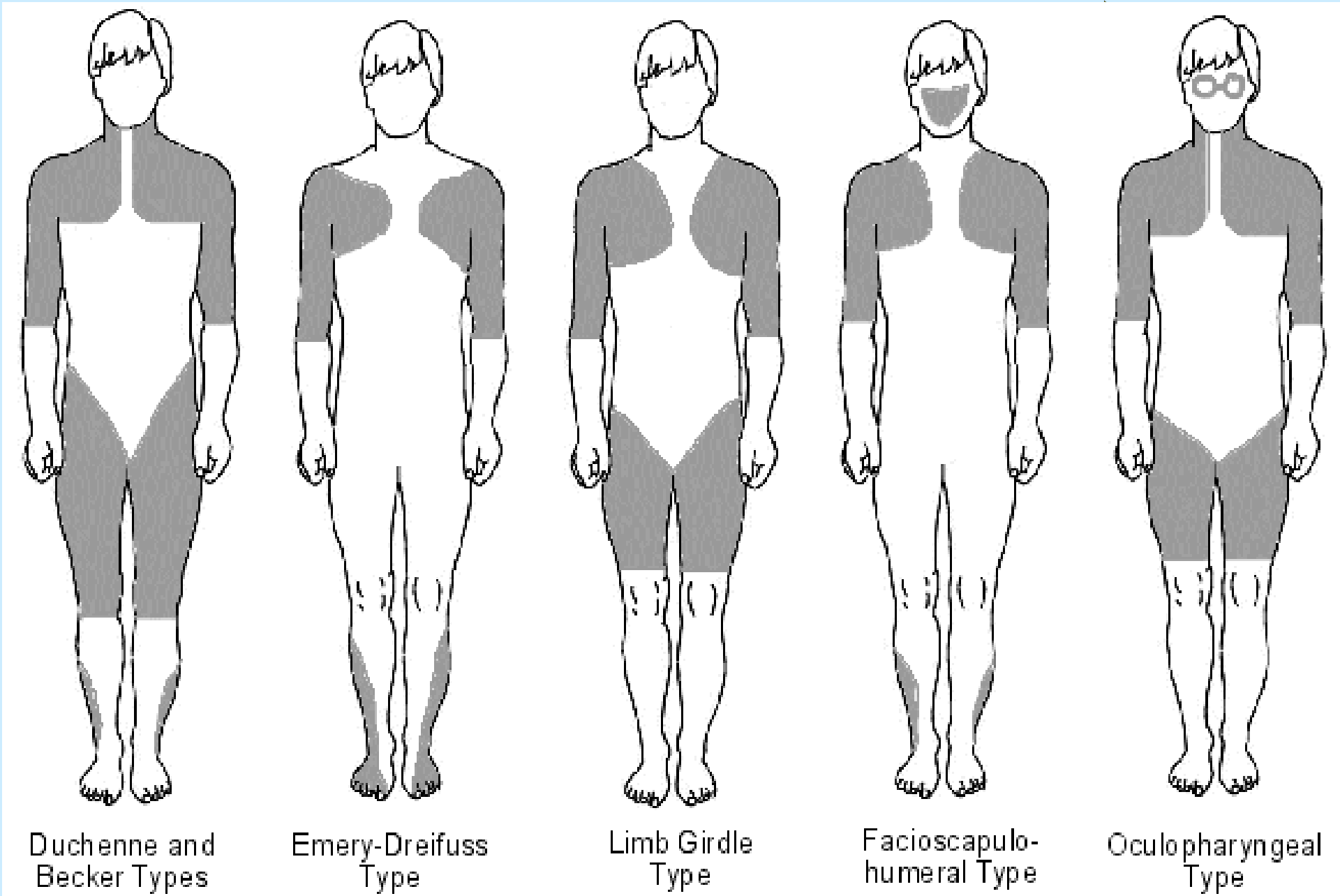

Becker Muscular Dystrophy
10 %
Facio-Scapulo-Humeral Dystrophy
13 %
Duchenne Muscular Dystrophy
23 %
Limb Girdle Muscular Dystrophy
10 %
Hereditary Neuropathy
12%
Congenital Myopathy
7 %
Spinal Muscular
12 %
Myotonic Dystrophy
13%
Congenital Muscular Dystrophy
3%


X-linked recessive, carrier mother
Unaffected father
Carrier mother
X Y
X X
Unaffected
Affected
Carrier
X Y
X X
X X
X Y
Unaffected son
Unaffected daughter
Carrier daughter
Affected son
U.S. National Library of Medicine


Autosomal dominant
Affected father
Unaffected mother
Unaffected
Affected
Affected son
Unaffected daughter
Unaffected son
Affected daughter
U.S. National Library of Medicine

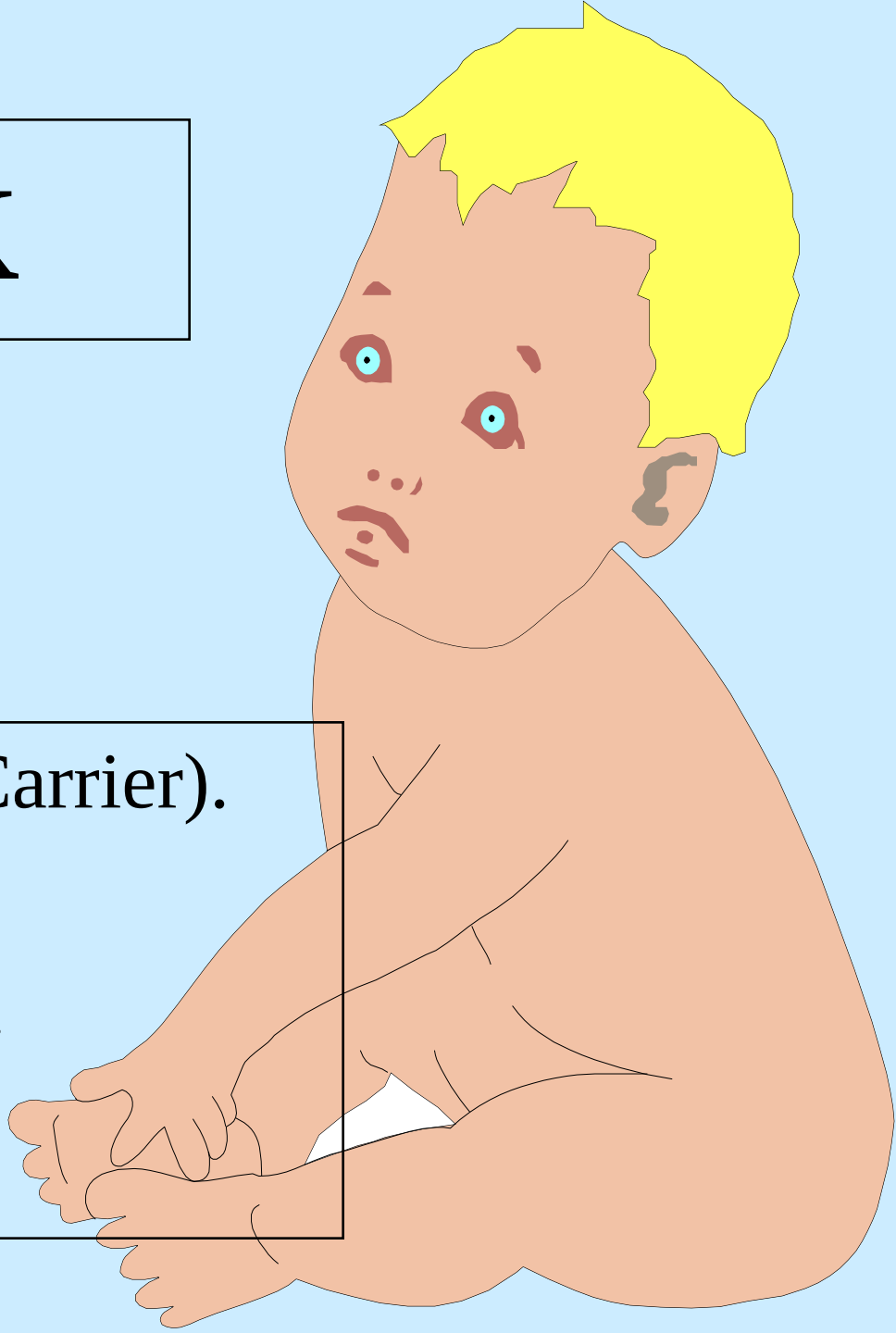

Autosomal recessive
Carrier father
Carrier mother
Unaffected
Affected
Carrier
Unaffected son
Carrier daughter
Carrier son
Affected daughter
U.S. National Library of Medicine





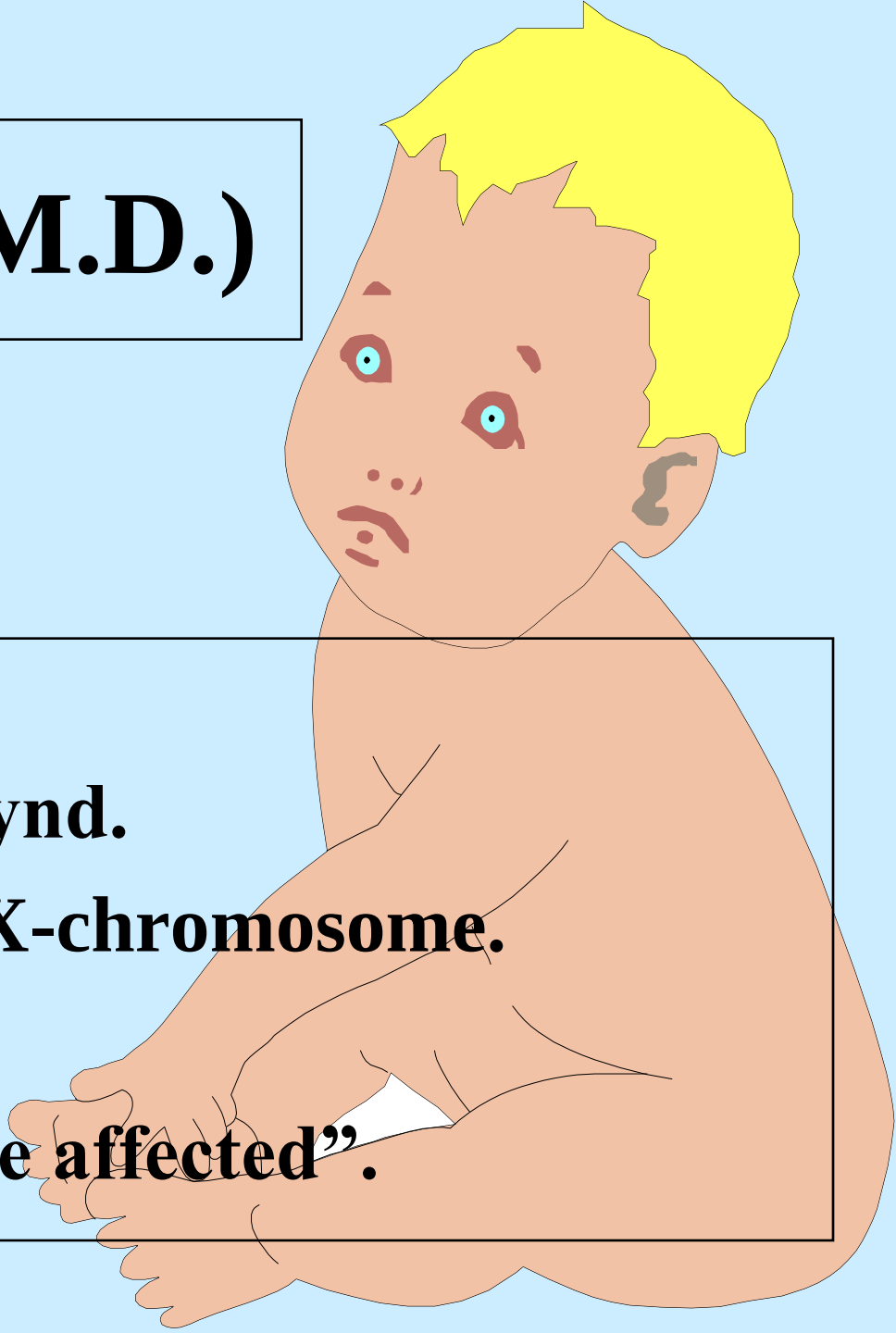
Main areas of muscle weakness in different types of dystrophy

High CPK



- DMD (Male, Female Carrier).
- Dermatomyositis.
- Limb girdle dystrophy.
- SMA type IV.

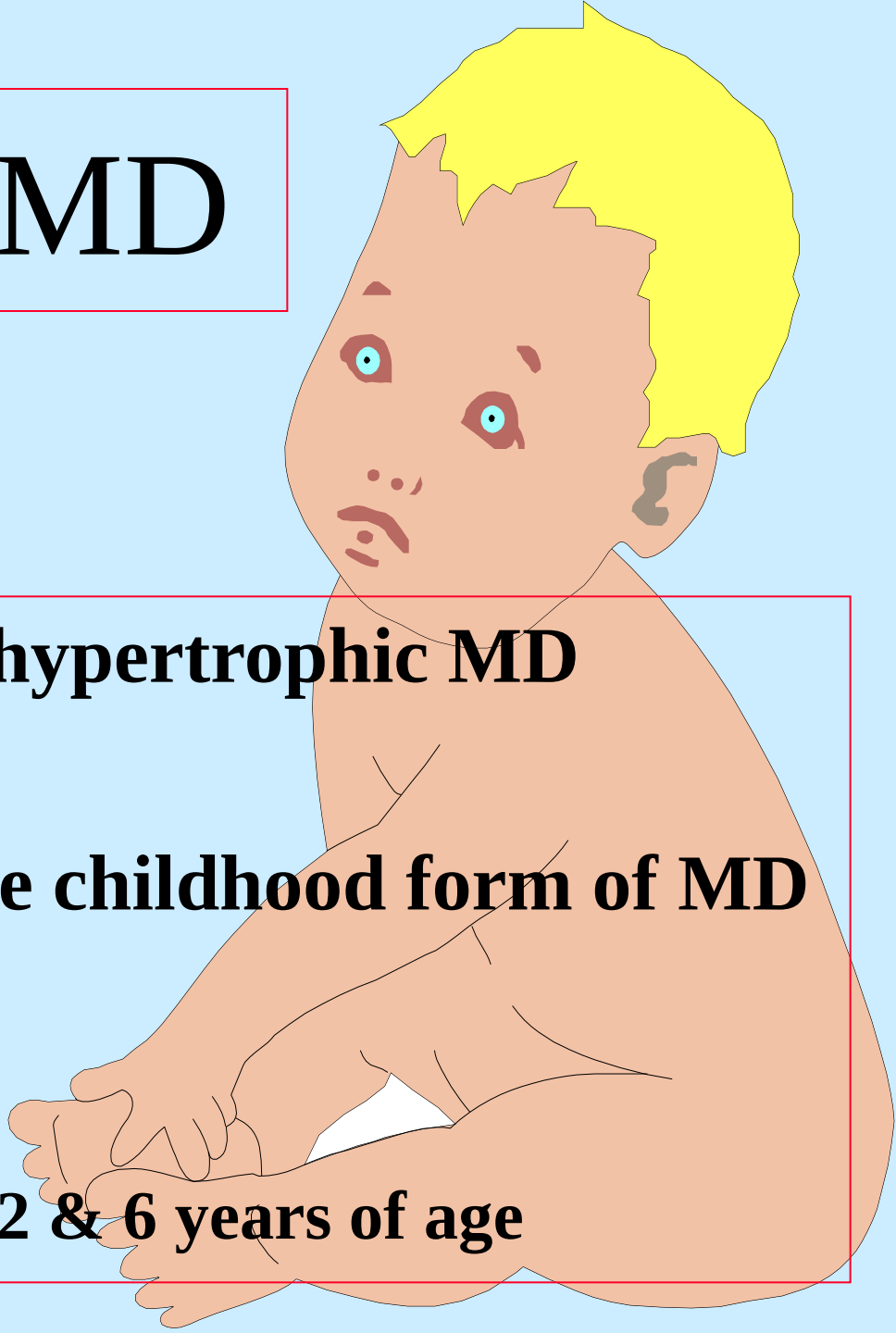
Duchenne (D.M.D.)



* Male only.
* Female - Turner's Synd.
  - Abnormal X-chromosome.
* Female Carrier

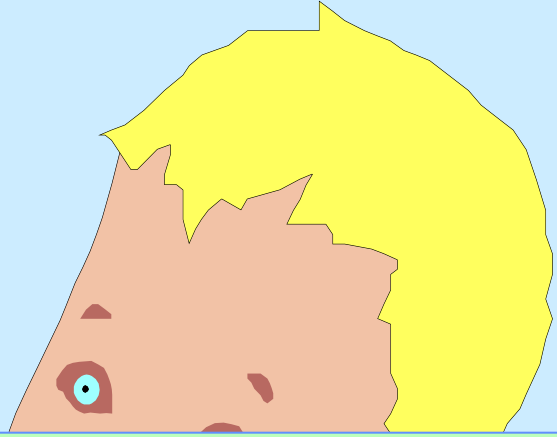
  " 50% of sons will be affected".

Duchenne MD



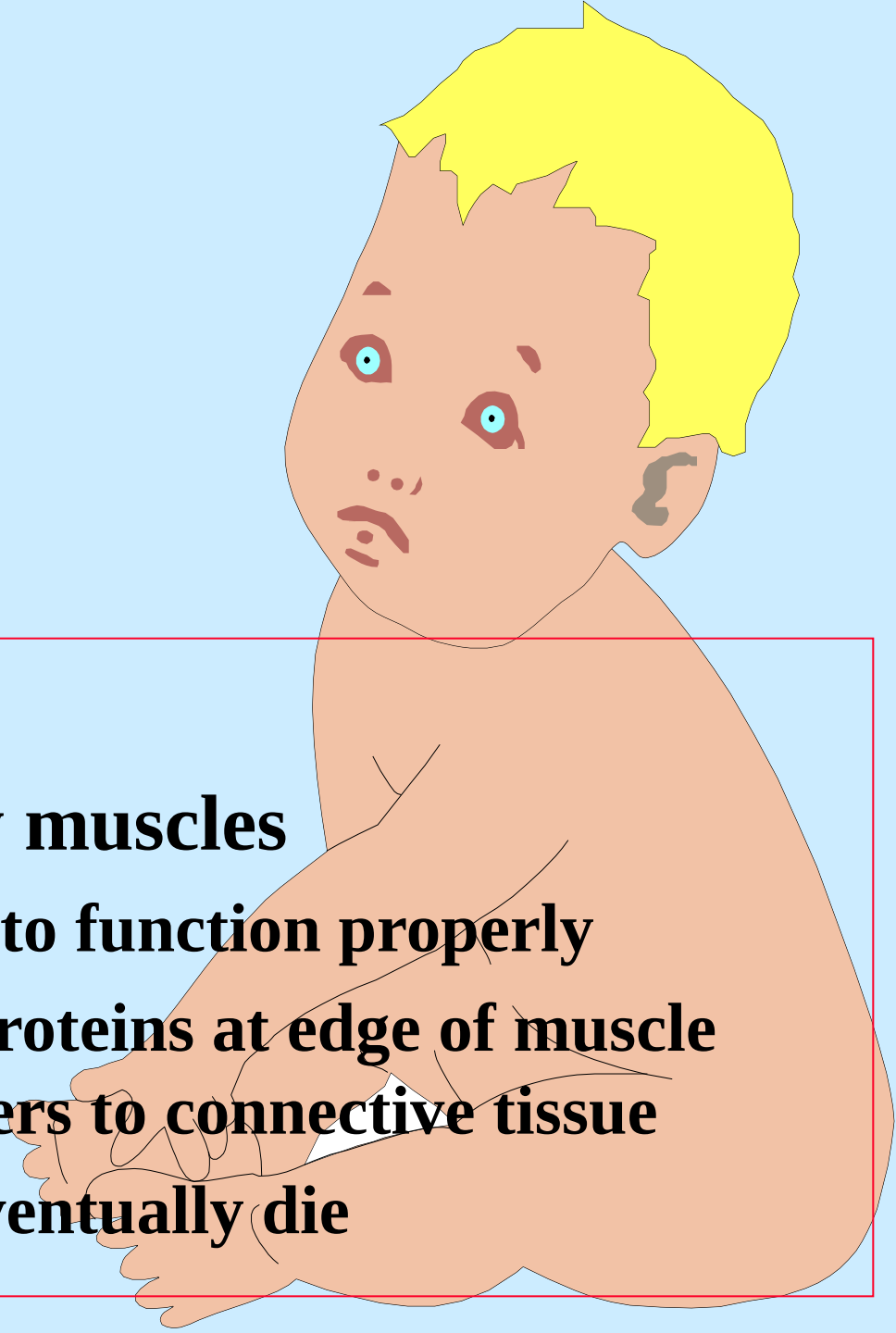
- **Also known as Pseudohypertrophic MD**
- **Genetic disease**
- **Most common & severe childhood form of MD**
- **Affects boys**
- **Symptoms:**
  - **Usually occur between 2 & 6 years of age**

Cause of DMD



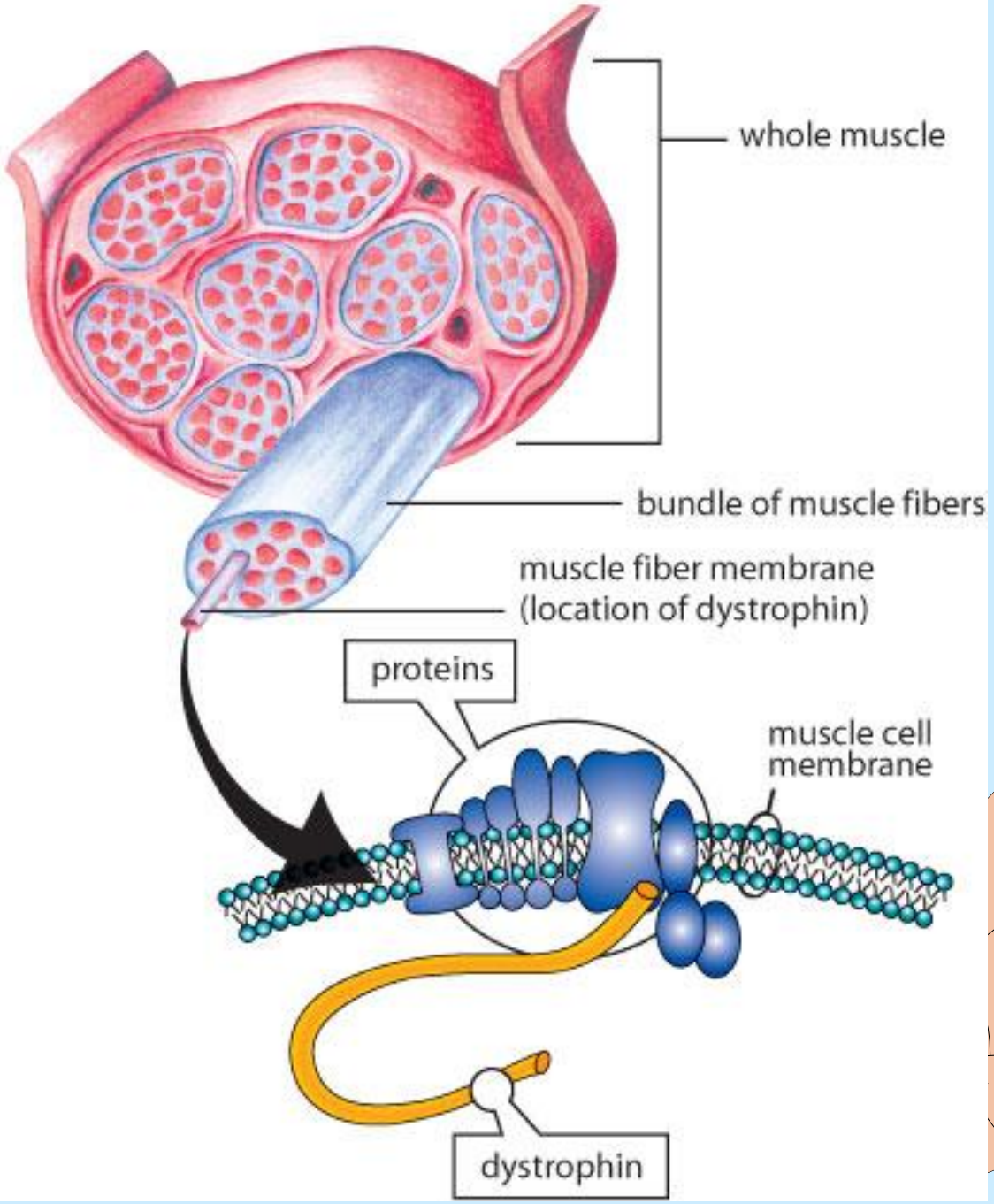
Gowers in 1886 concluded the genetic basis of disease, and in 1986 Kunkel identified dystrophin gene mutations that are responsible for disease development.

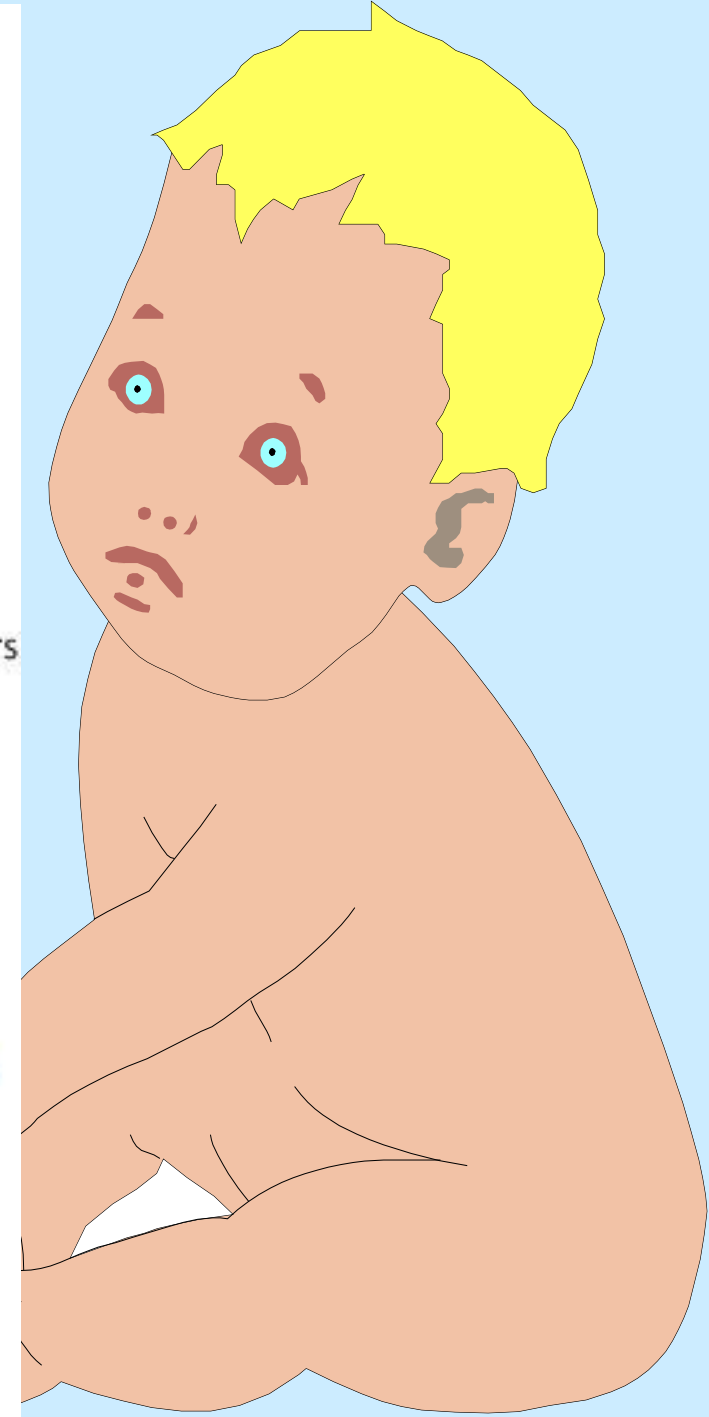
* **Genetic defect causing lack of Dystrophin (Protein on muscle membrane).**
* **Site of chromosomal abnormality.**
  - **Central area of short arm X-Chromosome (Band P21).**



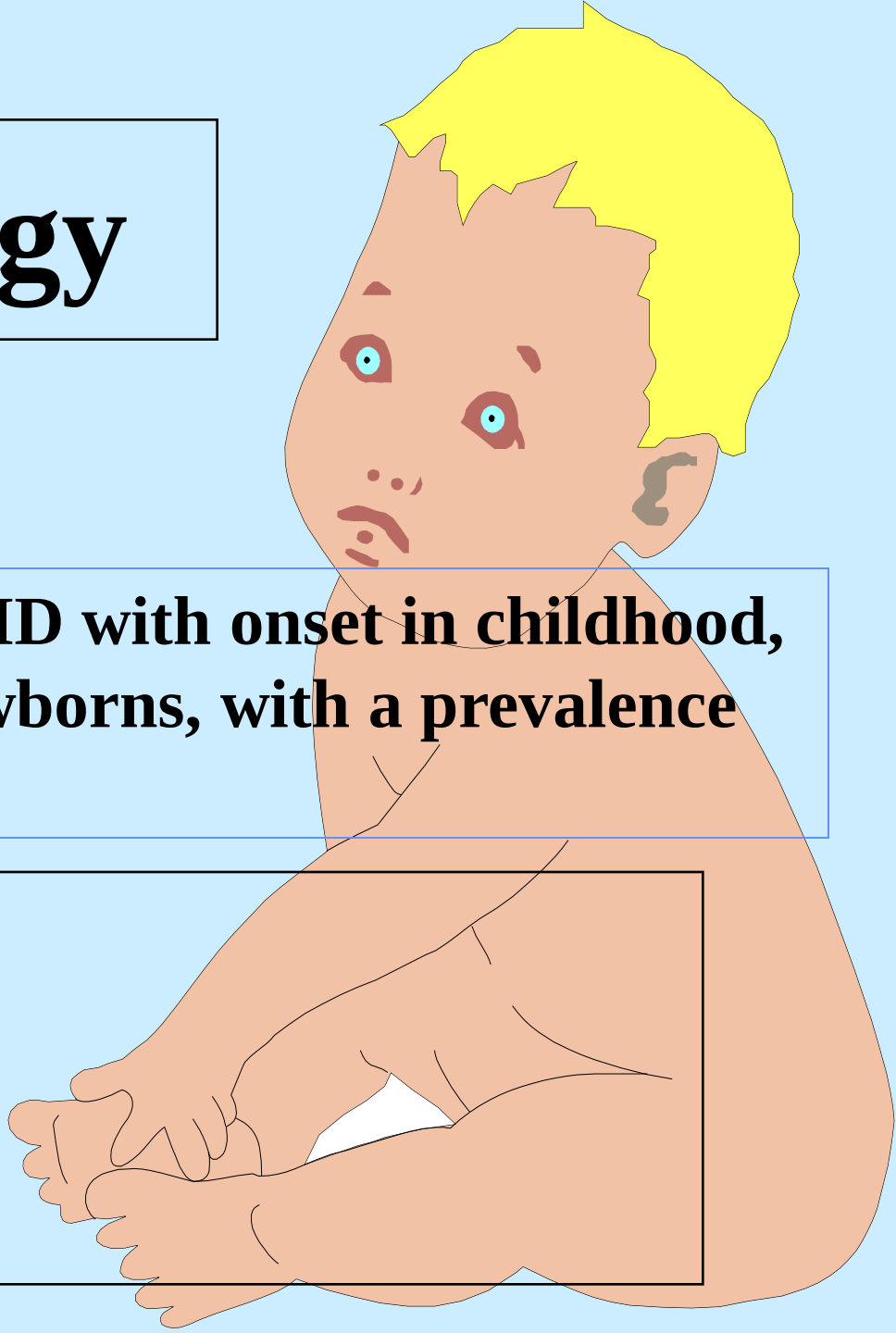
Dystrophin

- Protein required by muscles
  - Allows muscle cells to function properly
  - Attached to other proteins at edge of muscle fibers to anchor fibers to connective tissue
  - W/o muscle cells eventually die


whole muscle
bundle of muscle fibers
muscle fiber membrane
(location of dystrophin)
proteins
muscle cell
membrane
dystrophin



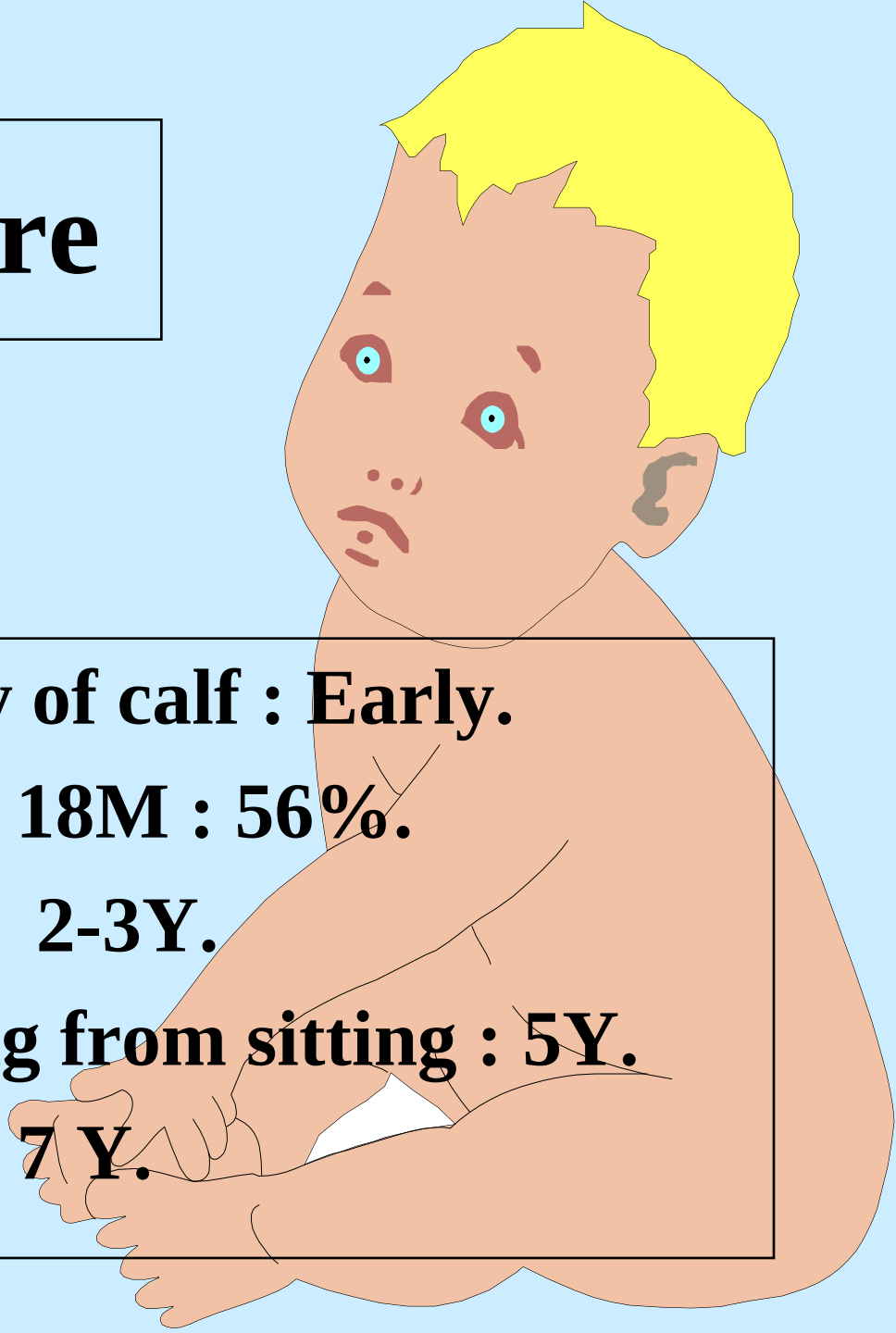
Epidemiology



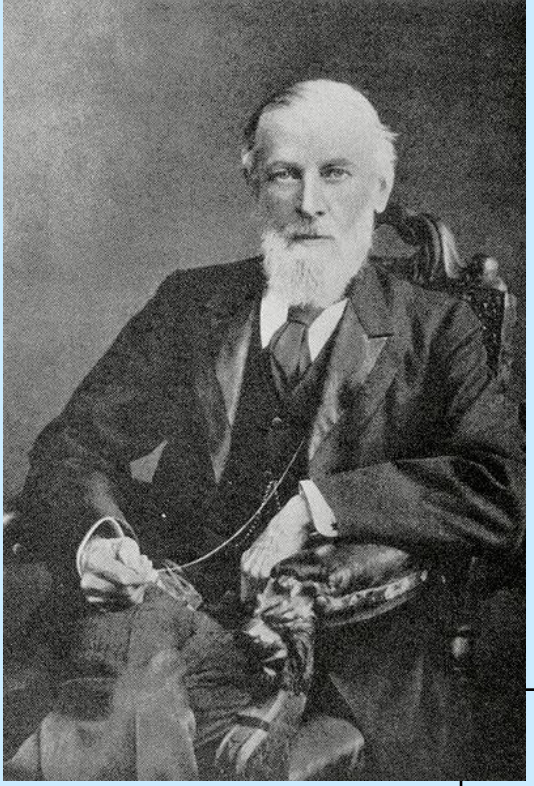
DMD is the most common MD with onset in childhood, affecting 1 in 3,300 male newborns, with a prevalence of 63 cases per 1 million.

- **60% Family history.**
- **30% Mutation.**

Clinical picture

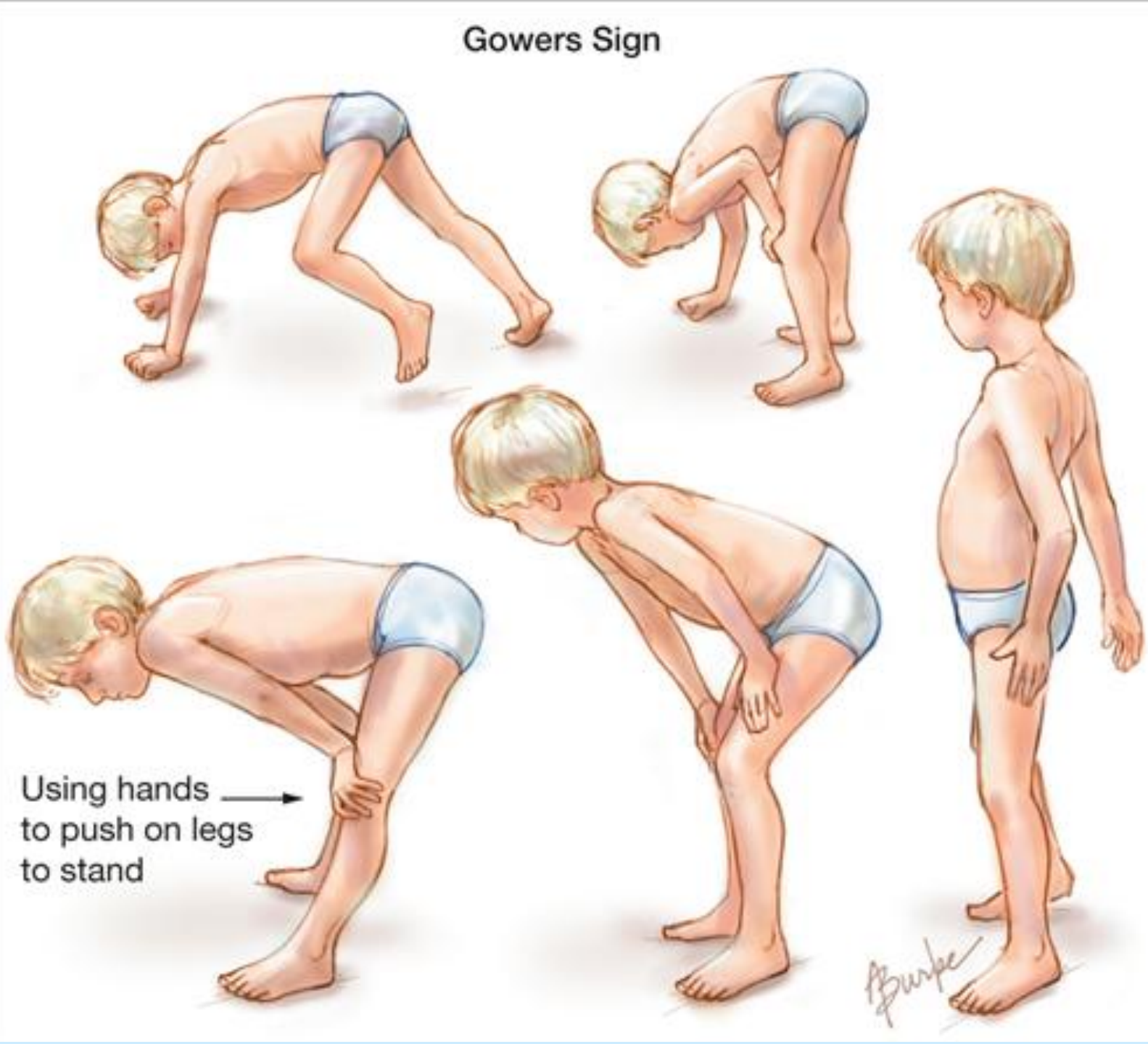


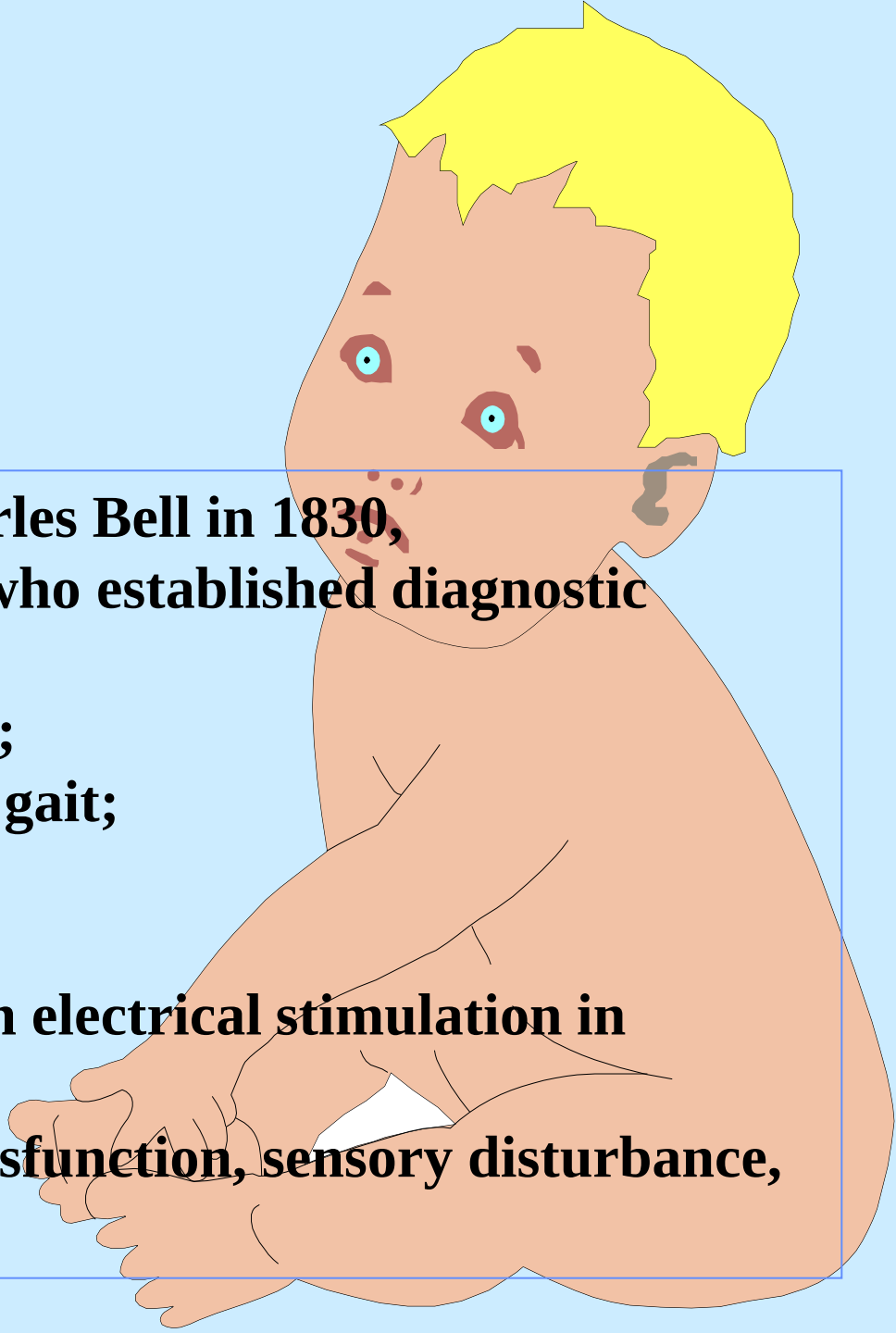
- Pseudo hypertrophy of calf : Early.
- Delayed walking : > 18M : 56%.
- Clumsy walking : 2-3Y.
- Difficulty in standing from sitting : 5Y.
- Tip toe walking : 7 Y.



Gower's Sign

- At 5 years.
- Weak hip extension & quadriceps.
- Prognostic sign when timed.


Gowers Sign
Using hands
to push on legs
to stand



DMD was first described by Charles Bell in 1830, but Duchenne in 1868 is the one who established diagnostic criteria that are valid today:

= Weakness with onset in the legs;

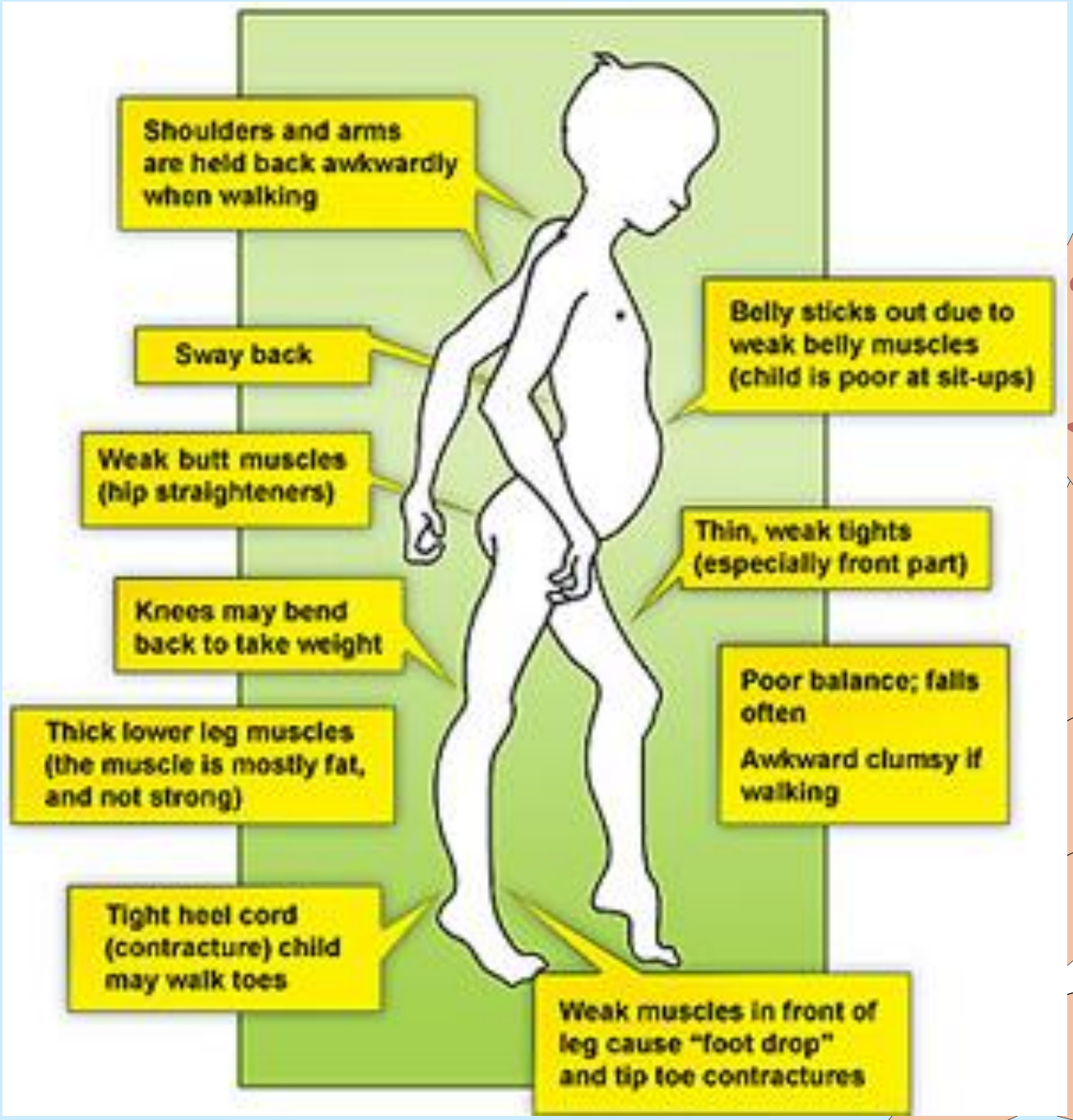
= Hyperlordosis with wide-based gait;

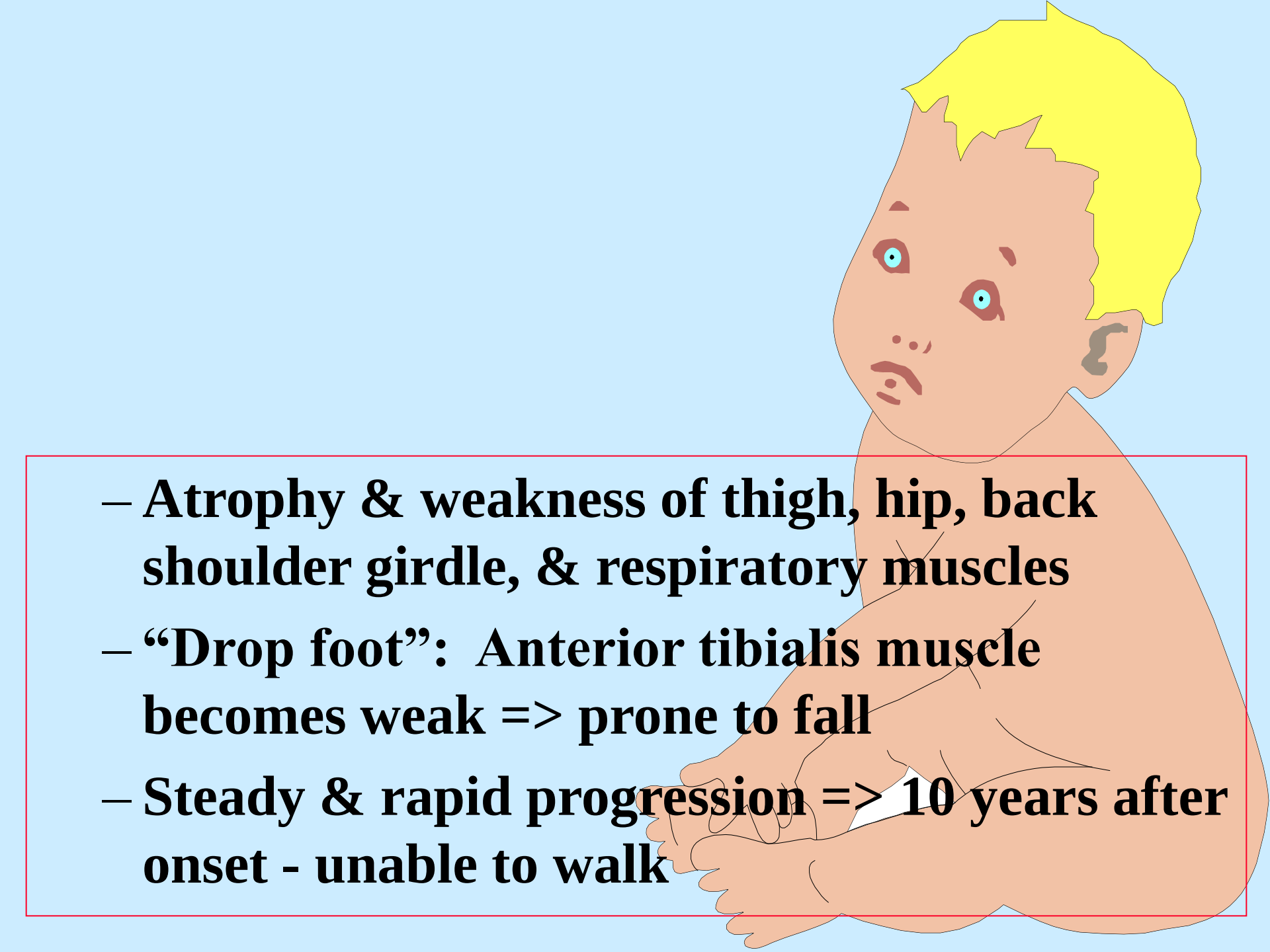
= Hypertrophy of weak muscles;

= Progressive course over time;

= Reduced muscle contractility on electrical stimulation in advanced stages of the disease;

= Absence of bladder or bowel dysfunction, sensory disturbance, or febrile illness.


Shoulders and arms are held back awkwardly when walking
Sway back
Weak butt muscles (hip straighteners)
Knees may bend back to take weight
Thick lower leg muscles (the muscle is mostly fat, and not strong)
Tight heel cord (contracture) child may walk toes
Belly sticks out due to weak belly muscles (child is poor at sit-ups)
Thin, weak tights (especially front part)
Poor balance; falls often
Awkward clumsy if walking
Weak muscles in front of leg cause "foot drop" and tip toe contractures

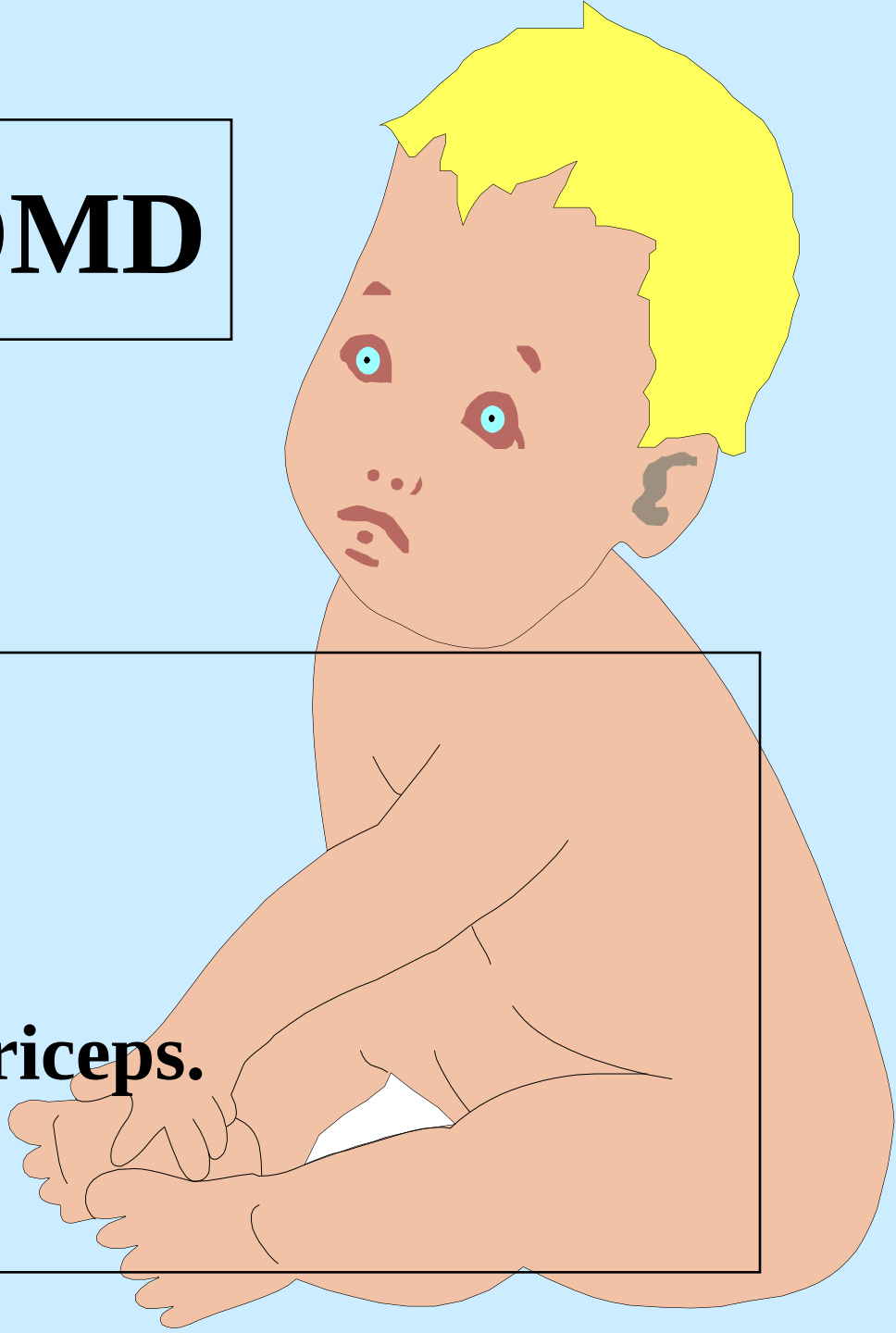


- Atrophy & weakness of thigh, hip, back shoulder girdle, & respiratory muscles
- “Drop foot”: Anterior tibialis muscle becomes weak => prone to fall
- Steady & rapid progression => 10 years after onset - unable to walk

- Lordosis: due to weakness of trunk muscles
- Orthopedic devices necessary as disease progresses (leg braces, walkers, etc.)
- Scoliosis usually develops when in wheelchairs

Diagnosis of DMD

- Male.
- High CPK.
- Needle Bx.
- Ultrasound to quadriceps.
- EMG!.



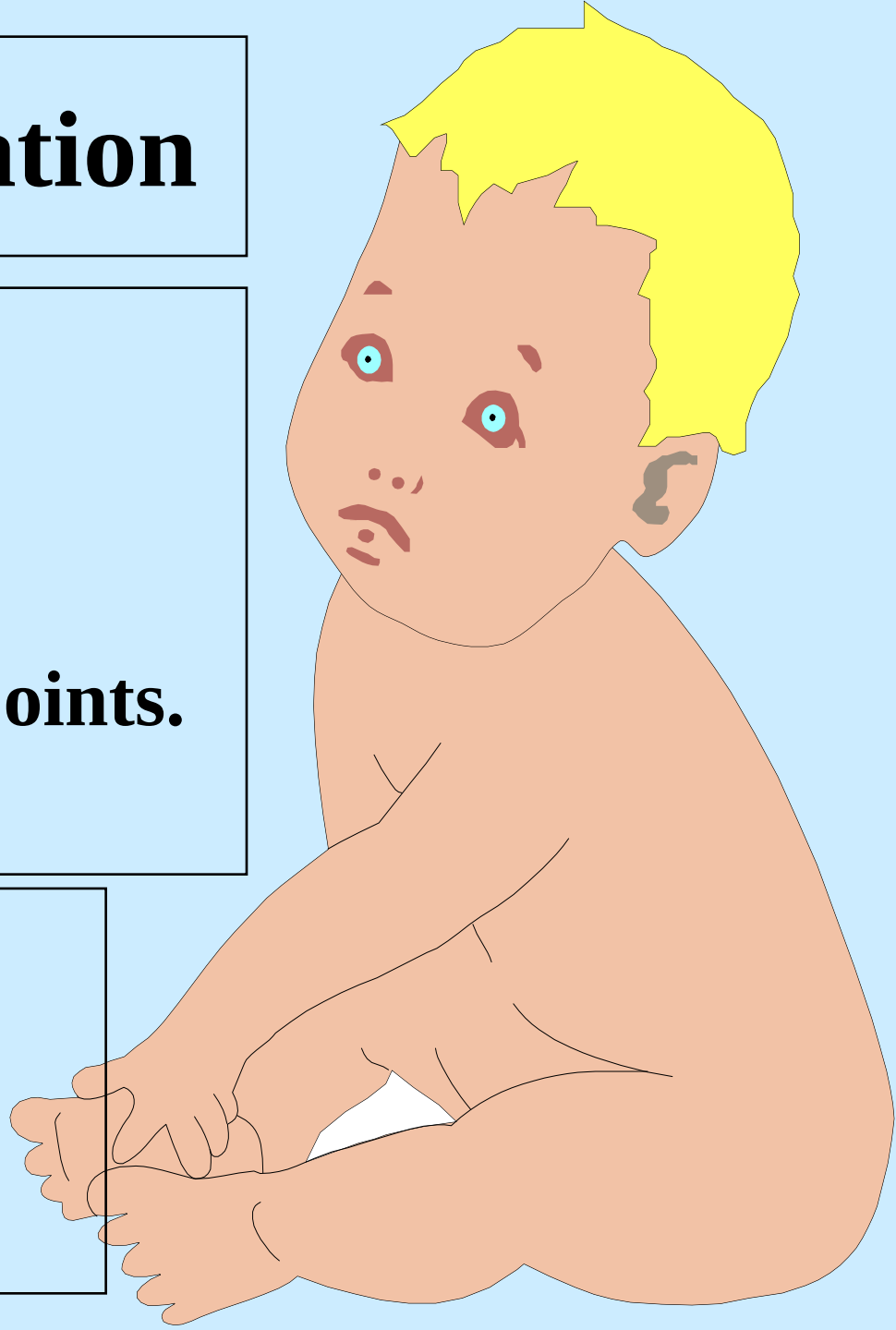
Routine Examination

1. Clinical

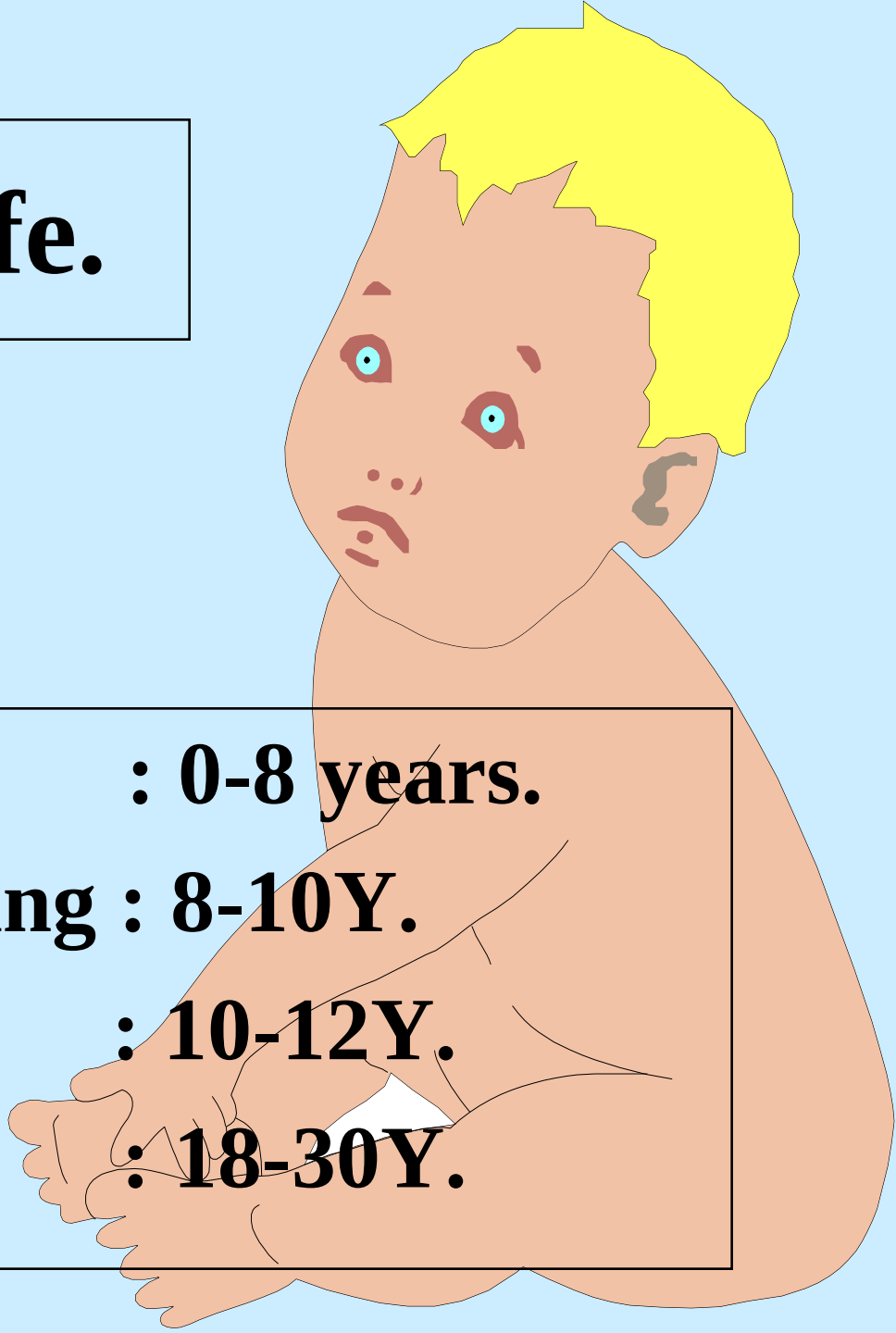
- Timed Gower's sign.
- Walking speed.
- Contractures of the joints.
- Spine.

2. Investigations

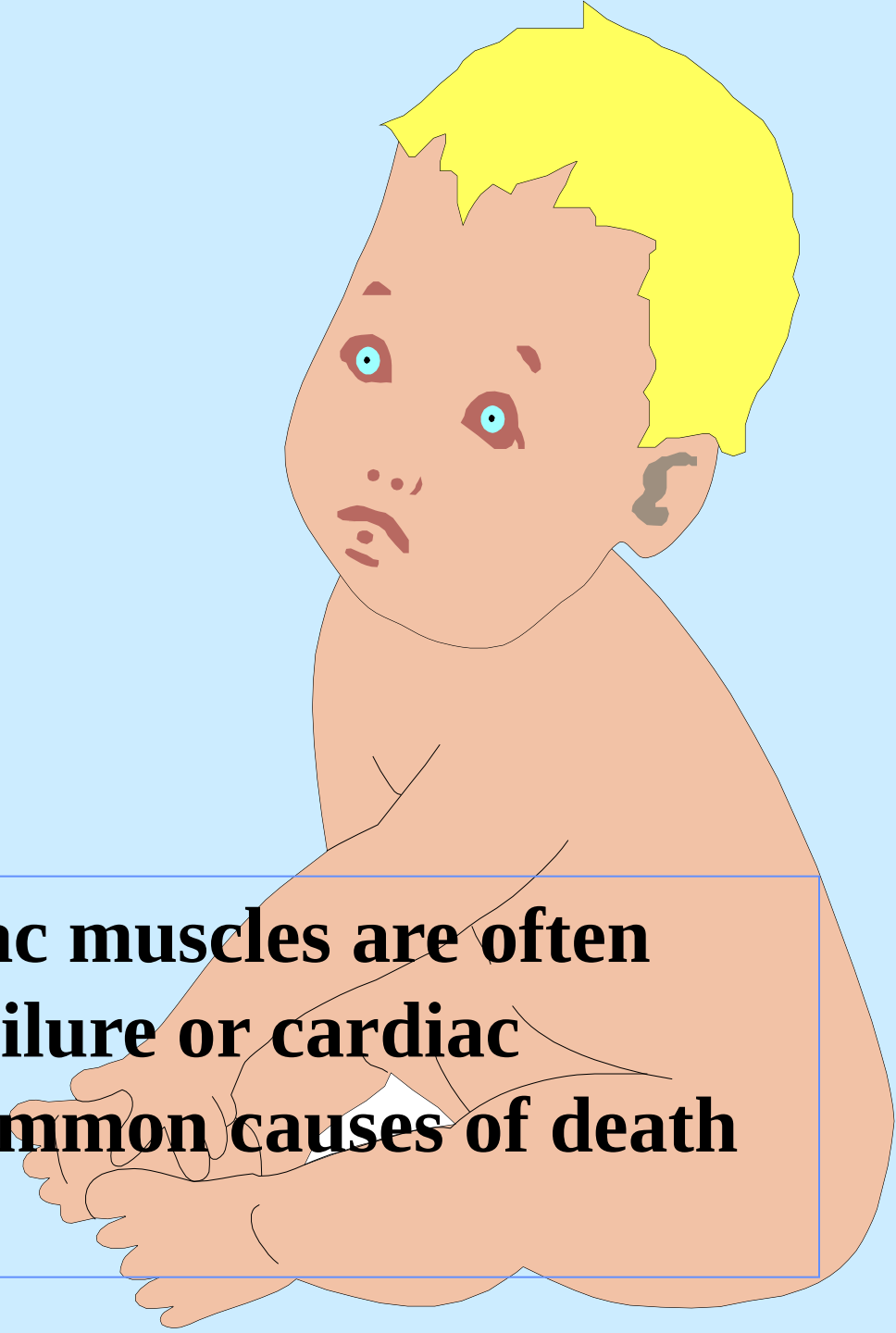
- FVC.
- Sitting spine x-ray.
- ECG.



Duchenne Life.

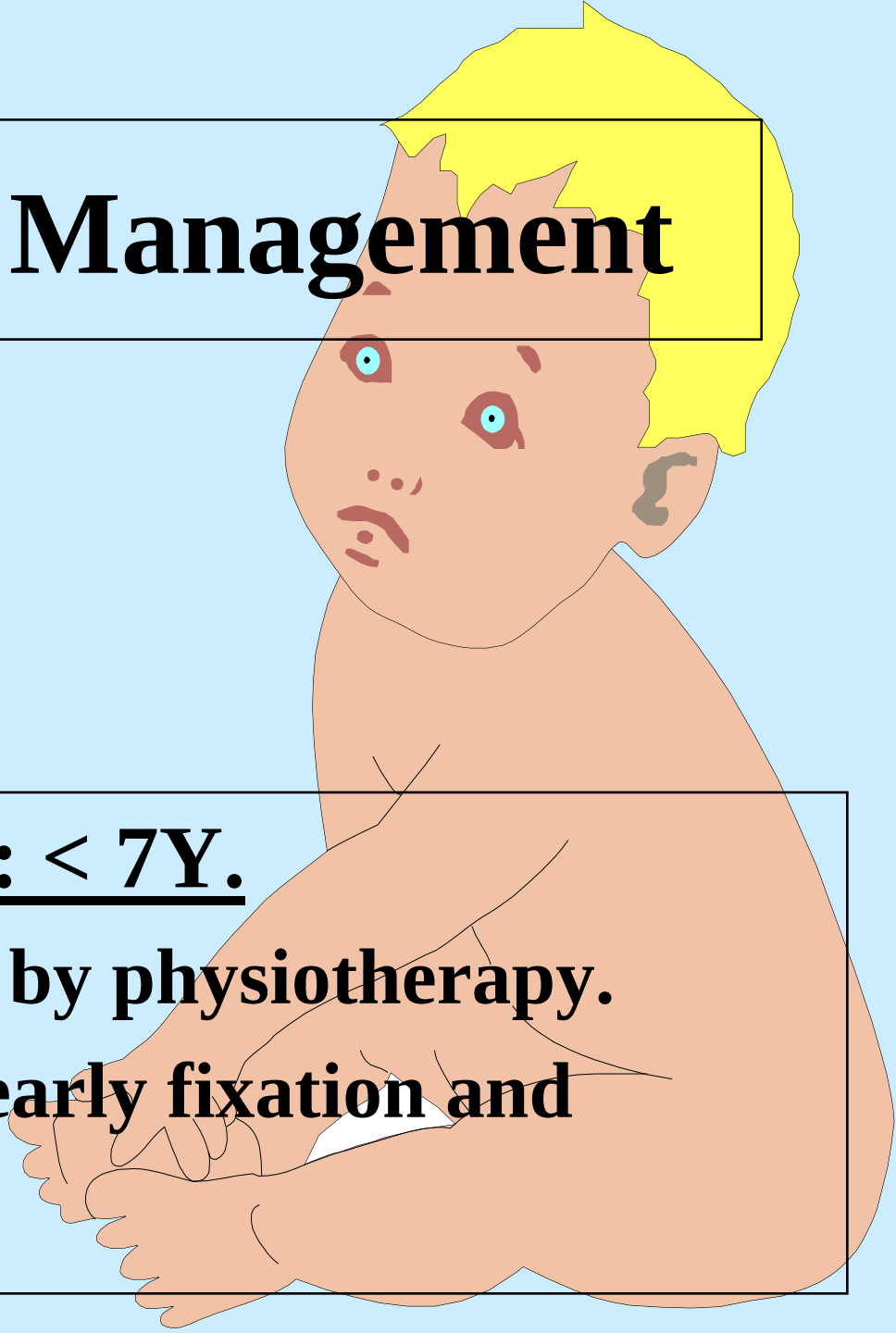


- Normal life : 0-8 years.
- Decreased walking : 8-10Y.
- Wheel chair : 10-12Y.
- Death : 18-30Y.



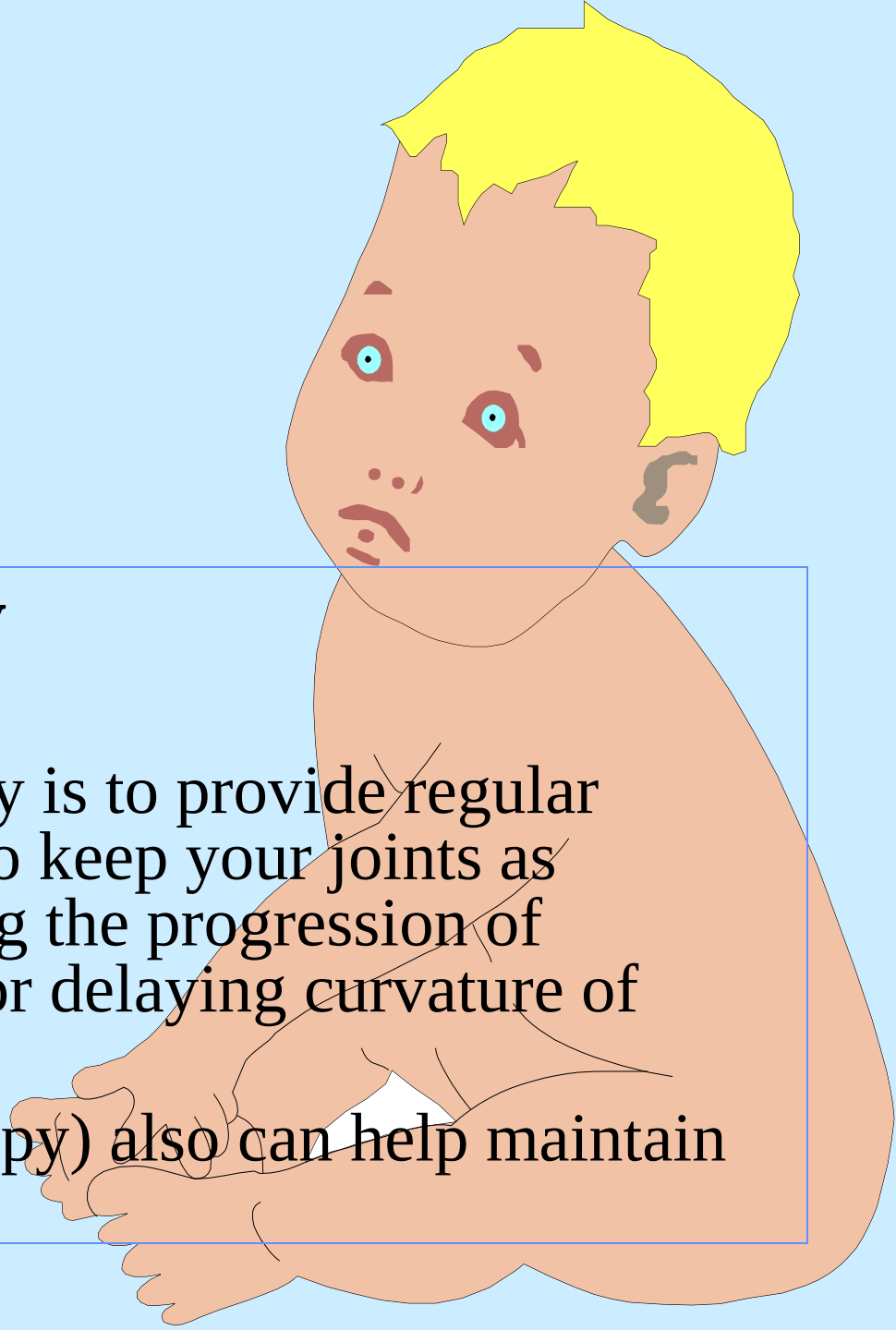
Respiratory and cardiac muscles are often affected, respiratory failure or cardiac decompensation are common causes of death in these patients.

Orthopaedic Management



1. Period of walking : < 7Y.

- Prevent contracture by physiotherapy.
- Any fracture needs early fixation and mobilization.

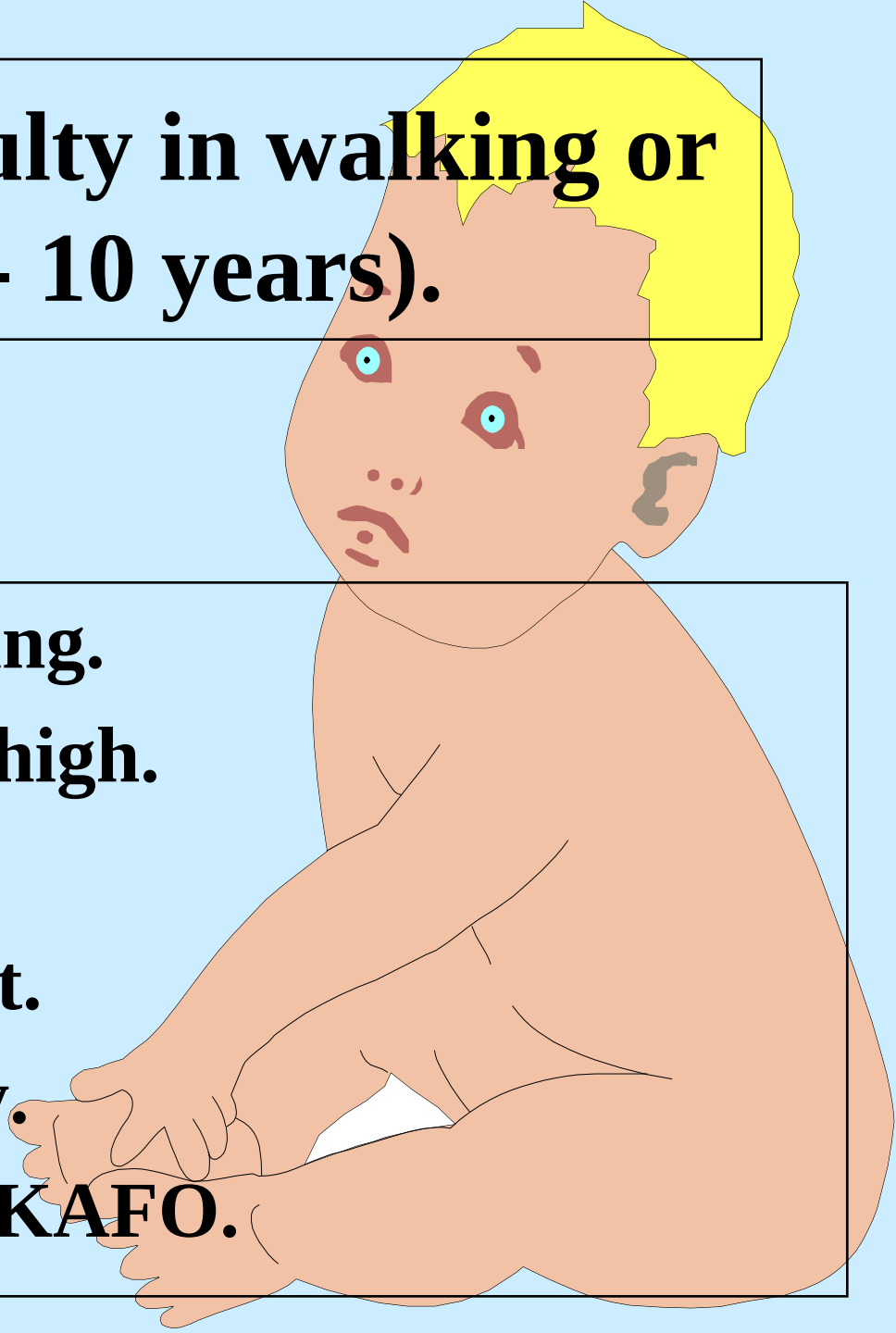


Physiotherapy

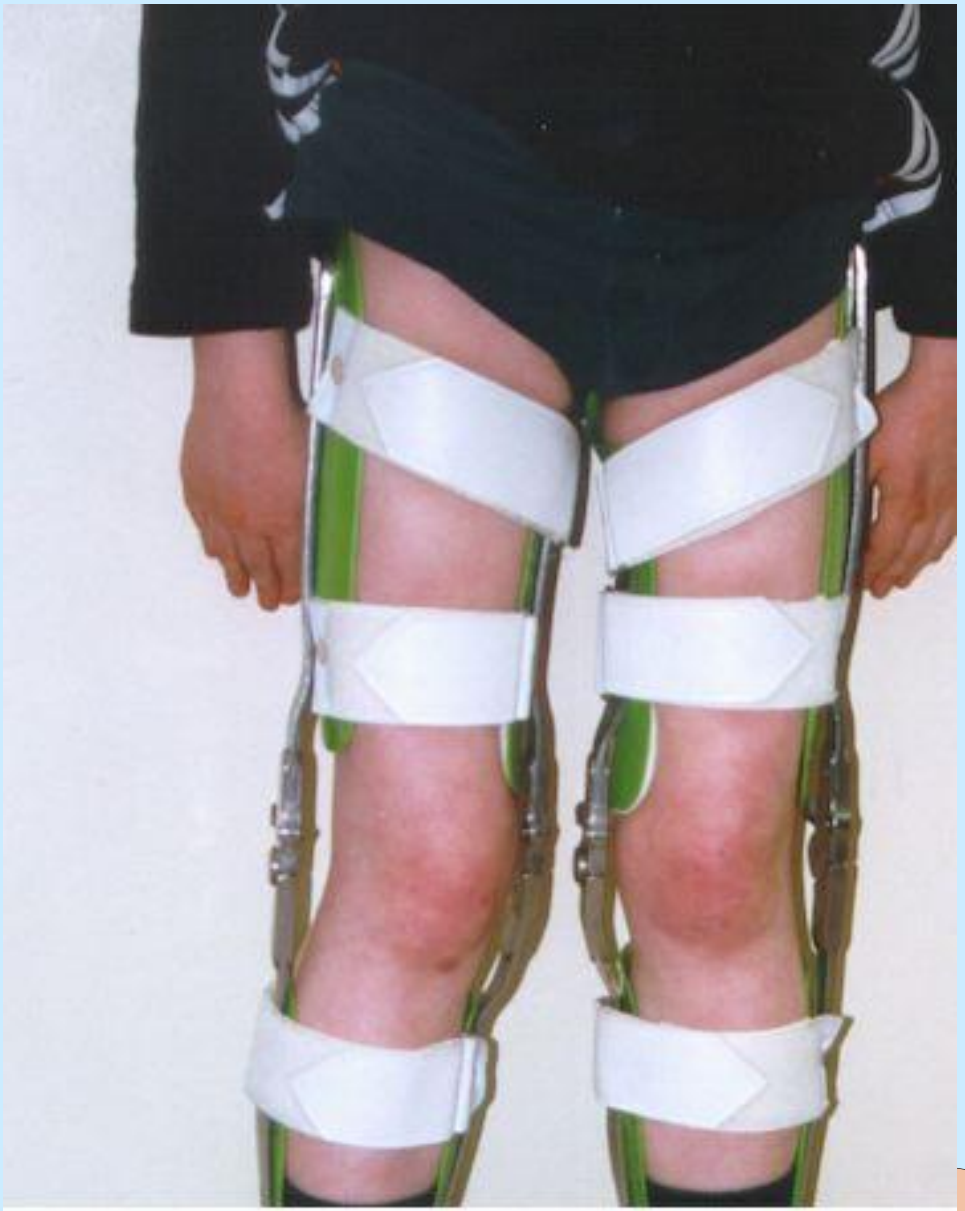
One goal of physical therapy is to provide regular range-of-motion exercises to keep your joints as flexible as possible, delaying the progression of contractures, and reducing or delaying curvature of your spine.

Using hot baths (hydrotherapy) also can help maintain range of motion in joints.

2. Period of difficulty in walking or standing (7 - 10 years).

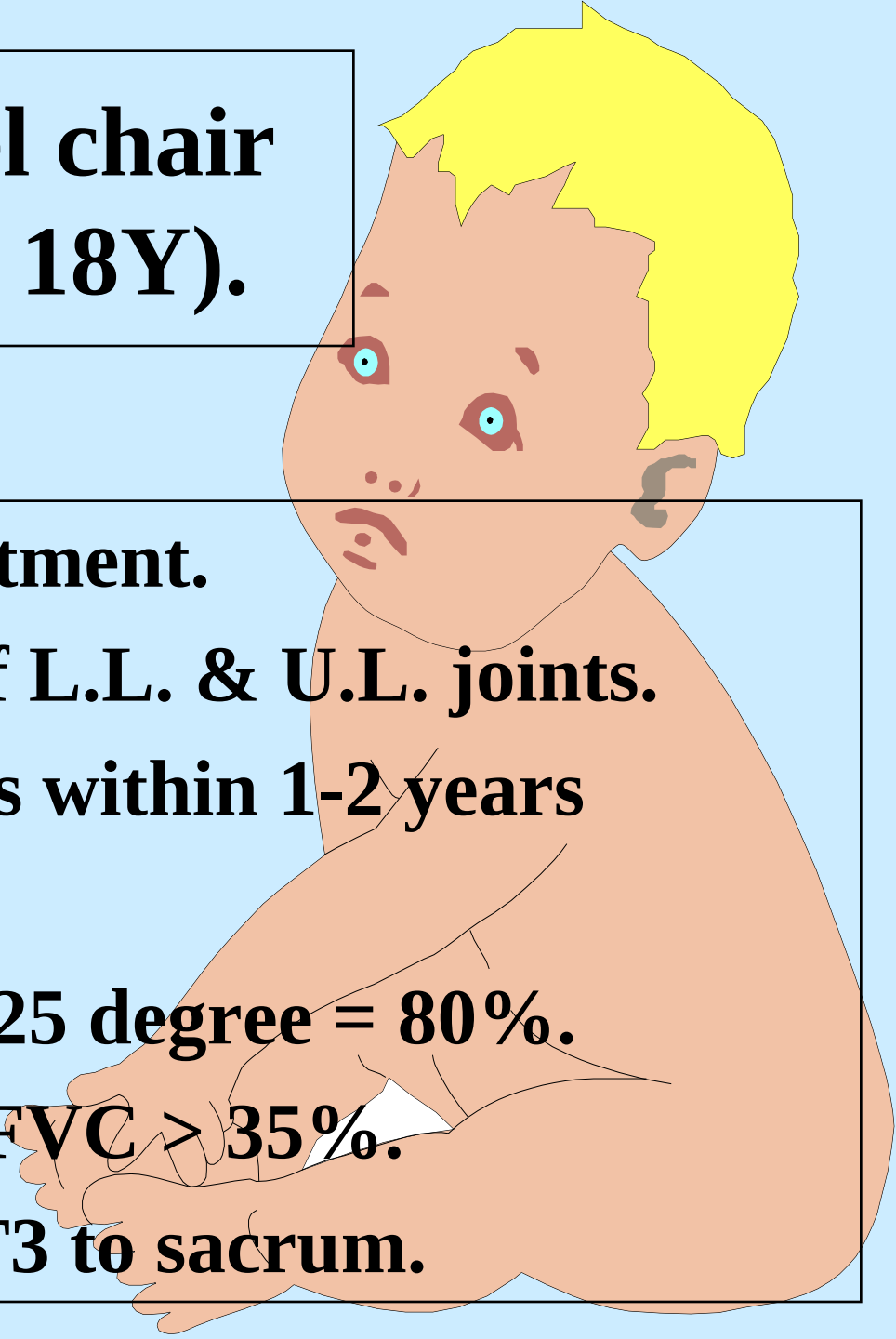


- Percut. TA lengthening.
- Release of ITB mid thigh.
- Post op.
  - * Bilat. long leg cast.
  - * Mobilize next day.
  - * After 2W -- light KAFO.



light KAFO

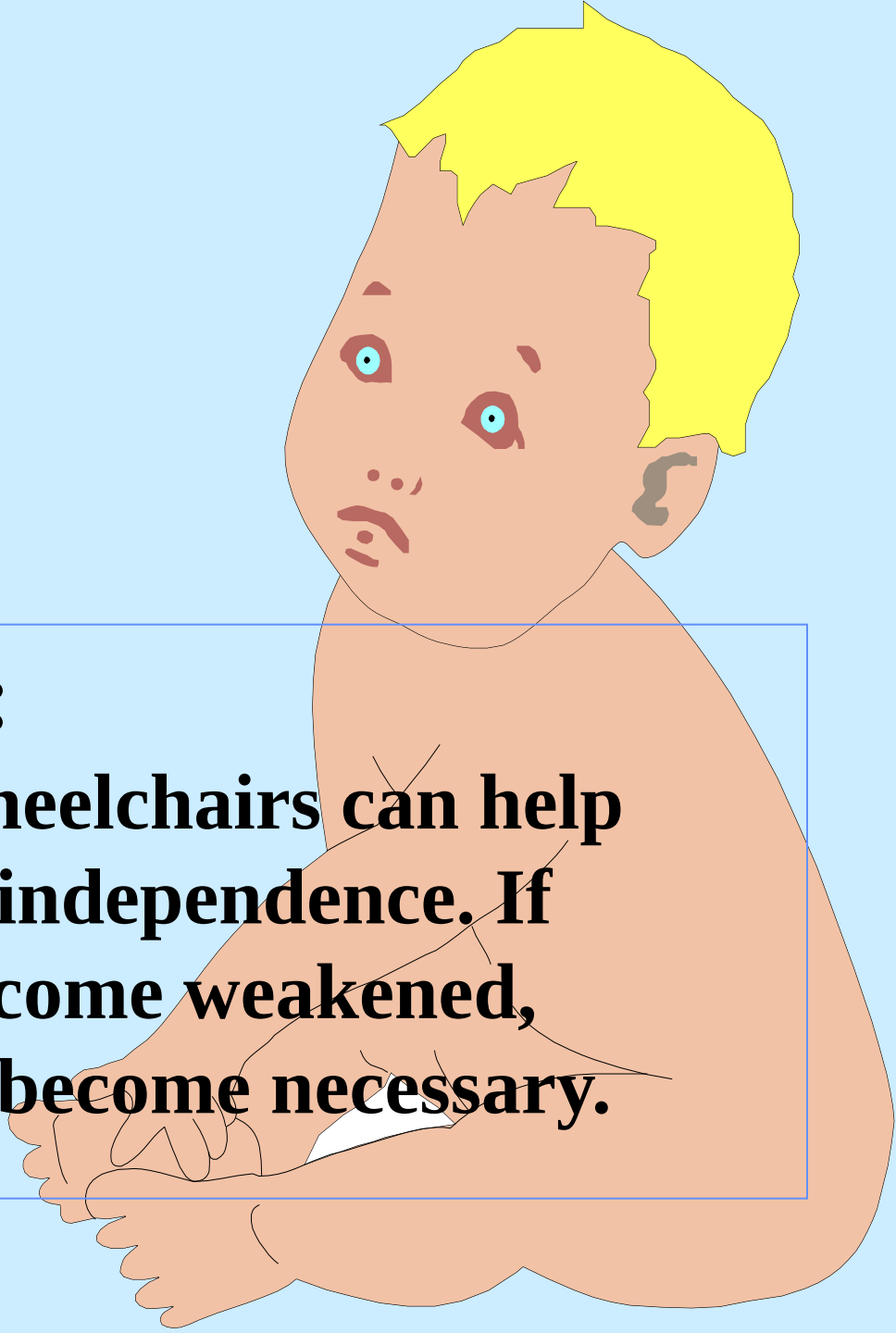
3. Period of wheel chair dependance (10 - 18Y).



- Proper seating adjustment.
- Regular stretching of L.L. & U.L. joints.
- 95% develop scoliosis within 1-2 years of loss of walking.
- Scoliosis surgery : > 25 degree = 80%.

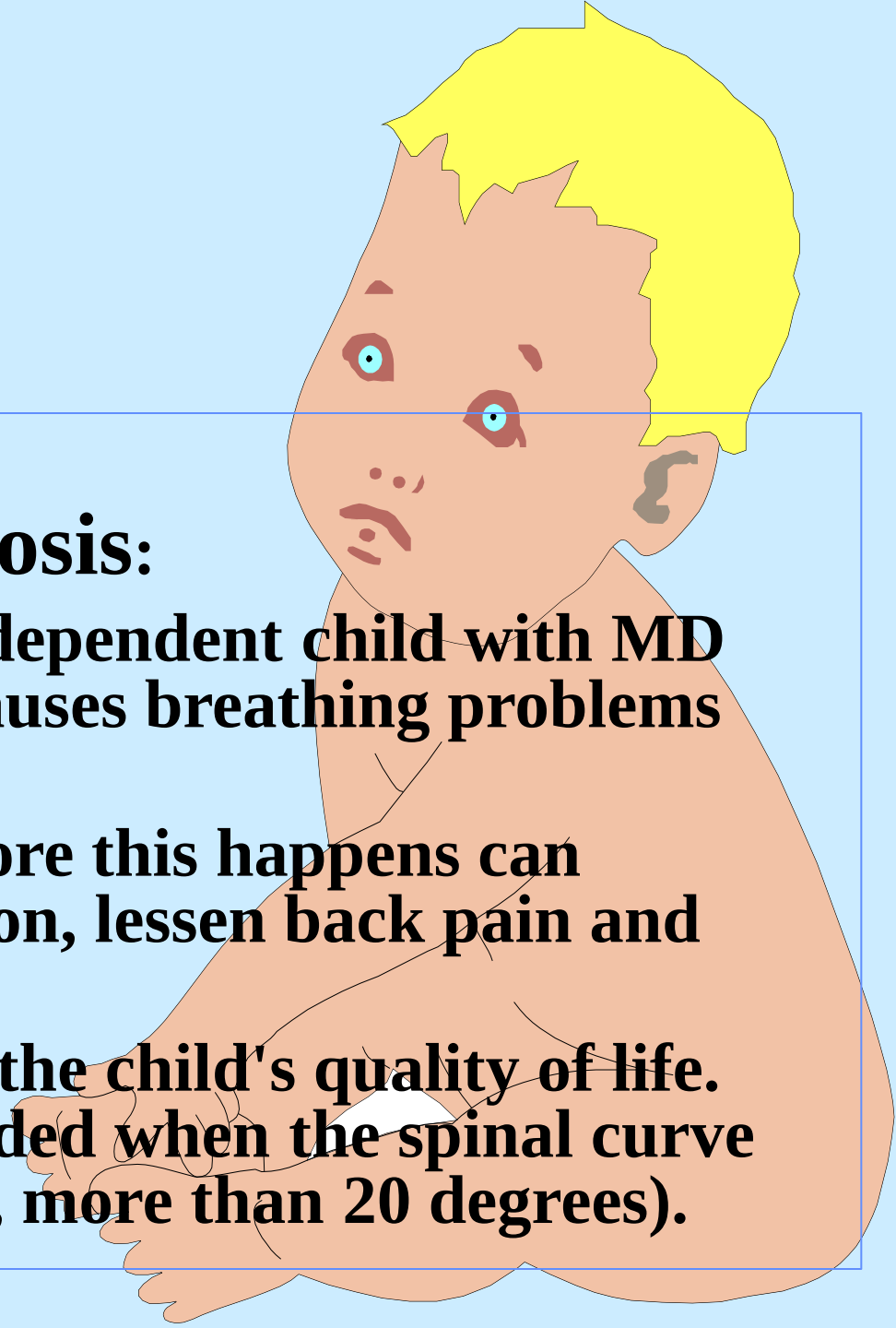
  FVC > 35%.

  T3 to sacrum.



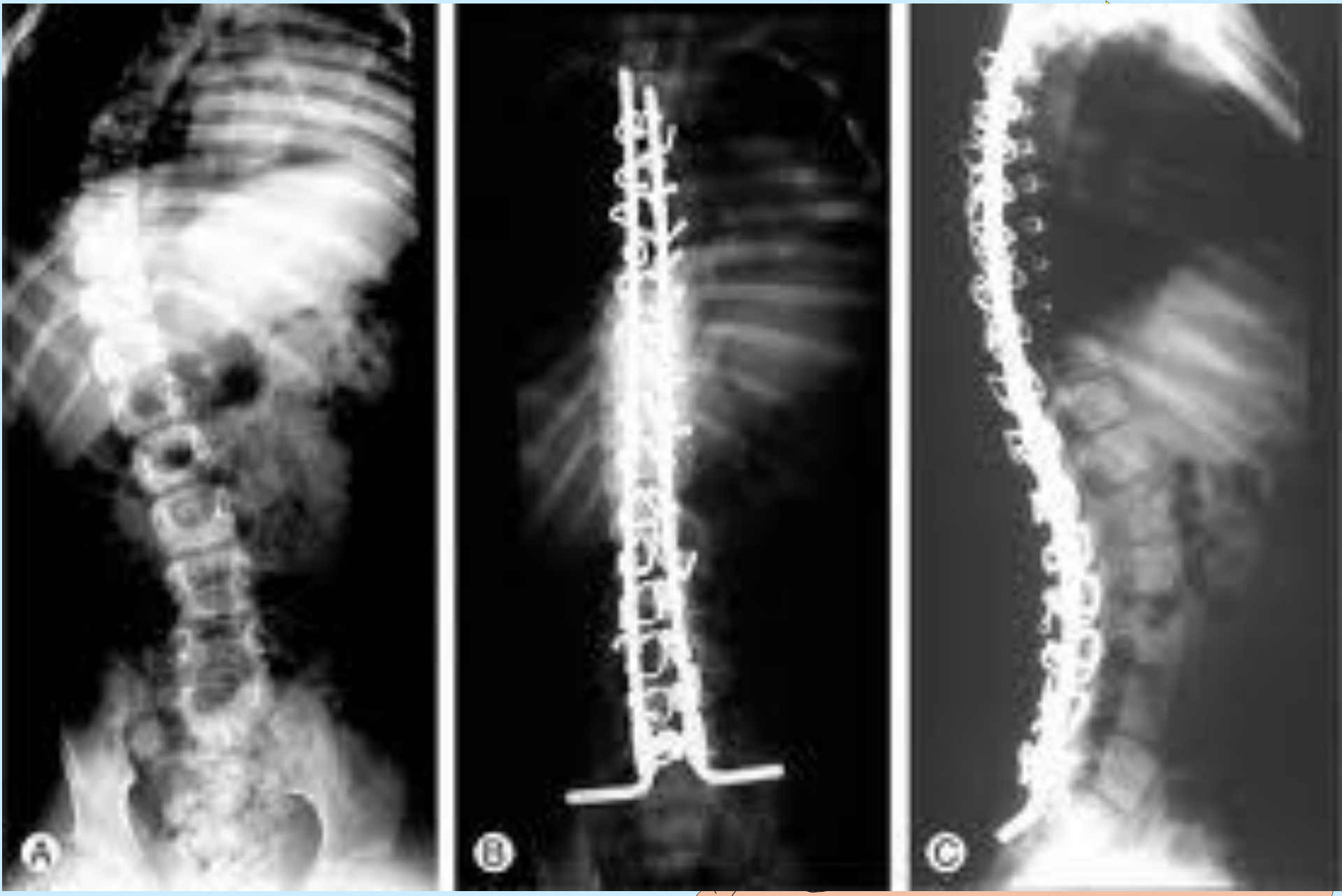
Assistive devices:

Canes, walkers and wheelchairs can help maintain mobility and independence. If respiratory muscles become weakened, using a ventilator may become necessary.



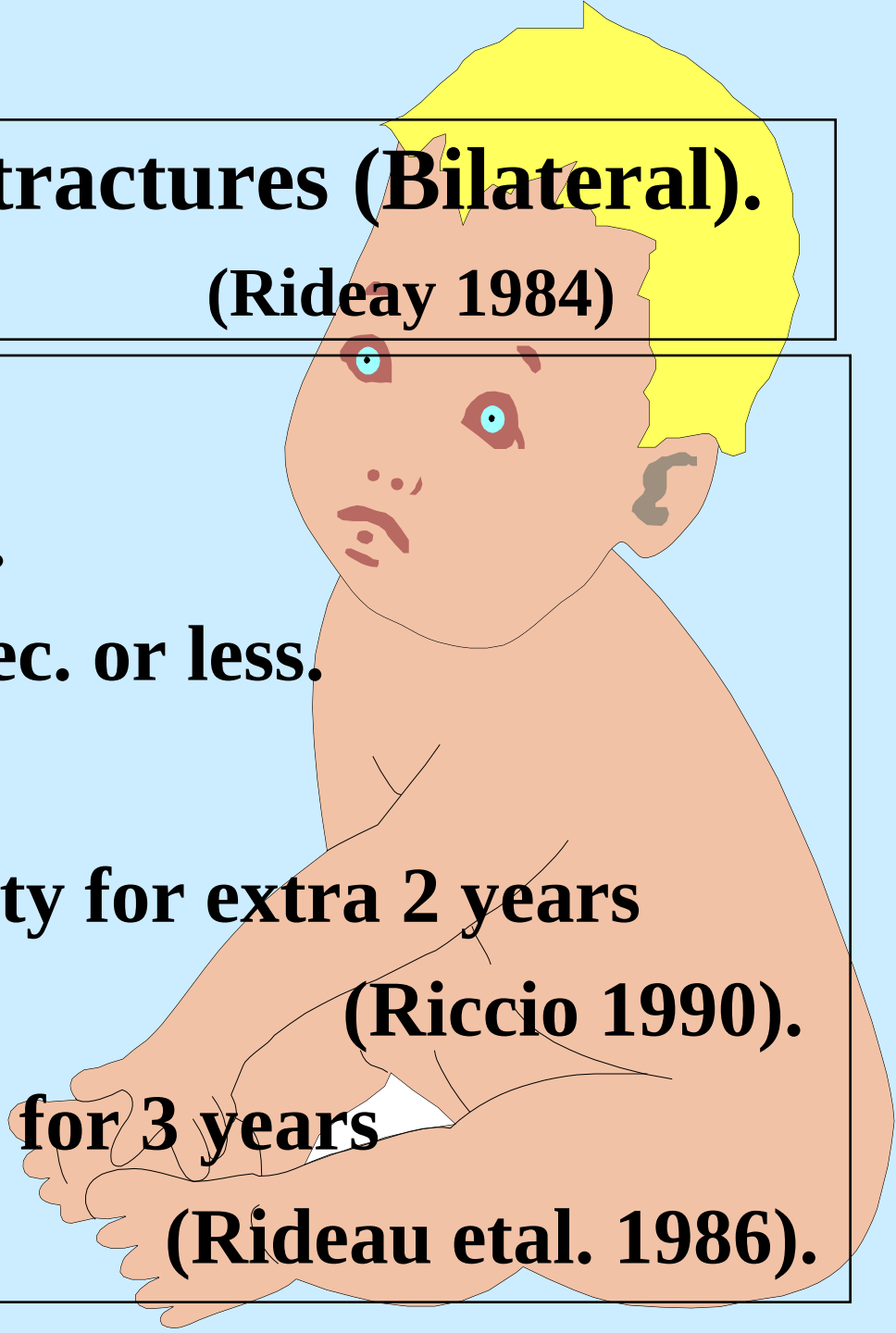
Spinal fusion for scoliosis:

- Scoliosis in a wheelchair-dependent child with MD can become so severe it causes breathing problems and pneumonia.
- Having spine surgery before this happens can preserve breathing function, lessen back pain and improve sitting balance.
- All these factors improve the child's quality of life. The surgery is recommended when the spinal curve reaches a certain size (i.e., more than 20 degrees).


A
B
C

Early release of contractures (Bilateral).

(Rideay 1984)



*** When?.**

- Usually bet. 4-6 years.
- Timed Gower's is 5 Sec. or less.

*** Why?.**

_ Prolong walking ability for extra 2 years

(Riccio 1990).

- Delay hip contracture for 3 years

(Rideau etal. 1986).

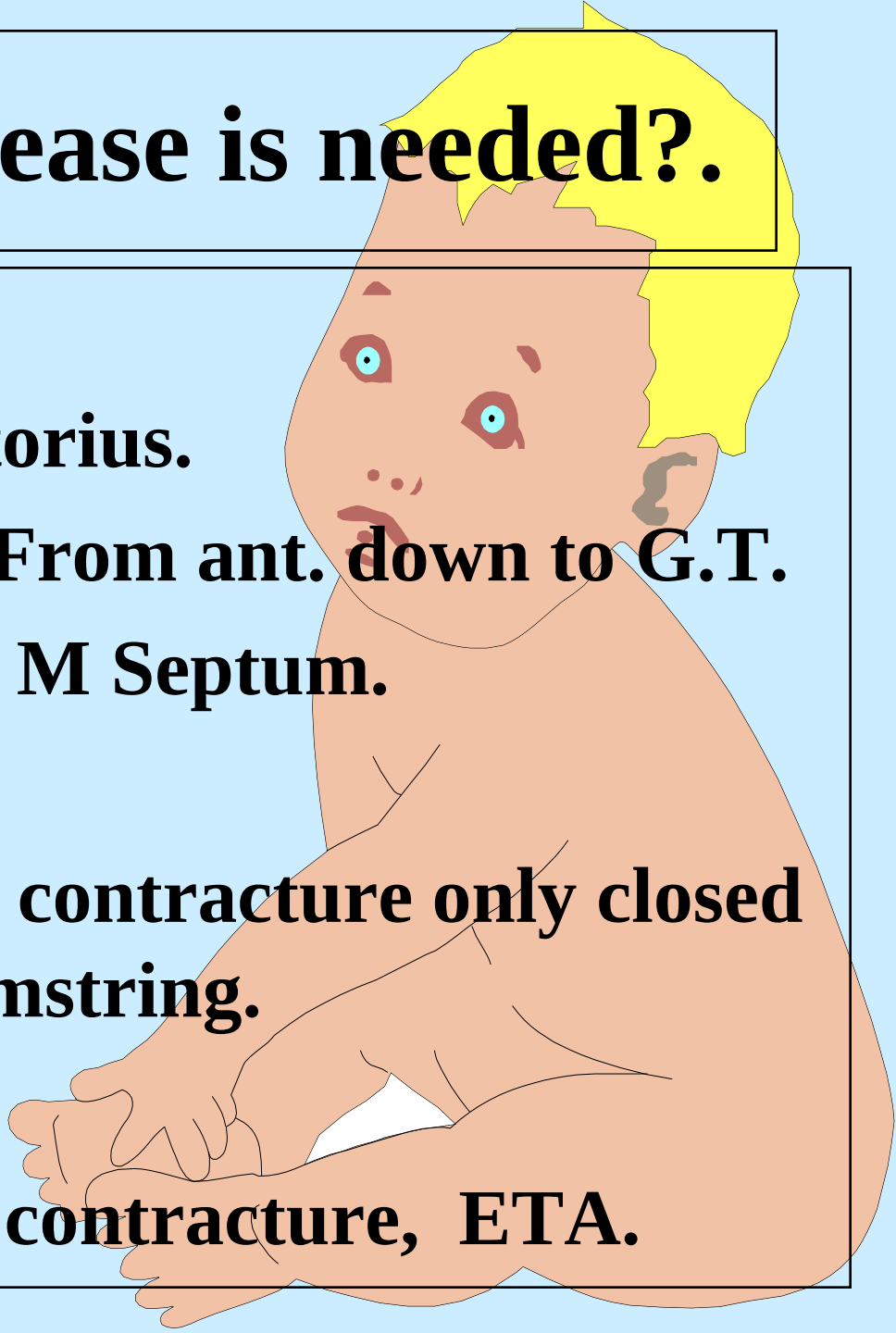
What early release is needed?.

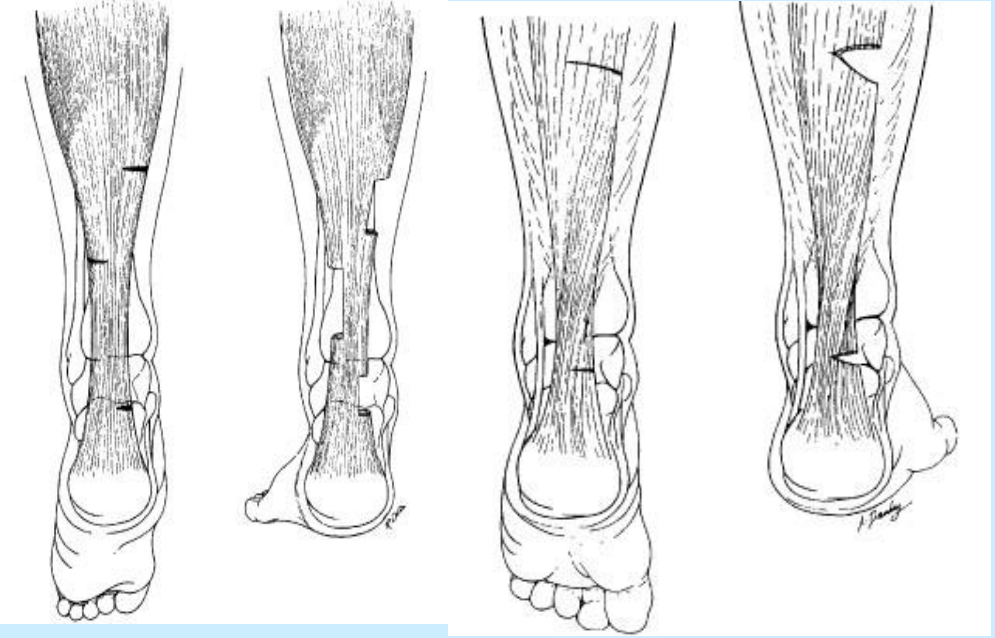
I. Hip

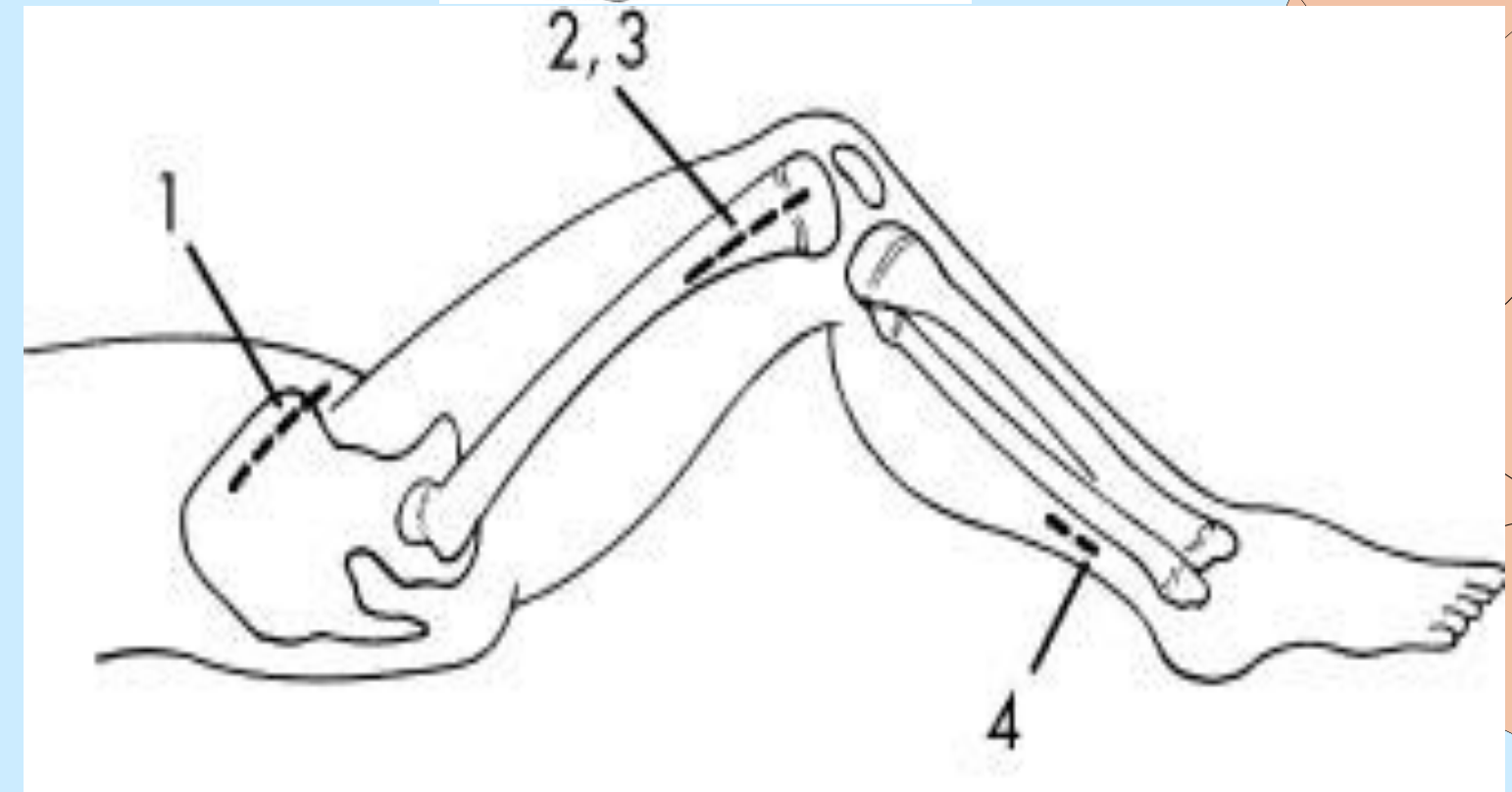
1. **division of TFL, Sartorius.**
2. **Division of F. Lata : From ant. down to G.T.**
3. **Resection of I TB + I M Septum.**

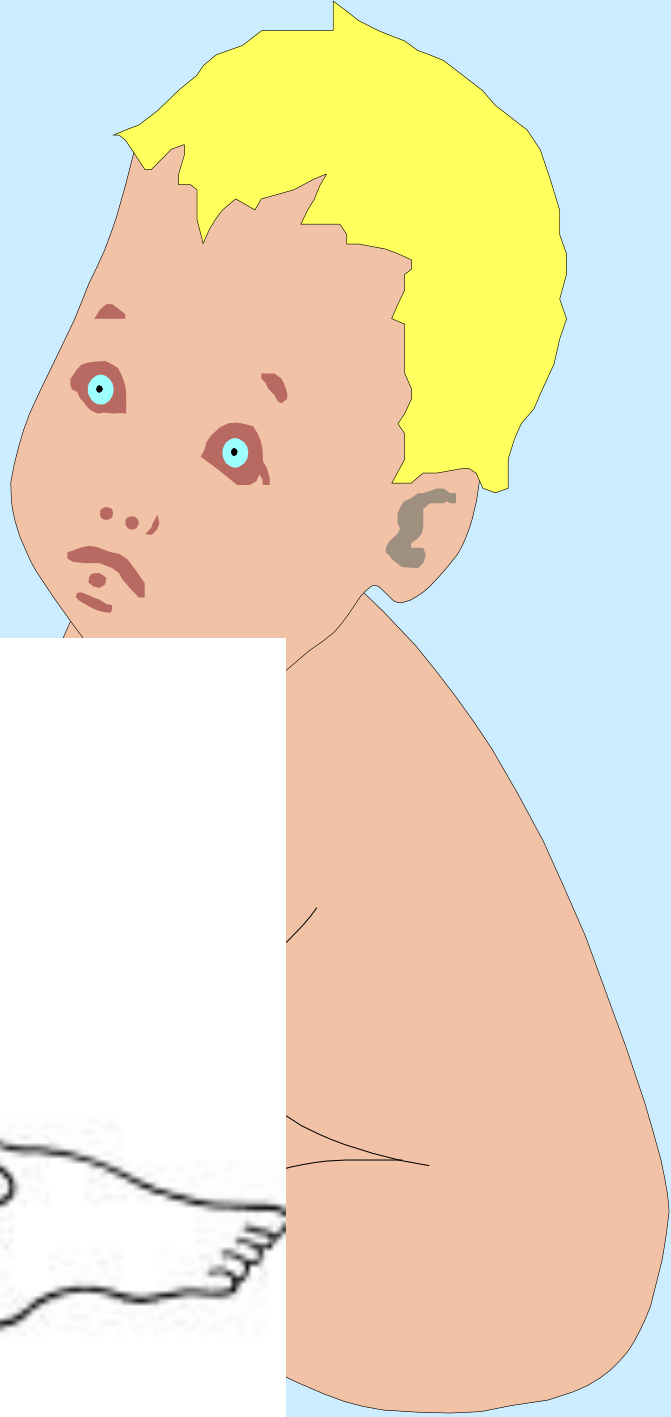
II Knee: **In Hamstring contracture only closed tenotomy of Med. Hamstring.**

III Ankle: **In early calf contracture, ETA.**

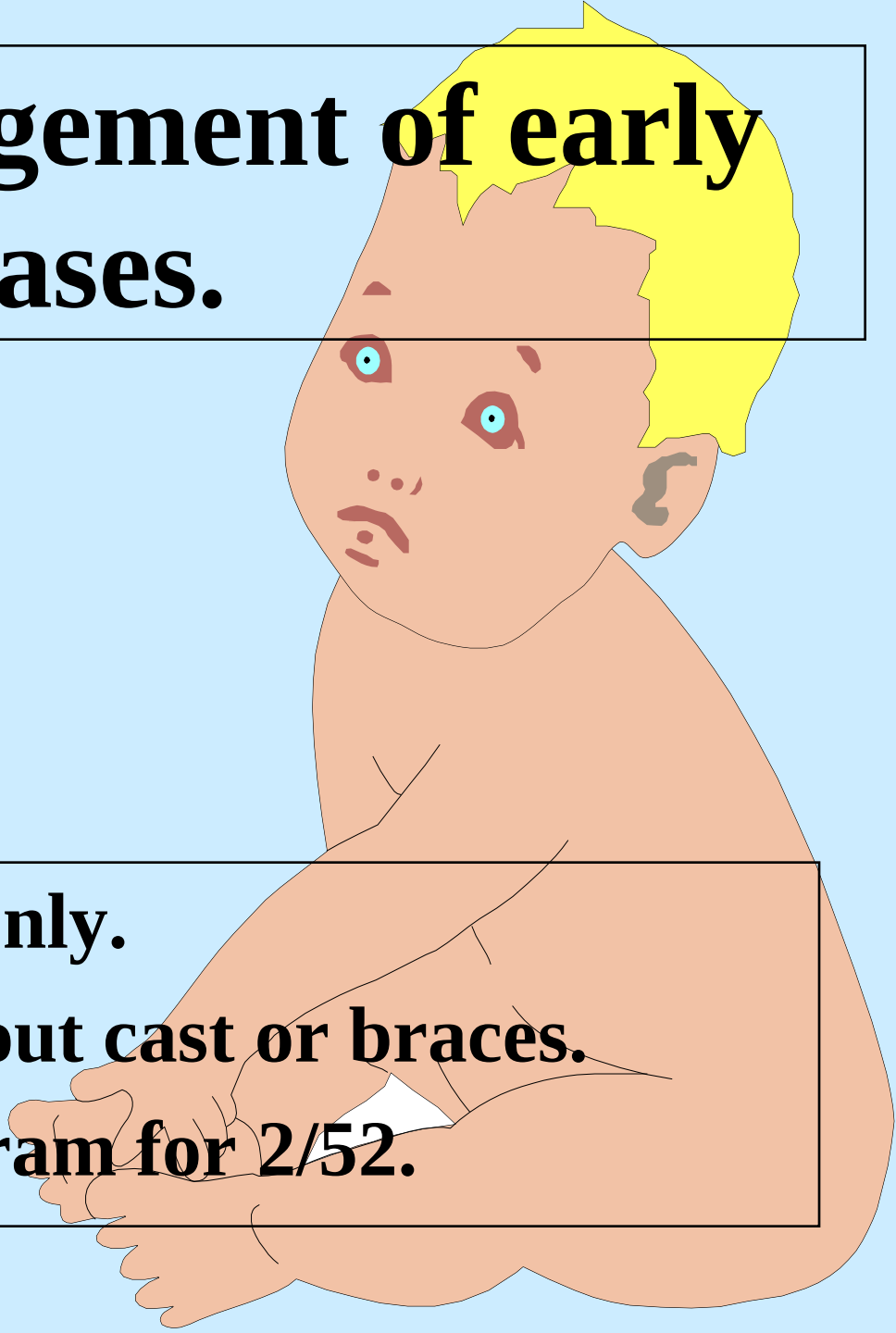





2, 3
1
4



Post op. management of early releases.



- Bed rest for 2 days only.
- Mobilize FWB without cast or braces.
- Physiotherapy program for 2/52.

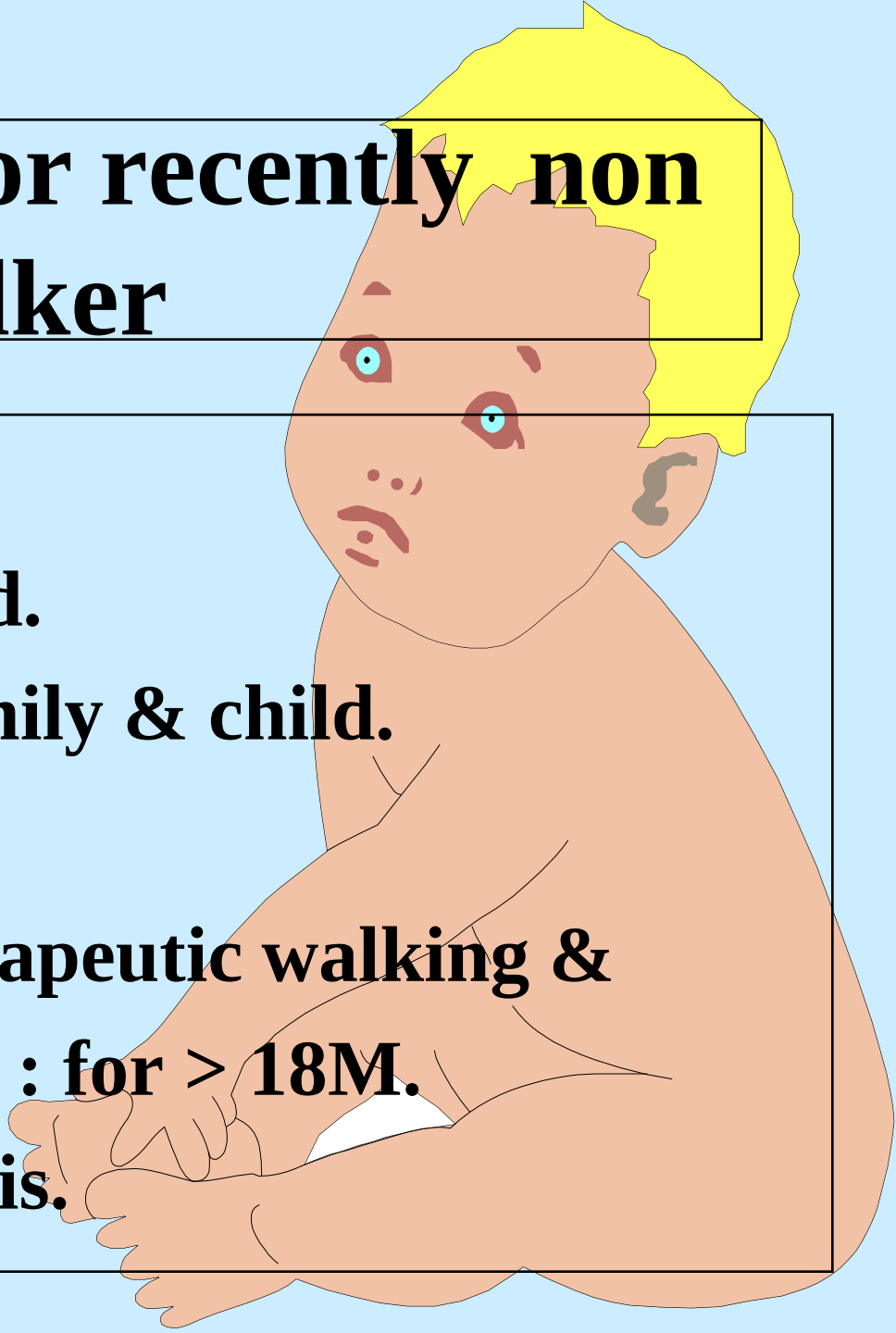
Late surgery for recently non walker

When?.

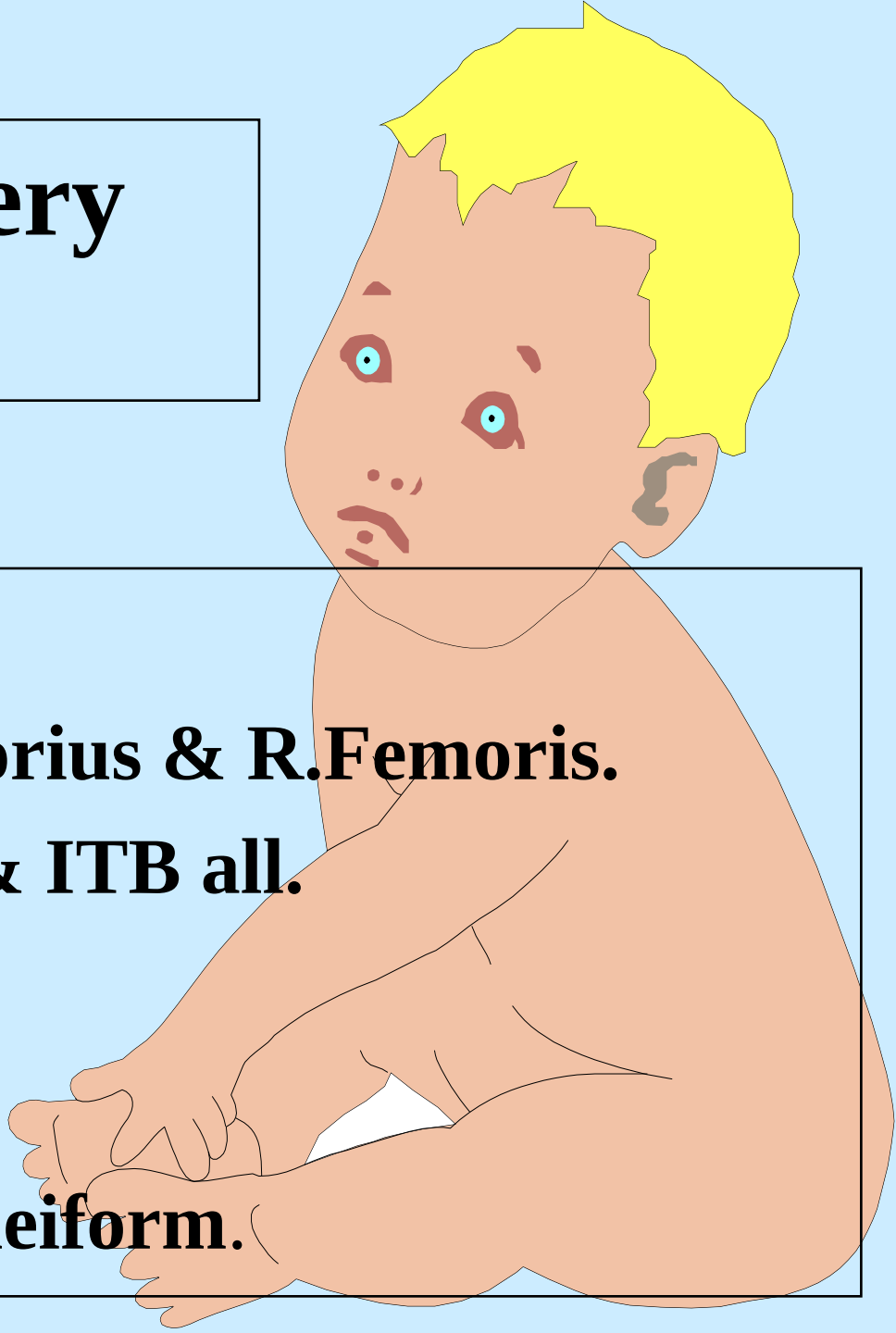
- **Non over weight child.**
- **Highly motivated family & child.**

why?

- **Bracing to allow therapeutic walking & limited independance : for > 18M.**
- **Delay onset of scoliosis.**



What late surgery needed?.



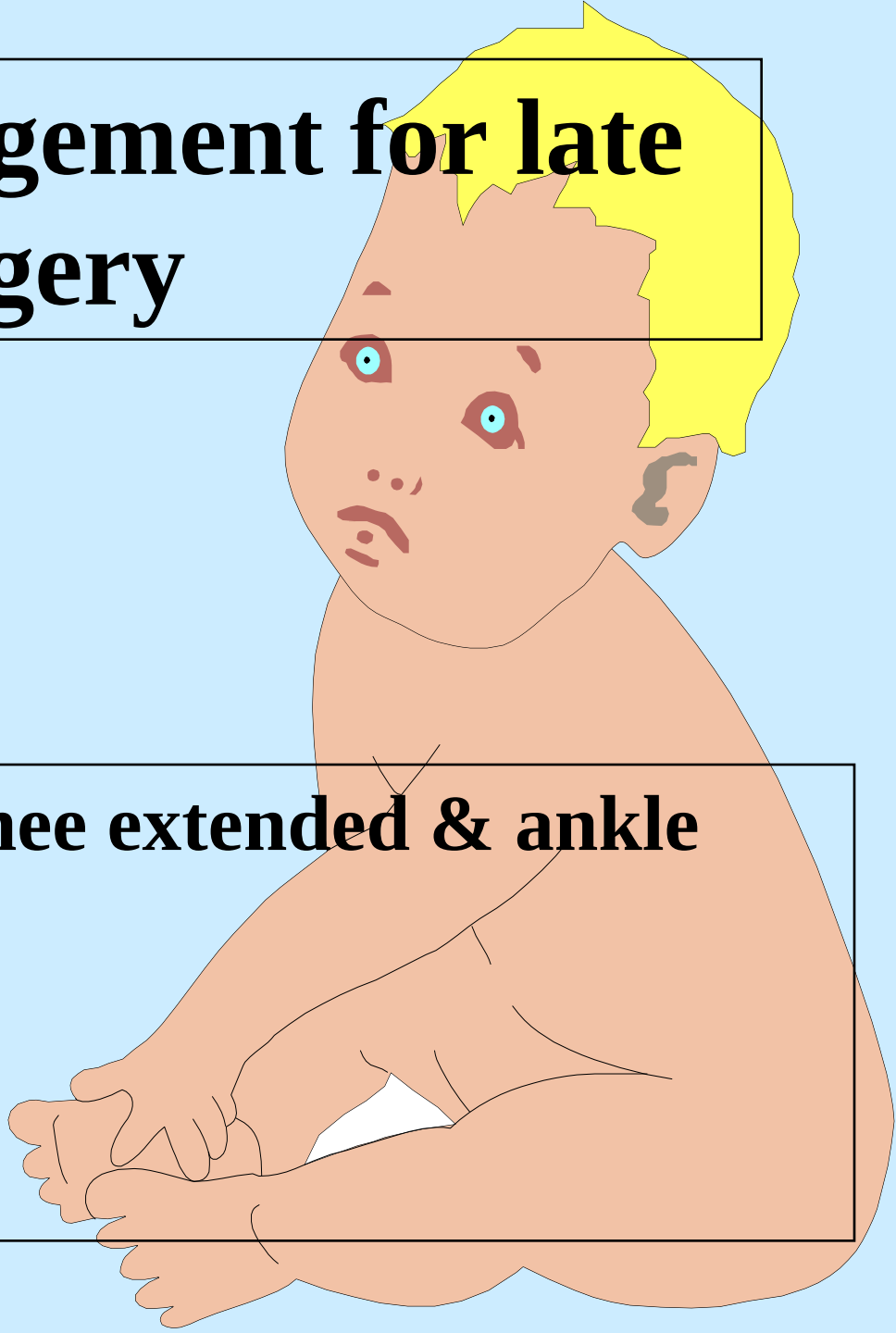
I. Hip

* Division of TFL, sartorius & R.Femoris.
* Resection of F. Lata & ITB all.

II. Ankle

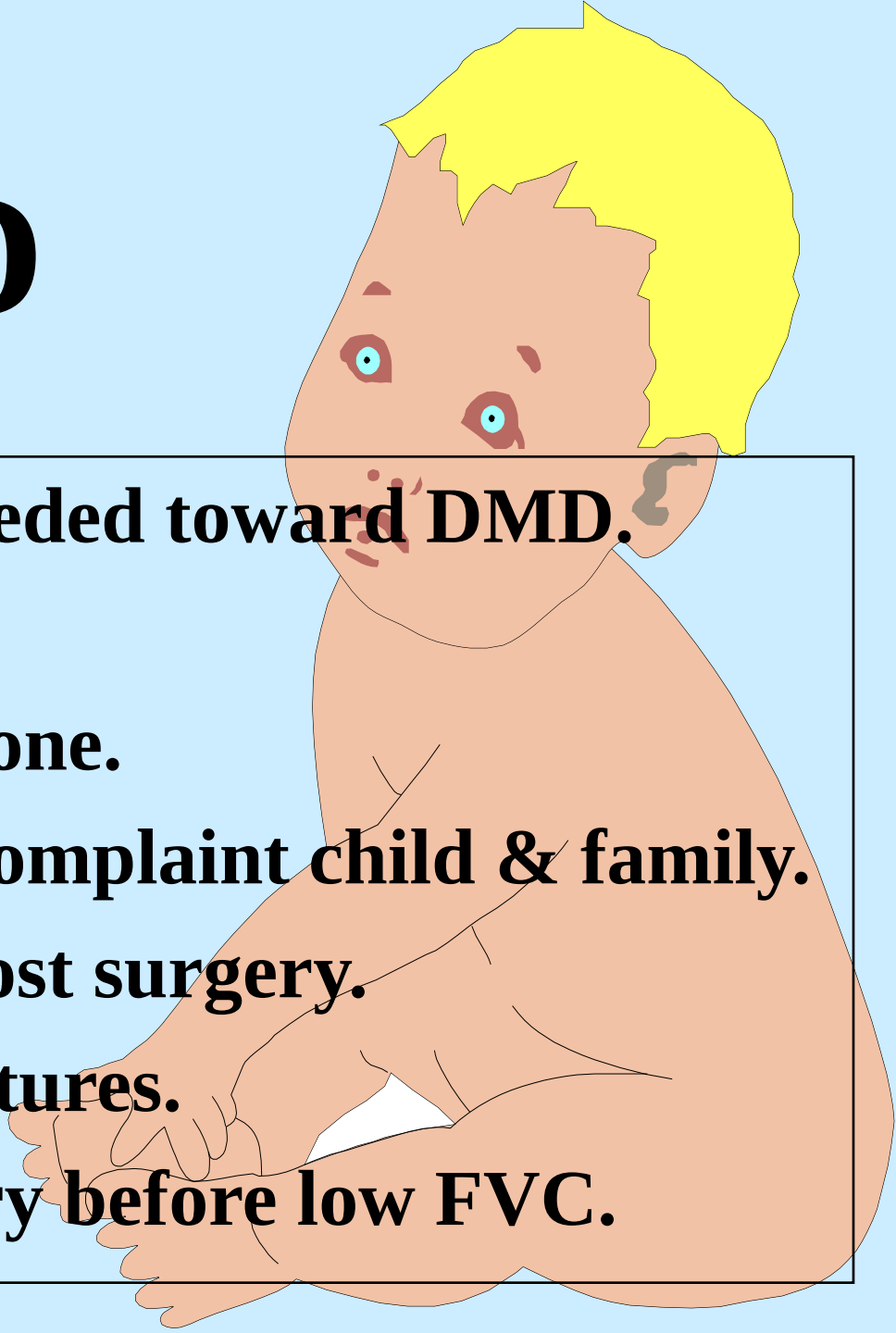
* ETA.
* T. Post to middle Cuneiform.

Post op. management for late surgery



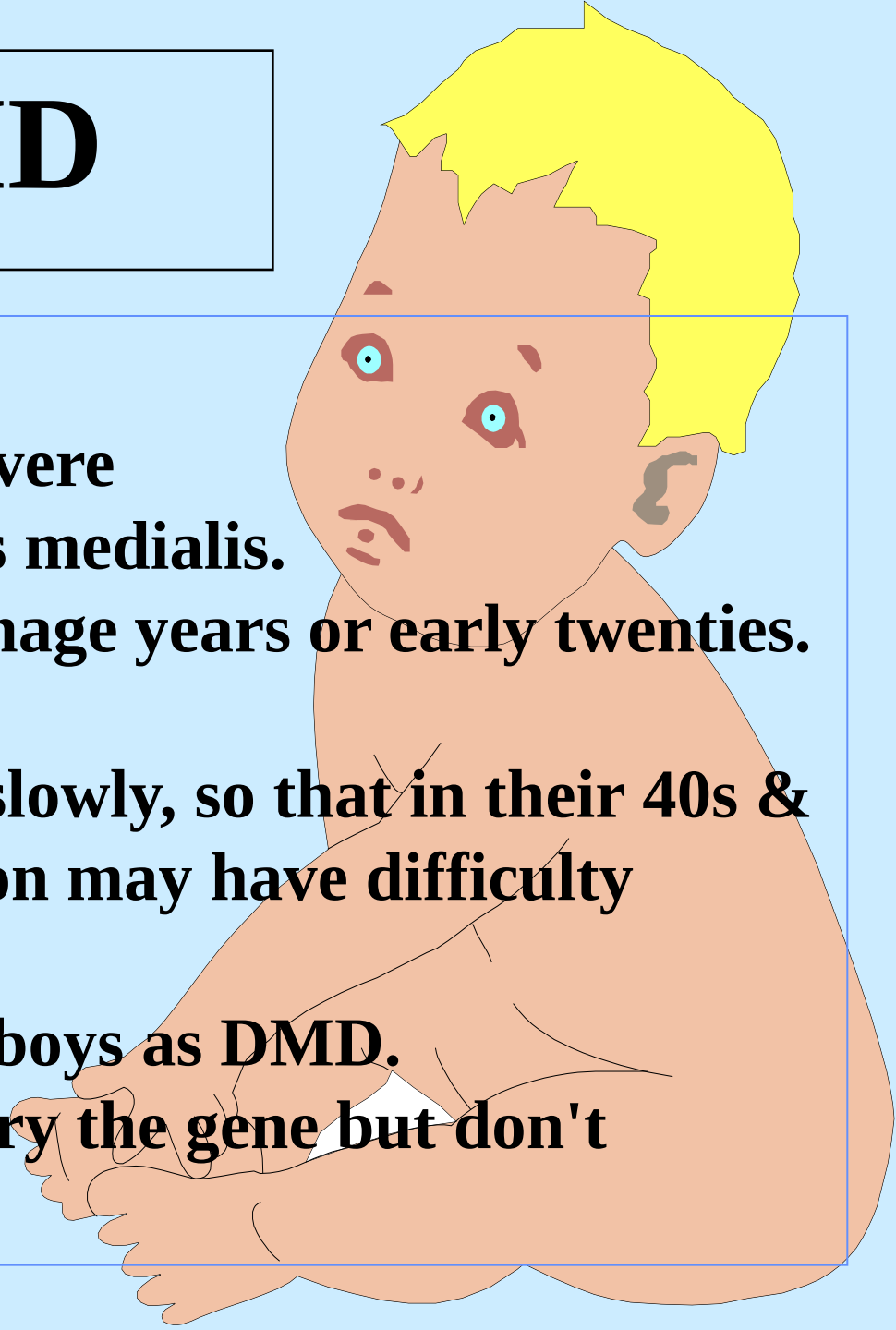
- Long leg cast with knee extended & ankle neutral.
- Early mobilization.
- KAFO before 4W.

DMD



1. High awareness is needed toward DMD.
2. Needle biopsy.
3. Never elongate TA alone.
4. Surgery helpful for complaint child & family.
5. Early mobilization post surgery.
6. Early fixation of fractures.
7. Early scoliosis surgery before low FVC.

Becker MD



As DMD Except

= Slowly progressive, less severe

= Selective wasting of vastus medialis.

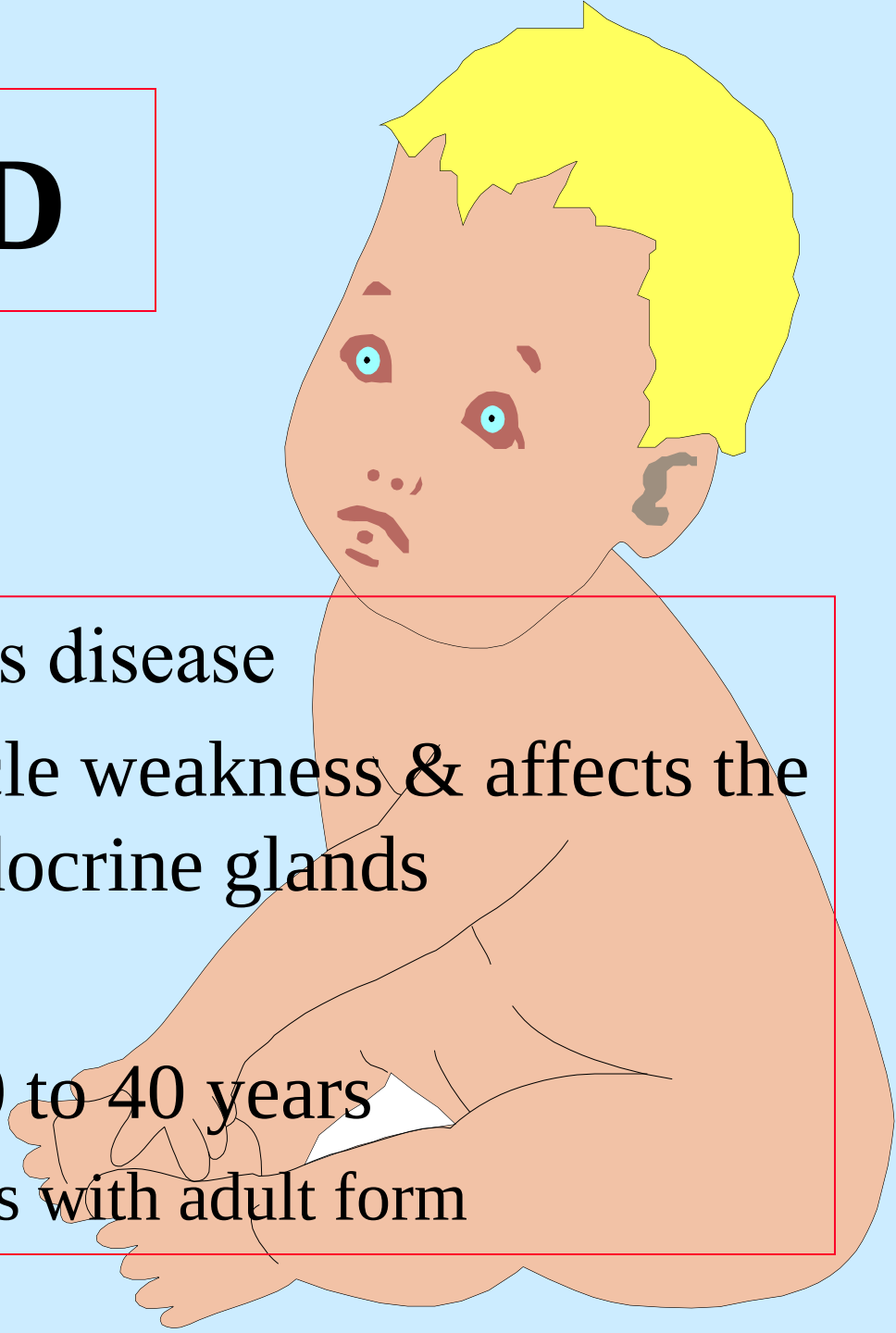
= Symptoms start in the teenage years or early twenties.

= The weakness progresses slowly, so that in their 40s & 50s, men with this condition may have difficulty walking.

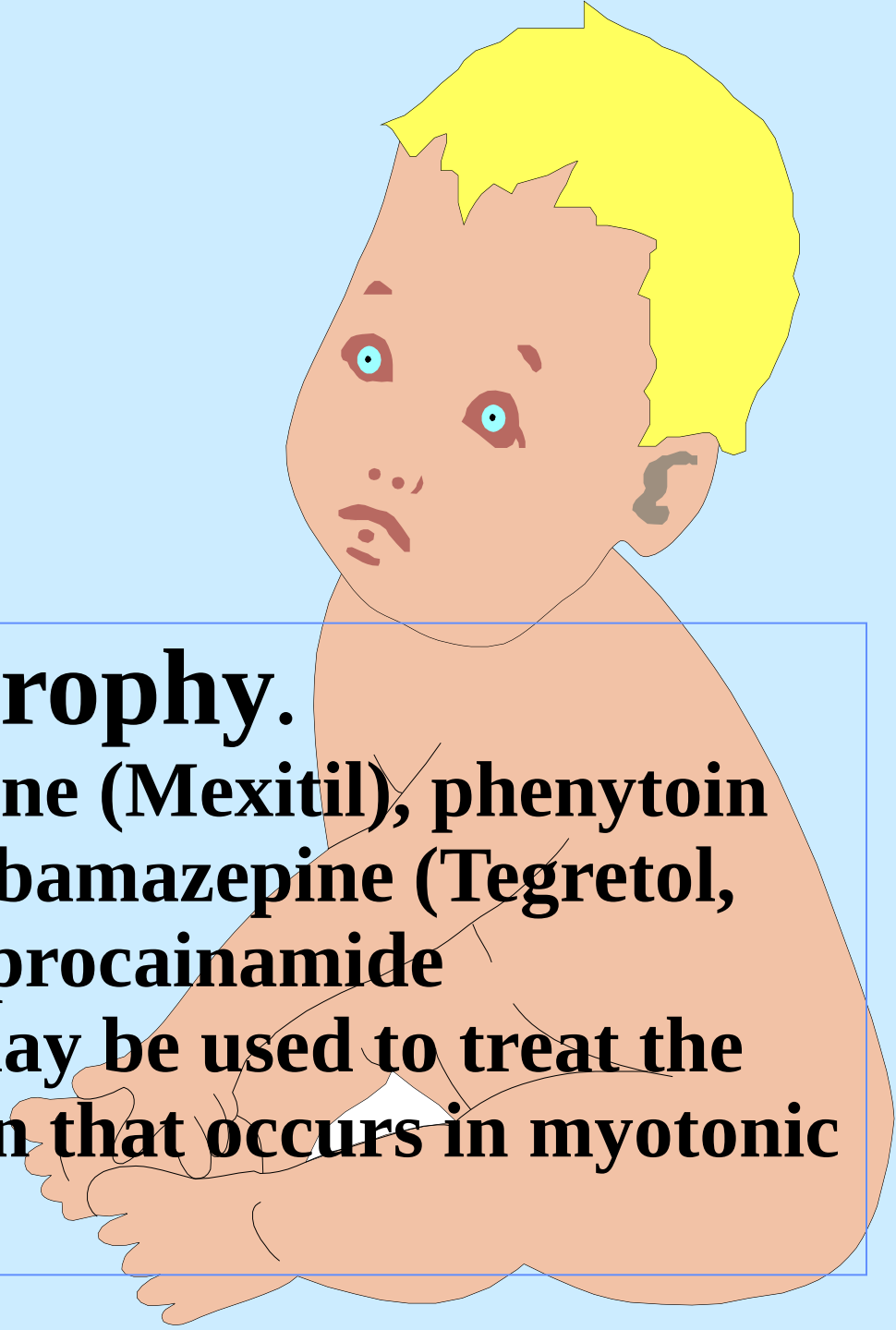
= BMD usually affects only boys as DMD.

= Girls and women may carry the gene but don't usually have symptoms.

Myotonic MD

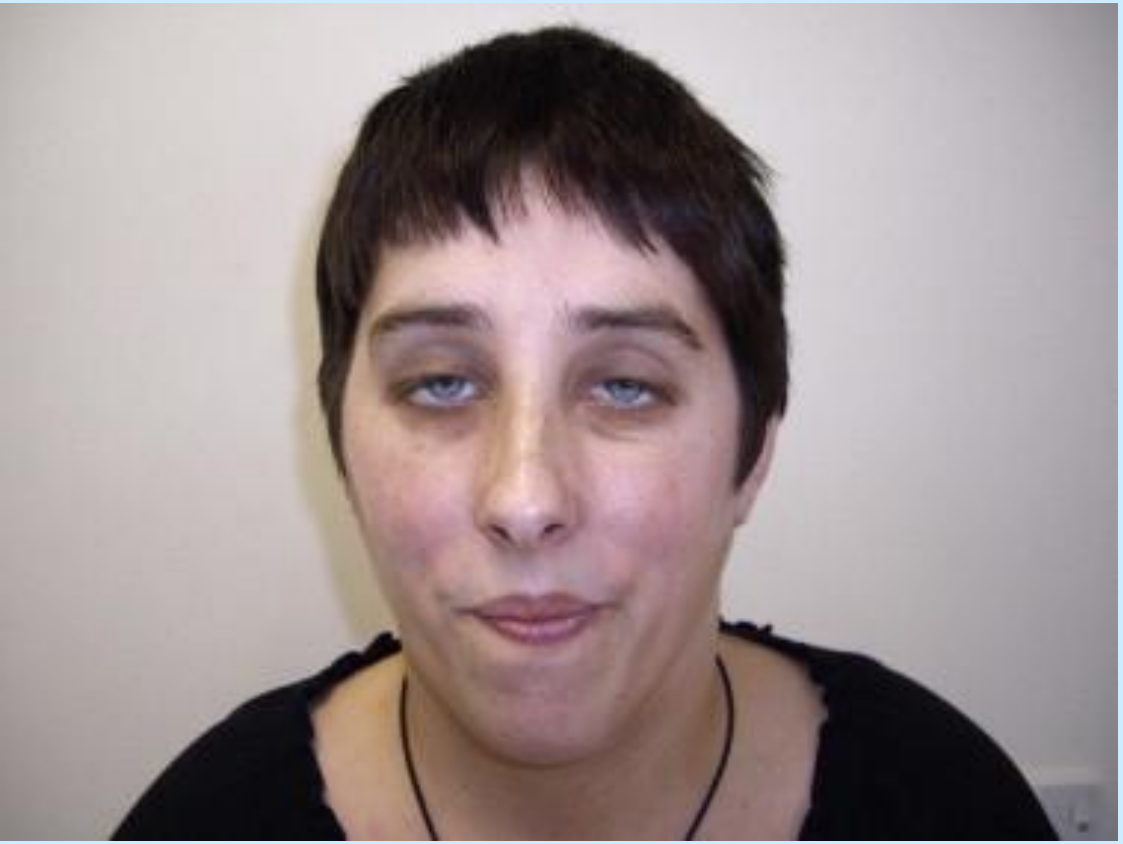


- Also known as Stienert's disease
- Manifests through muscle weakness & affects the CNS, heart, eyes, & endocrine glands
- Slowly progressive
- Occurs between ages 20 to 40 years
  - Congenital rare: mothers with adult form

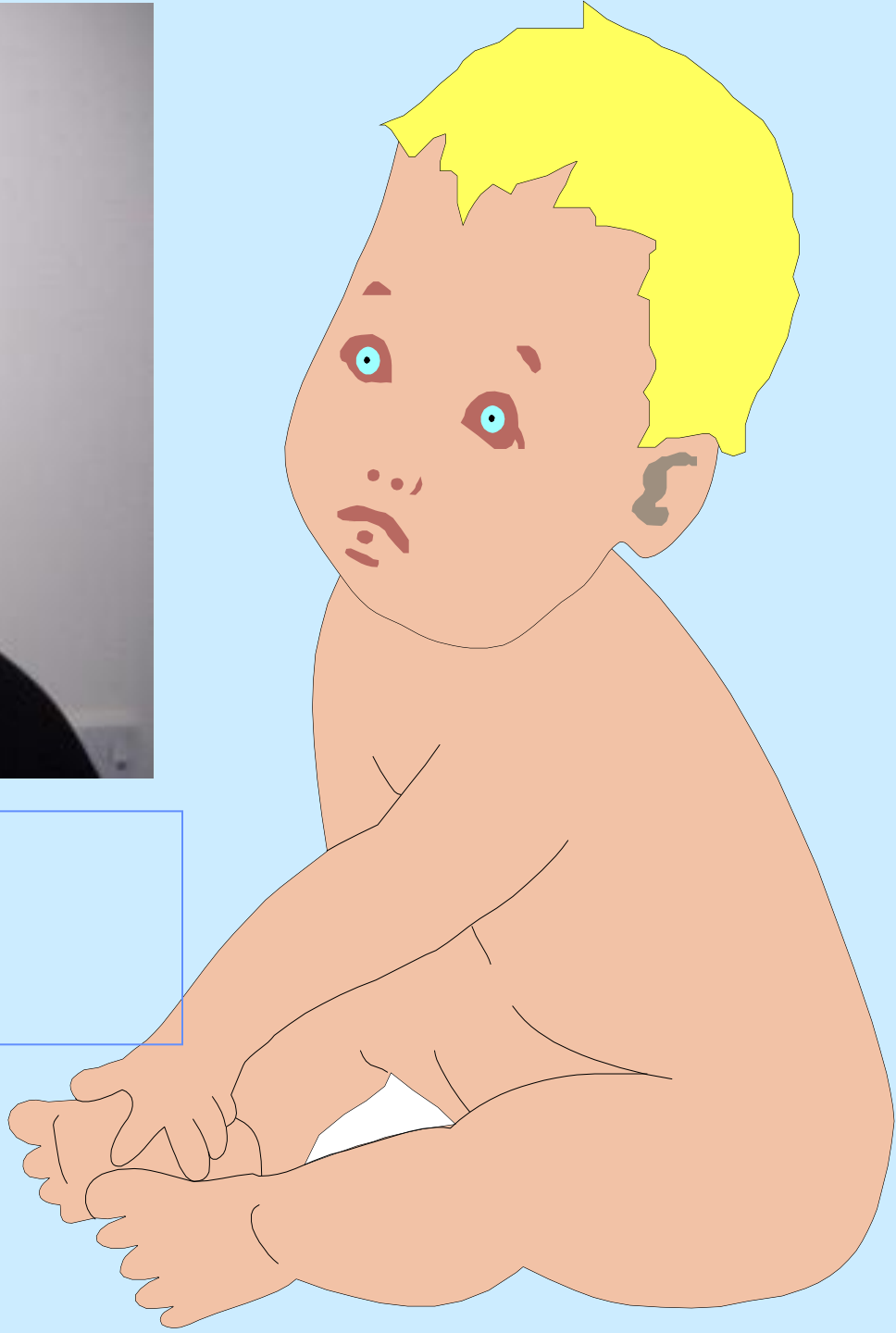


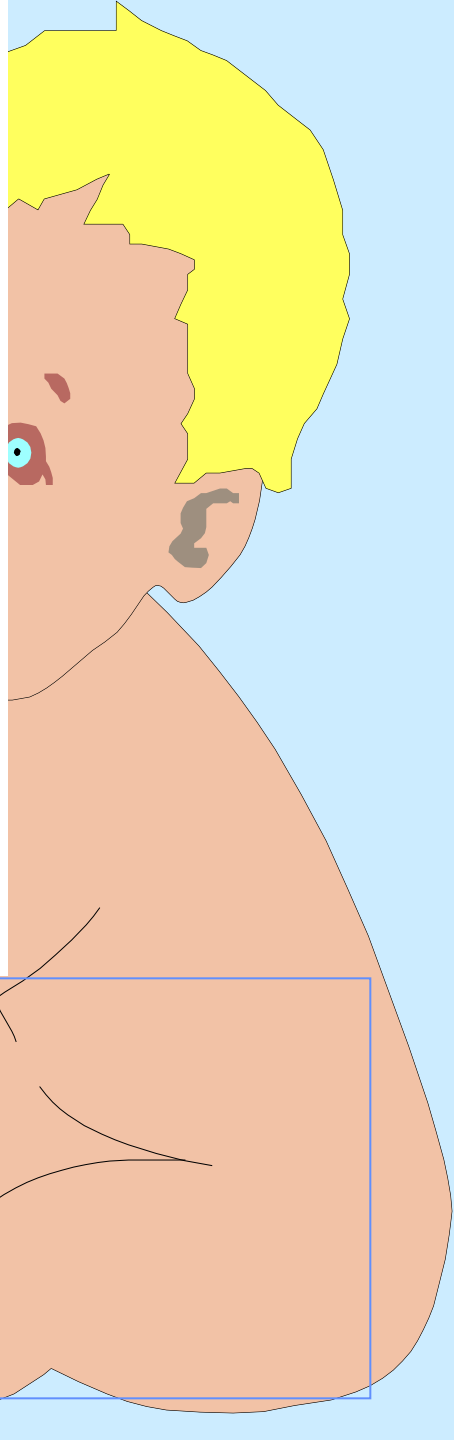
For myotonic dystrophy.

The medications mexiletine (Mexitil), phenytoin (Dilantin, Phenytek), carbamazepine (Tegretol, Carbatrol), quinine and procainamide (Procanbid, Pronestyl) may be used to treat the delayed muscle relaxation that occurs in myotonic dystrophy.



Myotonic dystrophy.
facial weakness, ptosis and temporalis atrophy

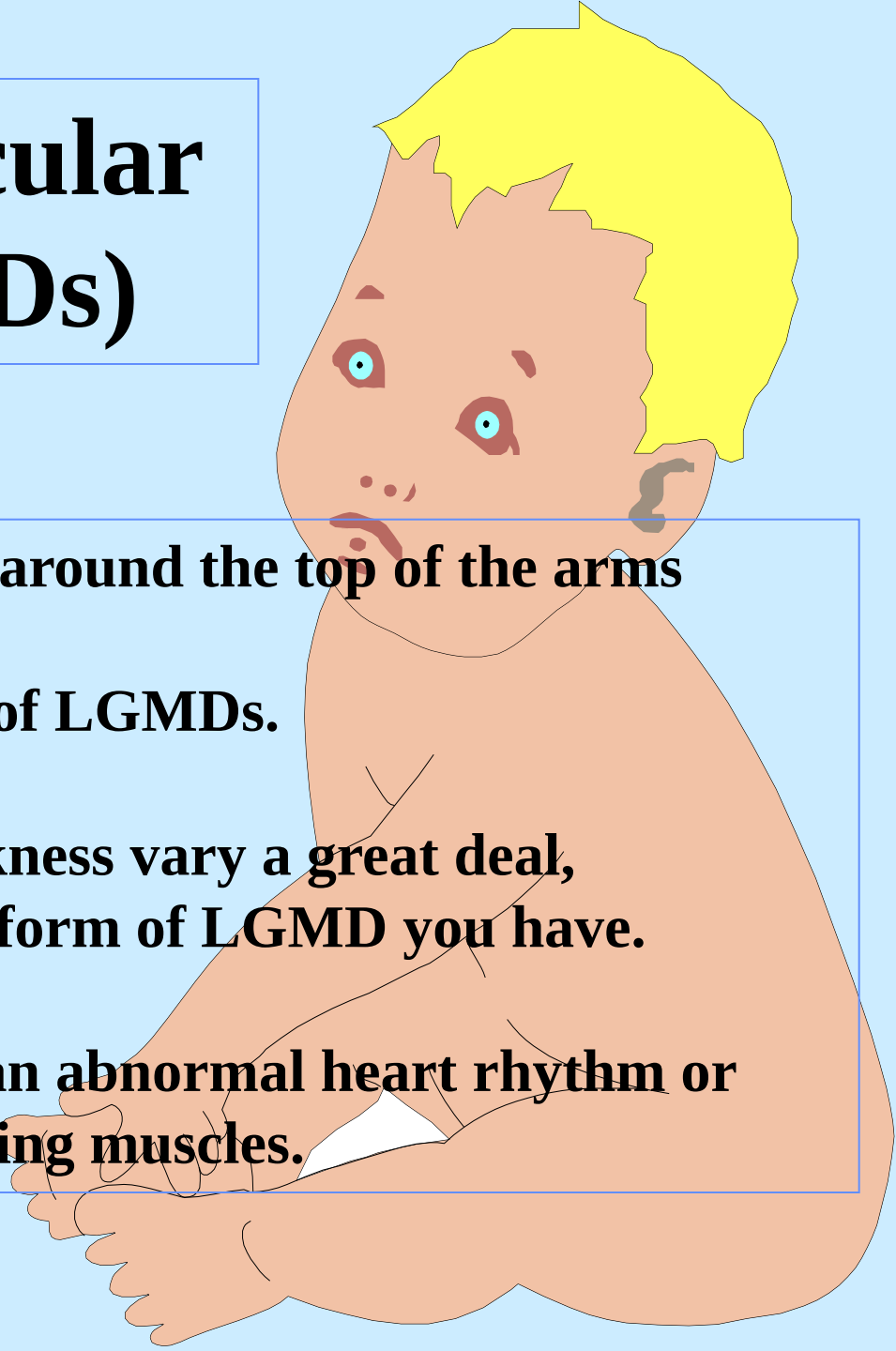




Myotonic dystrophy

Family with anticipation.
Grandmother with mild involvement.
Mother with myopathic face and mild weakness.
Child with congenital myotonic dystrophy.

Limb girdle muscular dystrophy (LGMDs)



= Cause weakness in the muscles around the top of the arms and lower limbs.

=There are many different types of LGMDs.

=They can affect men or women.

=The symptoms and muscle weakness vary a great deal, depending on which particular form of LGMD you have.

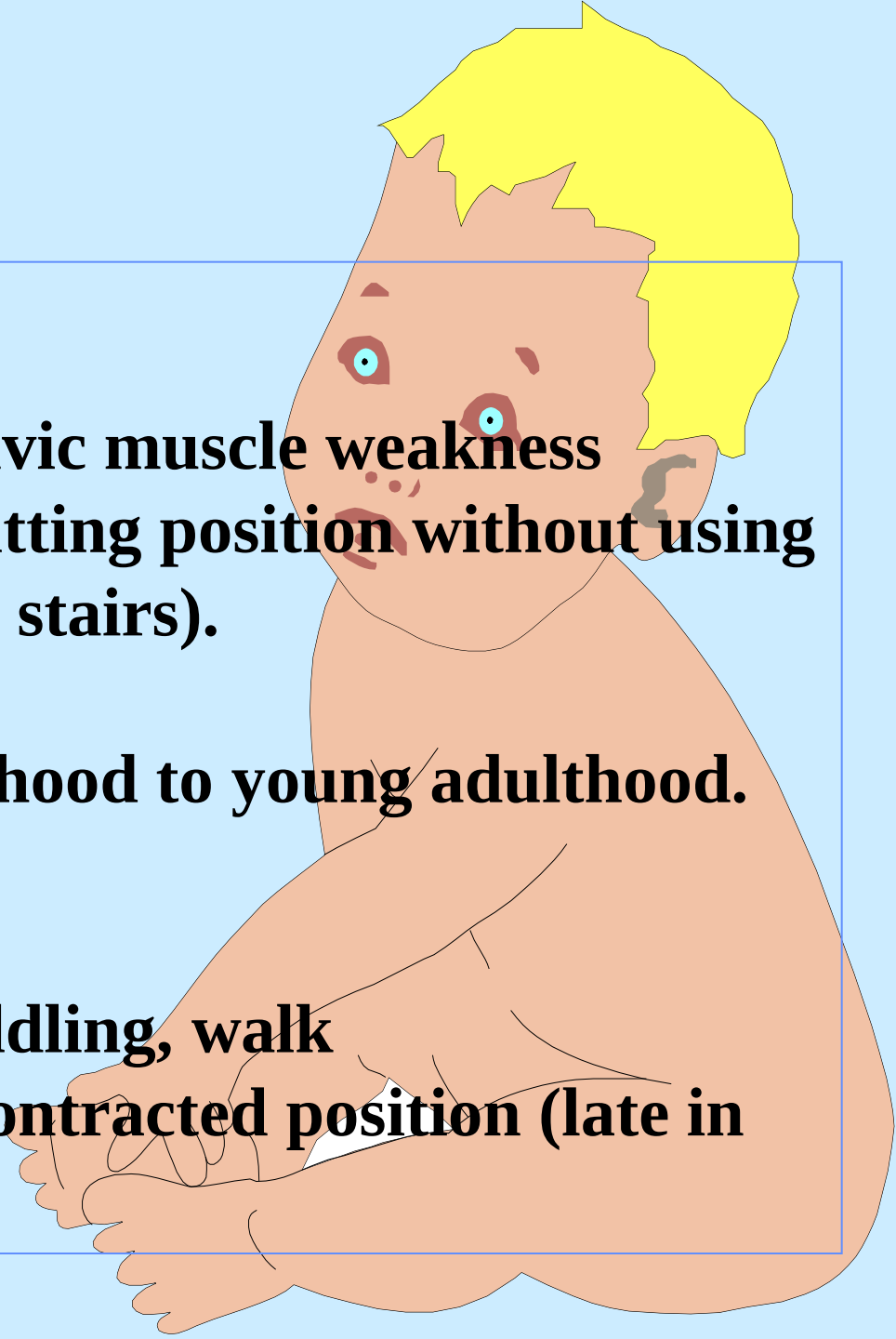
Some types of LGMD can cause an abnormal heart rhythm or weakness of the heart and breathing muscles.

Symptoms

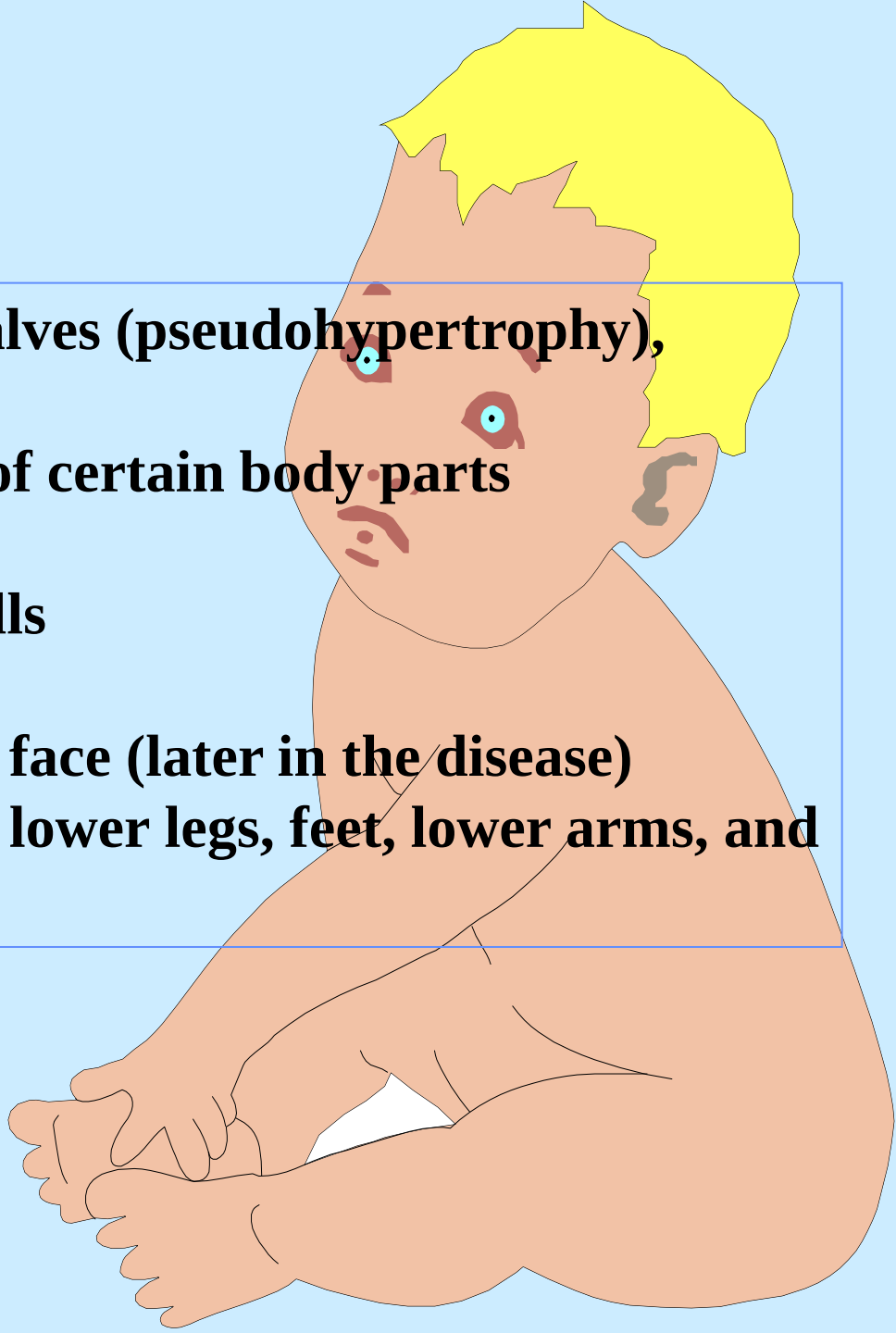
Typically, the first sign is pelvic muscle weakness (difficulty standing from a sitting position without using the arms, difficulty climbing stairs).

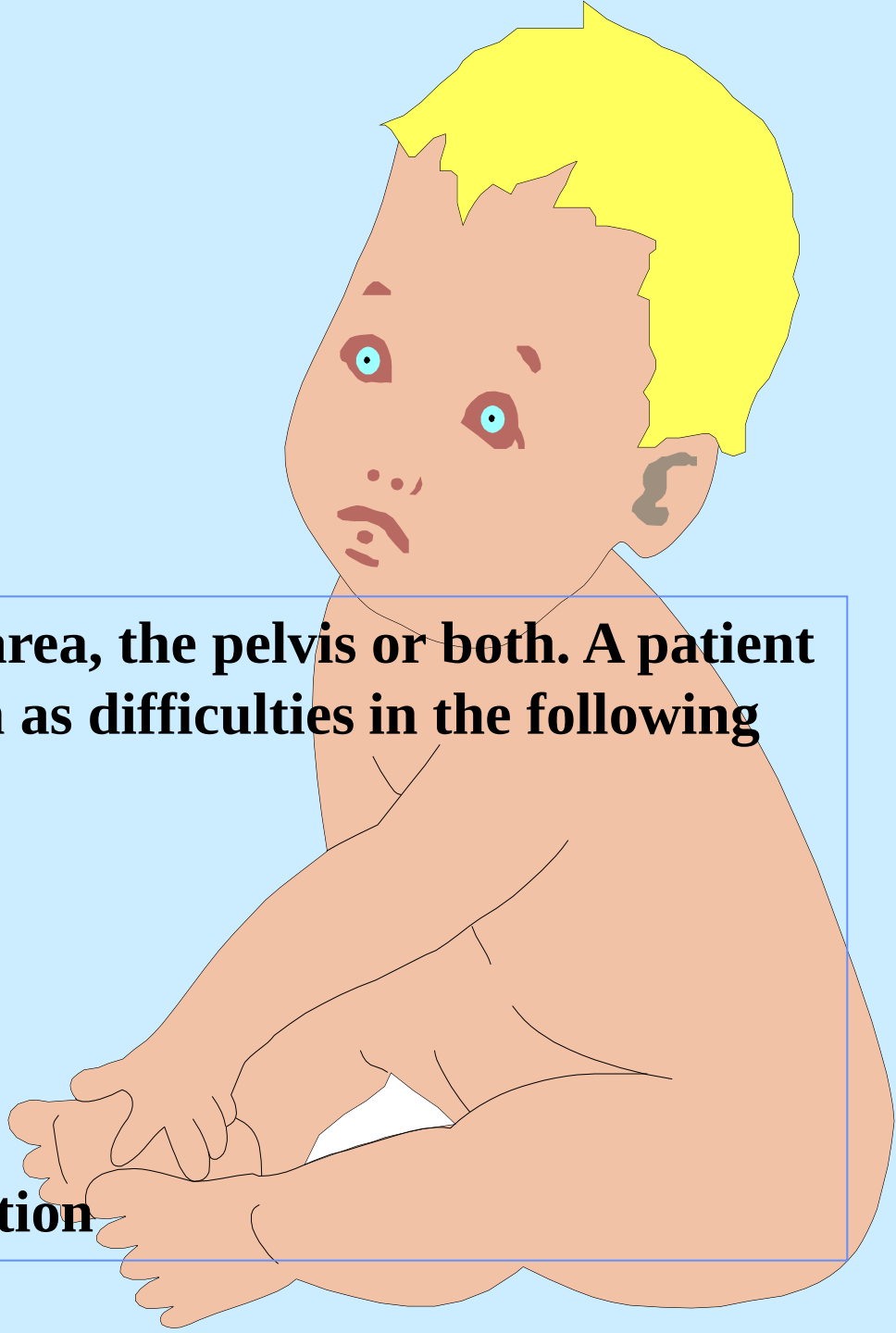
The weakness starts in childhood to young adulthood.

Other symptoms include:
1-Abnormal, sometimes waddling, walk
2-Joints that are fixed in a contracted position (late in the disease)



3-Large and muscular-looking calves (pseudohypertrophy), which are not actually strong
4-Loss of muscle mass, thinning of certain body parts
5-Low back pain
6-Palpitations or passing-out spells
7-Shoulder weakness
8-Weakness of the muscles in the face (later in the disease)
9-Weakness in the muscles of the lower legs, feet, lower arms, and hands (later in the disease)





LGMD may affect the shoulder area, the pelvis or both. A patient might experience symptoms such as difficulties in the following activities:

Walking

Running

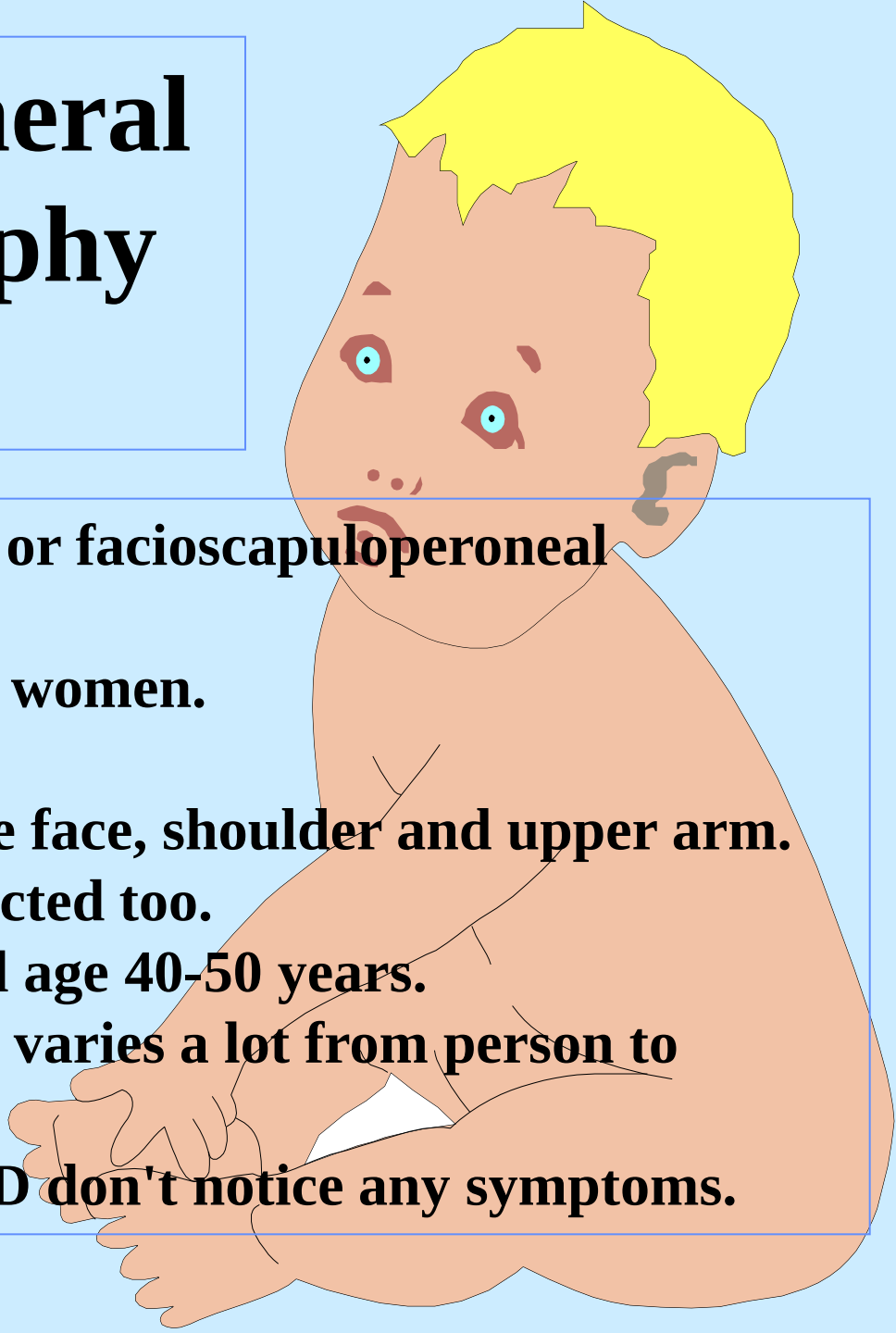
Going up and down the stairs

Getting up from the floor

Rising from a chair

Getting up from a squatting position

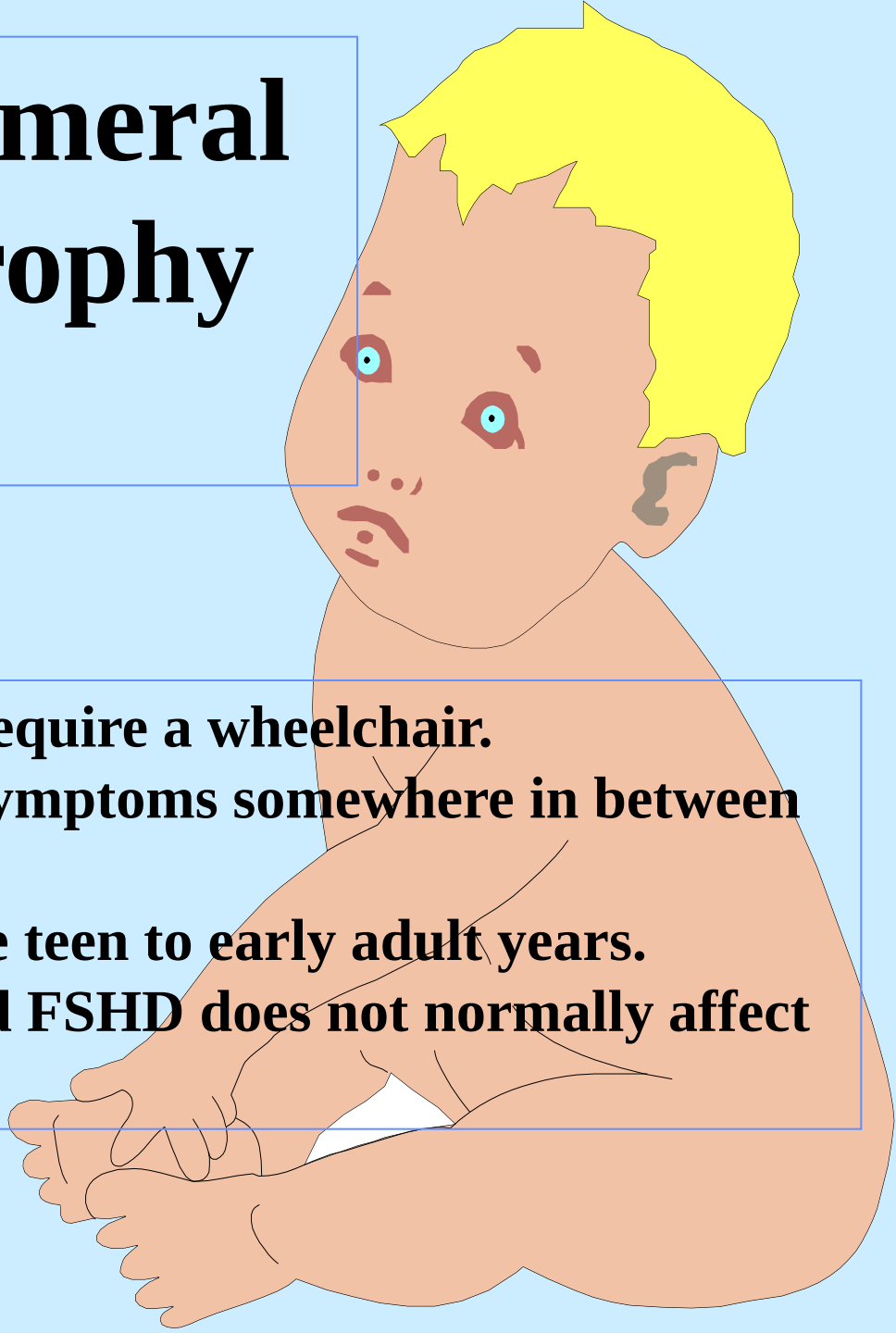
Facioscapulohumeral muscular dystrophy (FSHD)



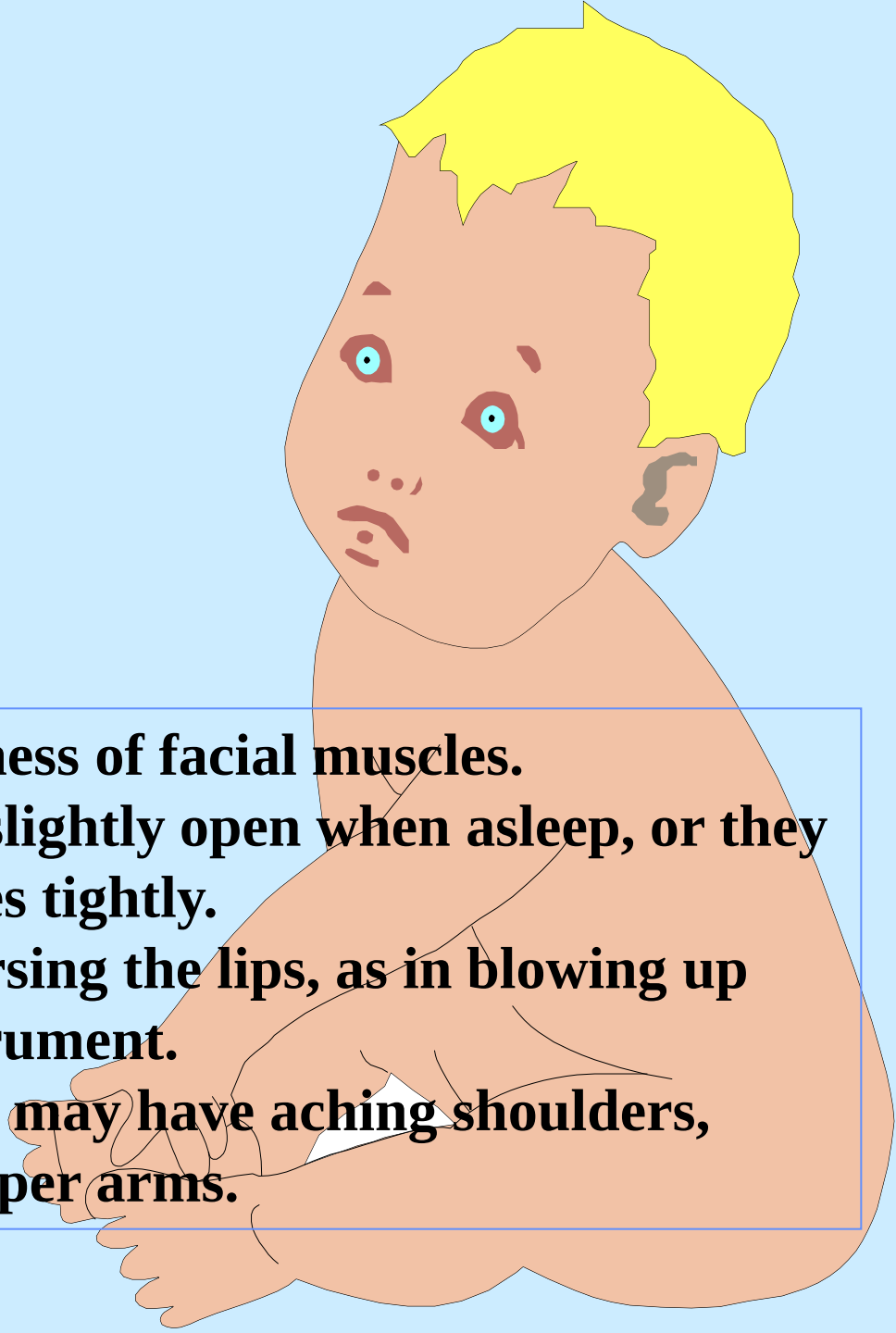
= Also called Landouzy-Dejerine or facioscapuloperoneal muscular dystrophy.
= FSHD can affect both men and women.

= FSHD affects the muscles of the face, shoulder and upper arm.
= Sometimes the legs may be affected too.
= Symptoms usually start around age 40-50 years.
= The degree of muscle weakness varies a lot from person to person.
= About 3 in 10 people with FSHD don't notice any symptoms.

Facioscapulohumeral muscular dystrophy (FSHD)

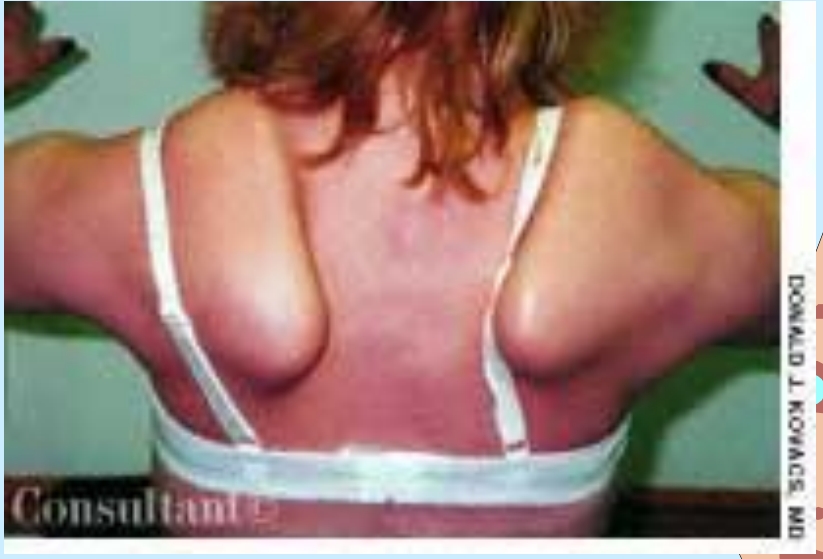


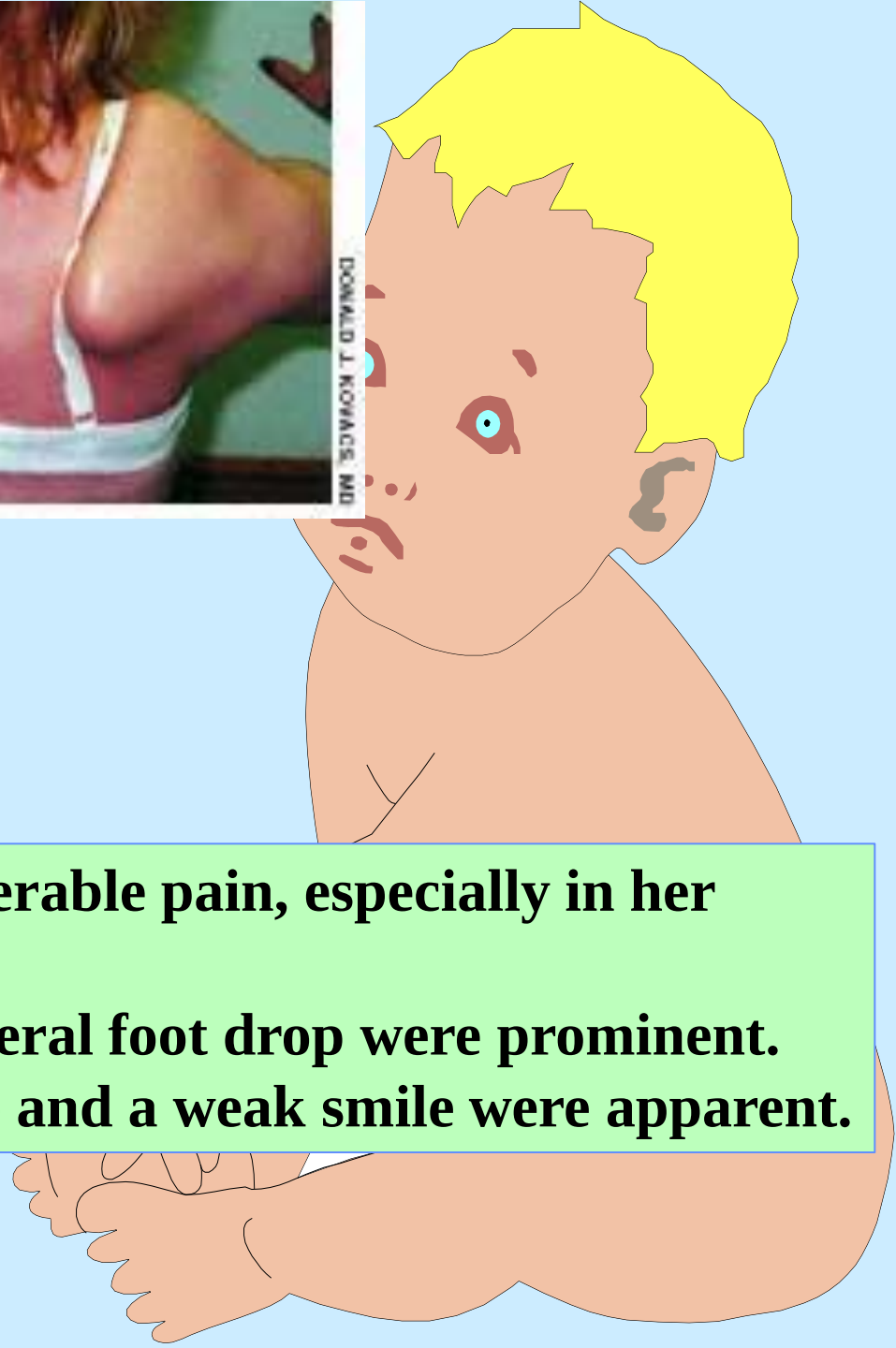
= About 10% people eventually require a wheelchair.
= Most people with FSHD have symptoms somewhere in between these two extremes.
= Onset usually occurs during the teen to early adult years.
= Overall, the outlook is good and FSHD does not normally affect a person's lifespan.



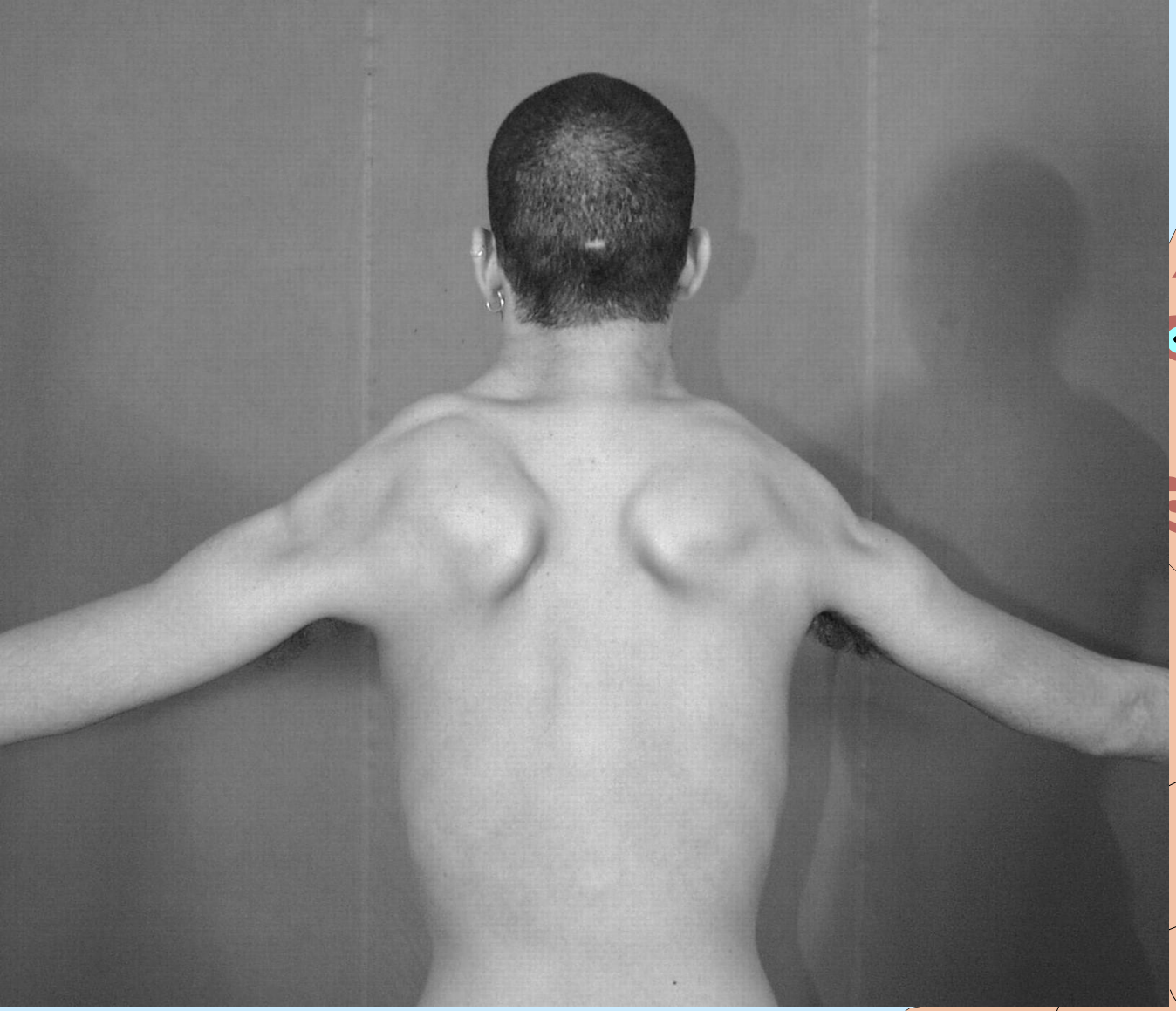
= FSHD may be noticed as weakness of facial muscles.
= The person's eyes may remain slightly open when asleep, or they may be unable to close their eyes tightly.
= They may have difficulty in pursing the lips, as in blowing up balloons or playing a wind instrument.
= Teenagers or adults with FSHD may have aching shoulders, rounded shoulders and thin upper arms.

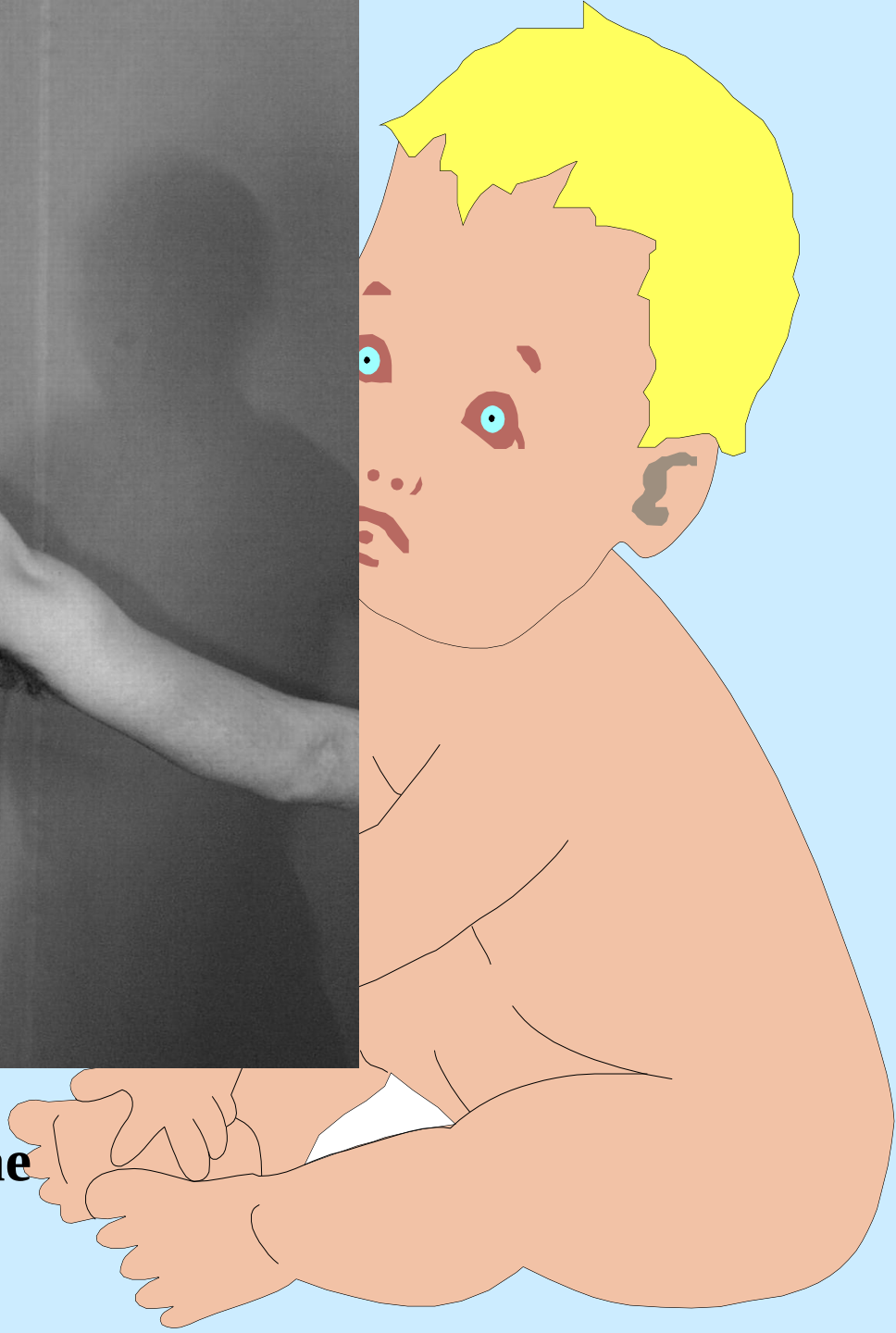
The patient is trying to abduct her arms to shoulder height but is unable to do so, not because of deltoid weakness but because of scapular instability – the ‘humps’ in the region of trapezius are the tops of her scapulae, riding upwards. Note that the facial weakness is not marked.





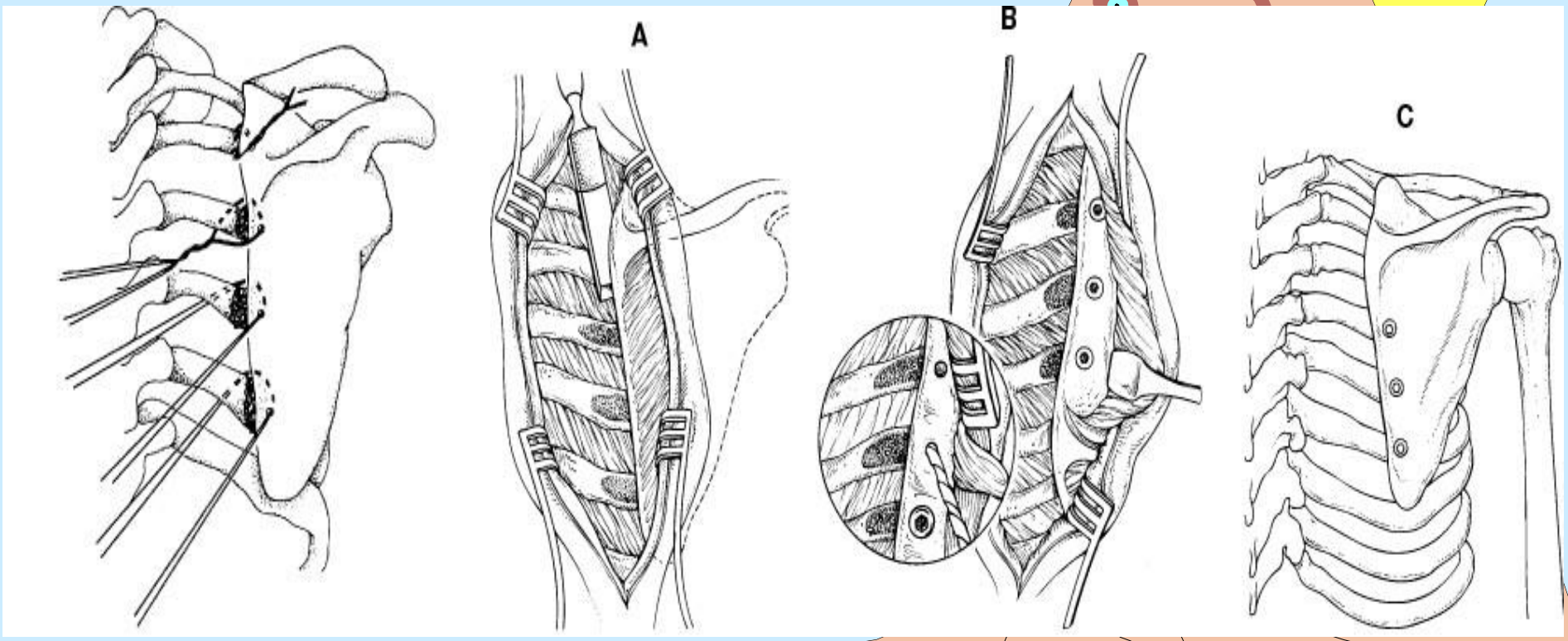
The patient complained of considerable pain, especially in her shoulders and hips.
Winging of the scapulae and bilateral foot drop were prominent.
Moderate weakness of eye closure and a weak smile were apparent.





Winging of the scapulae

Scapulothoracic Fusion

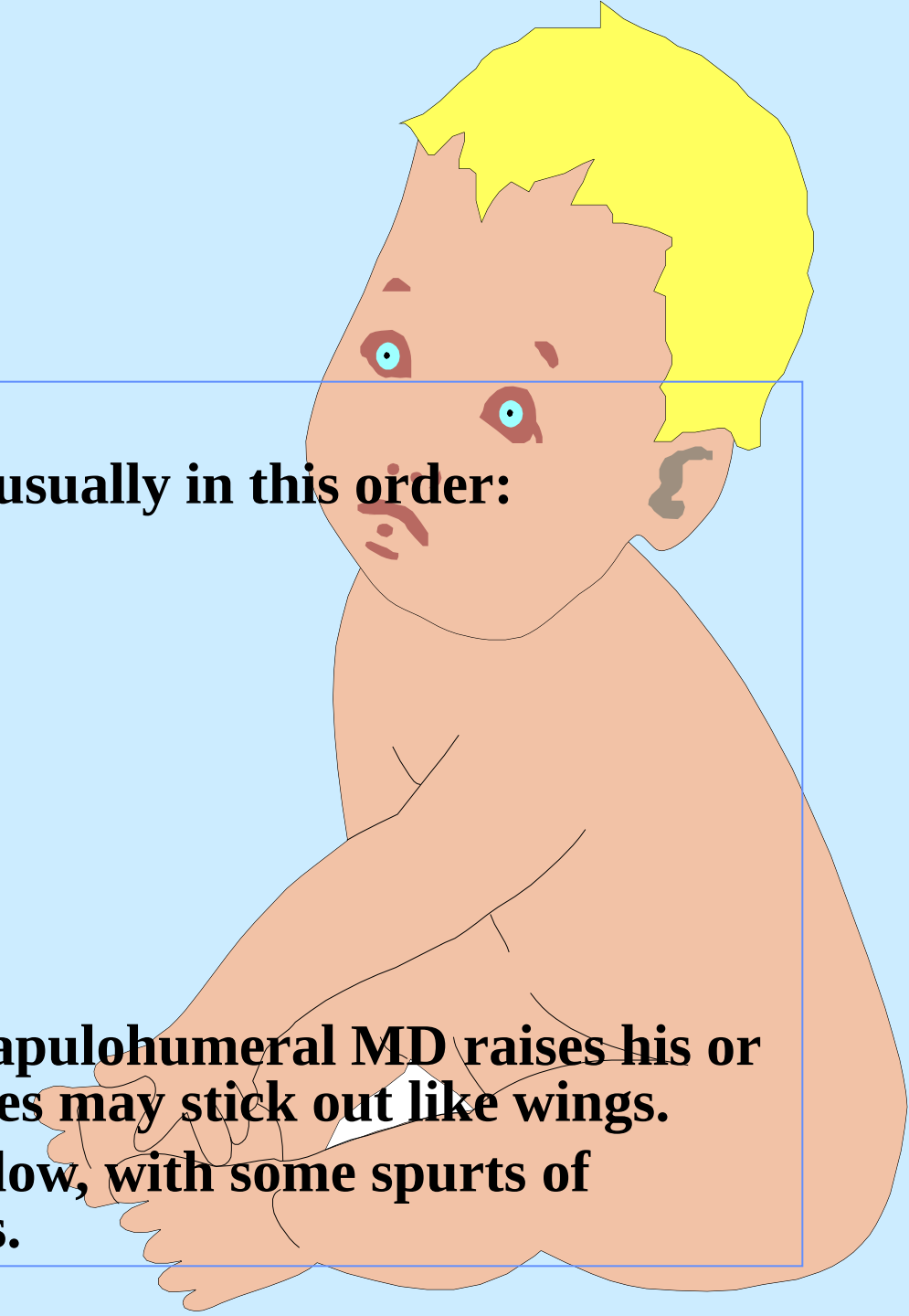


Progressive muscle weakness, usually in this order:

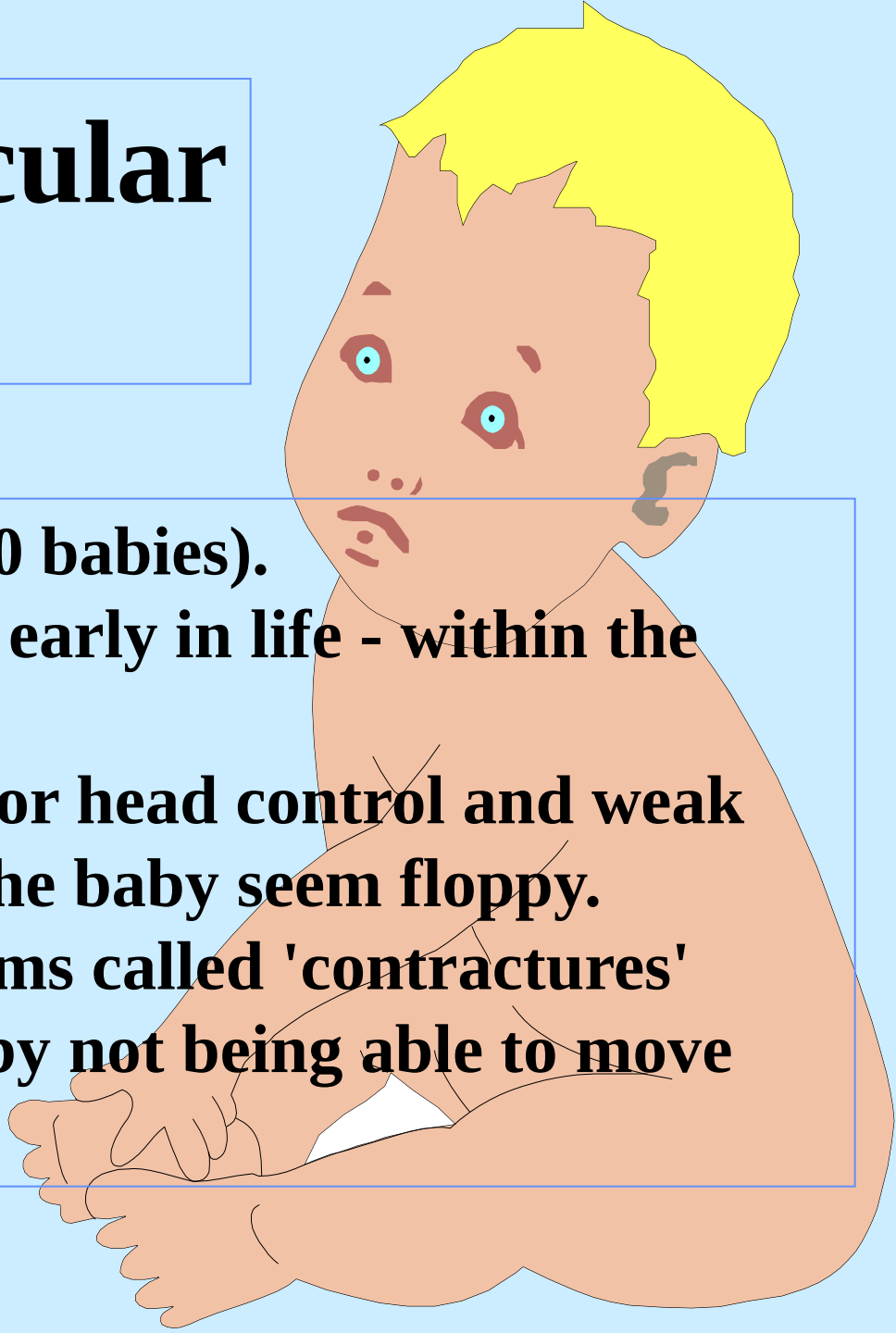
- **Face**
- **Shoulders**
- **Abdomen**
- **Feet**
- **Upper arms**
- **Pelvic area**
- **Lower arms**

When someone with facioscapulohumeral MD raises his or her arms, the shoulder blades may stick out like wings.

Progression of this form is slow, with some spurts of rapidly increasing weakness.



Congenital muscular dystrophy



= (affecting about 1 in 50,000 babies).

= It causes muscle weakness early in life - within the first six months of birth.

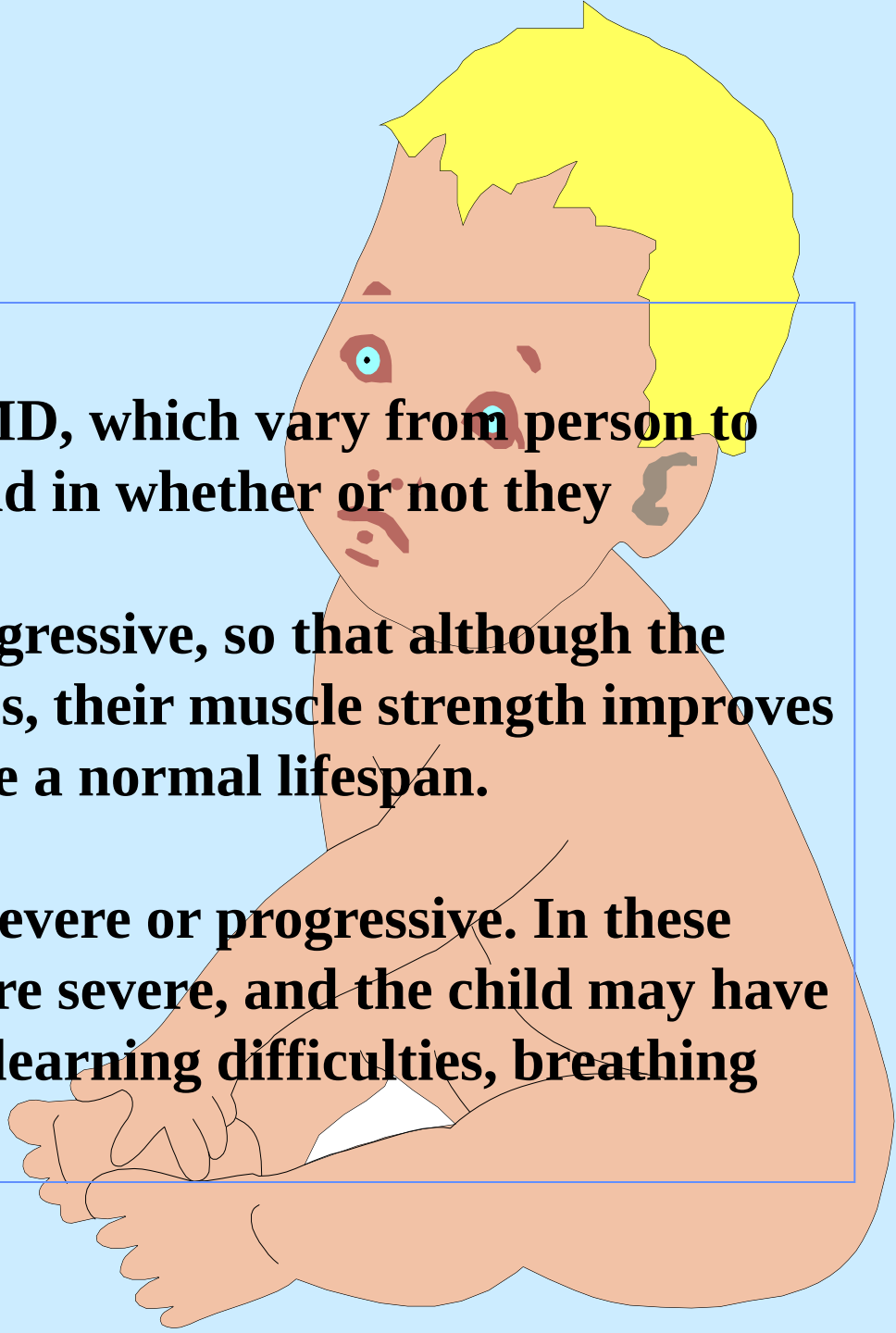
= The first symptoms are poor head control and weak muscles, which make the the baby seem floppy.

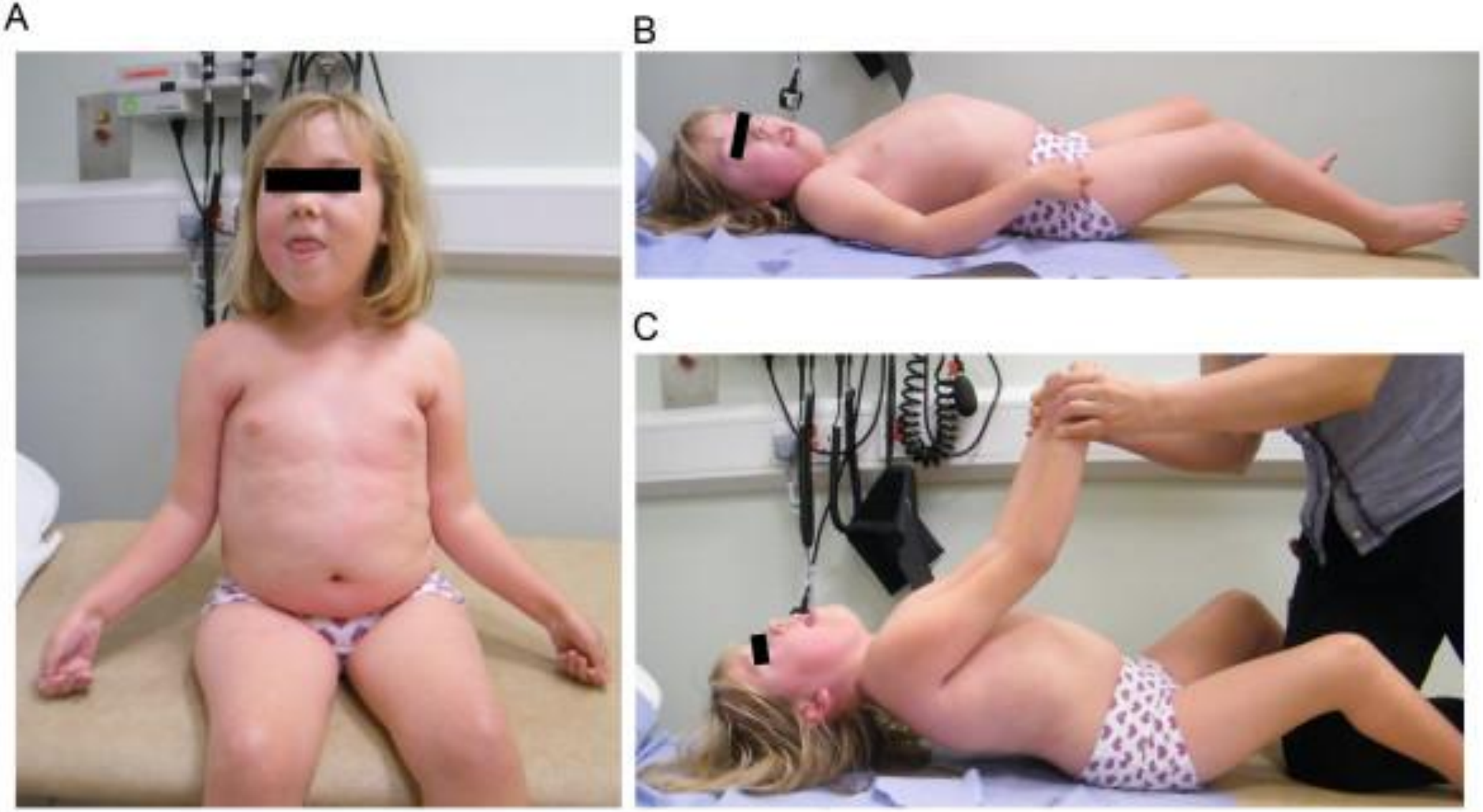
= There may be joint problems called 'contractures' (stiff joints), due to the baby not being able to move the joints enough.

= There are different types of CMD, which vary from person to person in how severe they are, and in whether or not they progress (get worse).

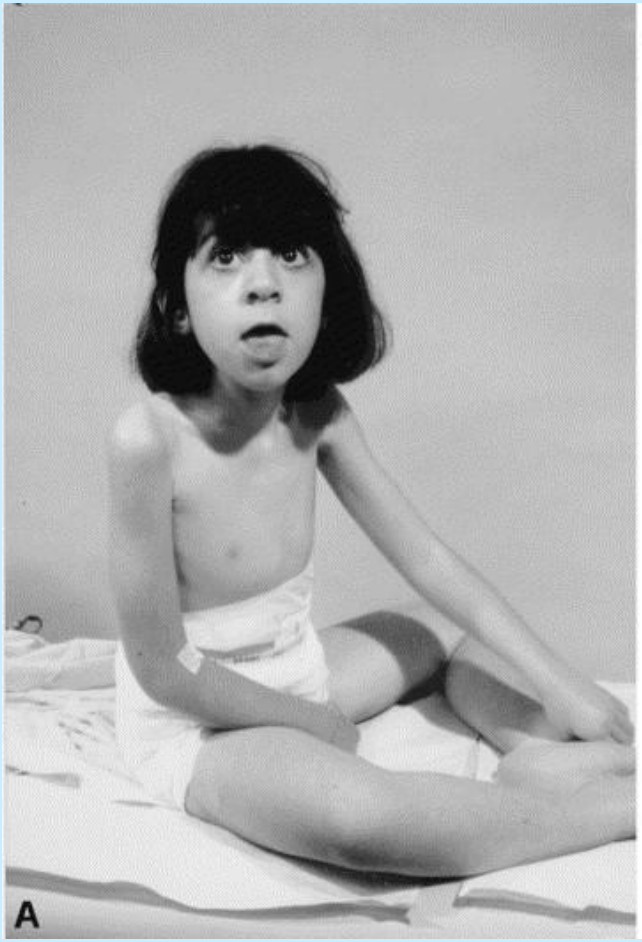
= In many cases, CMD is not progressive, so that although the child continues to have difficulties, their muscle strength improves with time, and the child may have a normal lifespan.

= Some types of CMD are more severe or progressive. In these cases, the muscle weakness is more severe, and the child may have other problems such as seizures, learning difficulties, breathing problems, and a poorer outlook.





(A) Facial weakness with an open mouth and reduced facial expression.
(B) Patient has developed bilateral elbow-flexion contractures, (B) Bilateral knee-flexion contractures and lumbar hyperlordosis. (C) Truncal weakness and neck-flexion weakness (lack of head control) when the patient is pulled up from a lying position



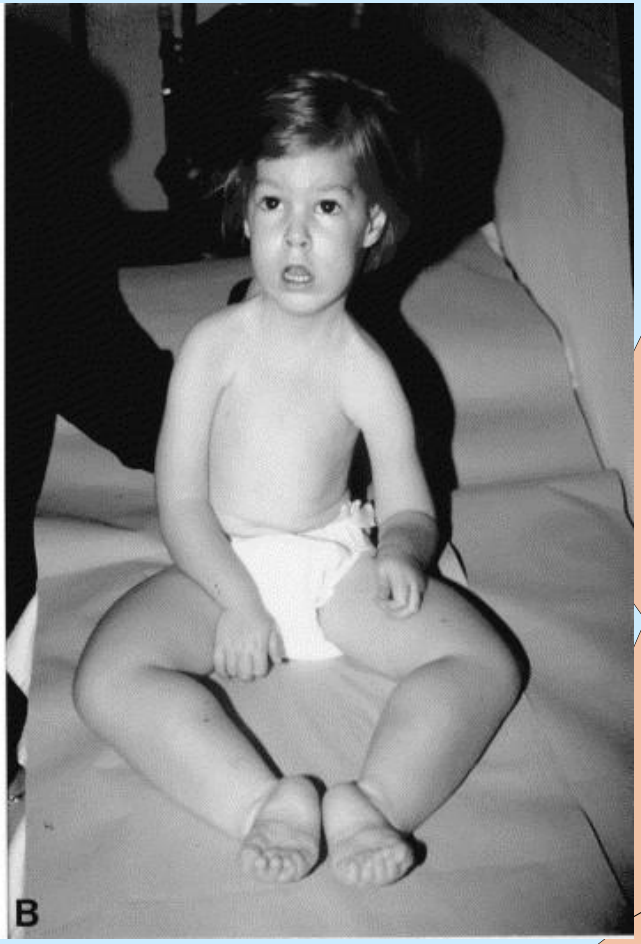
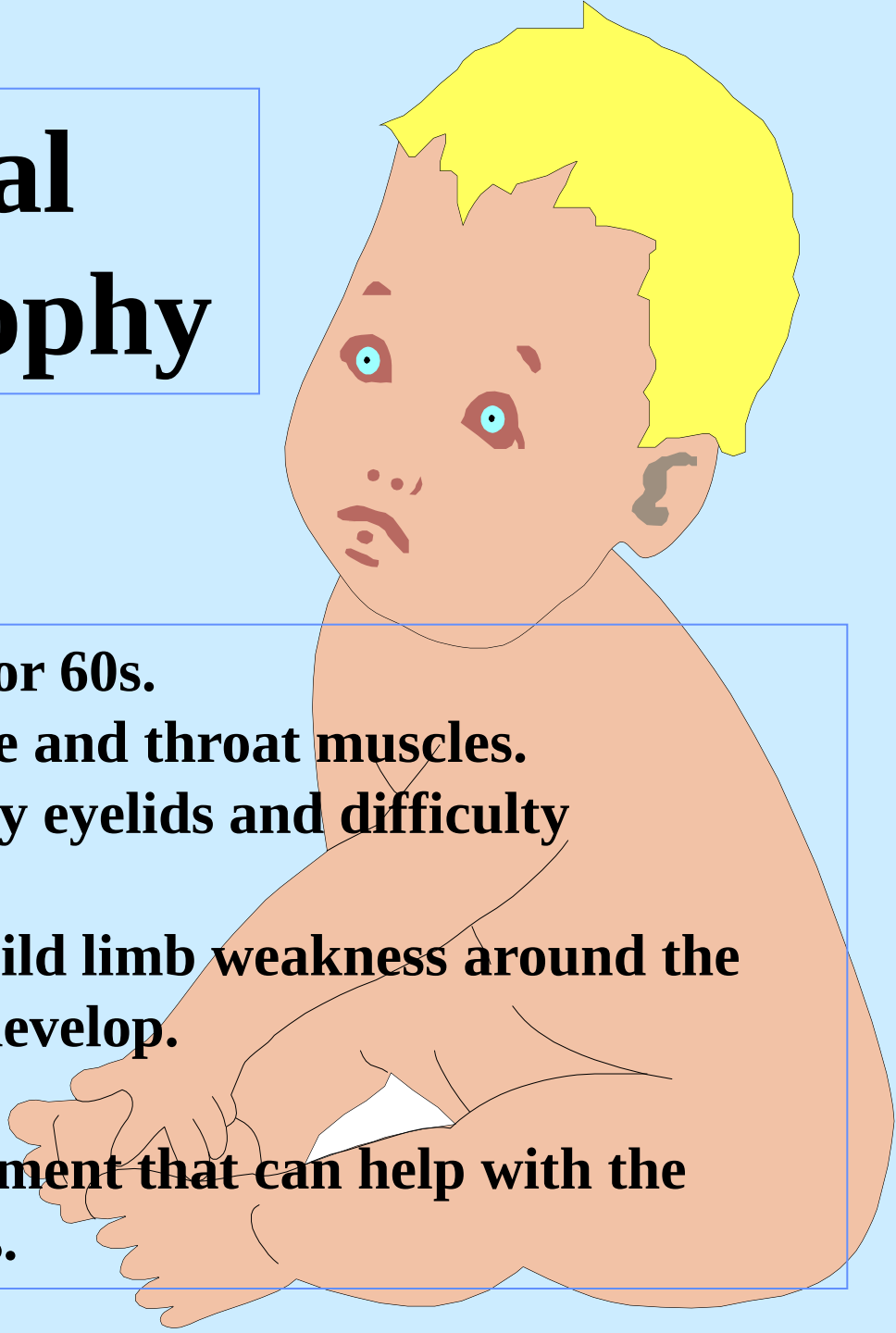


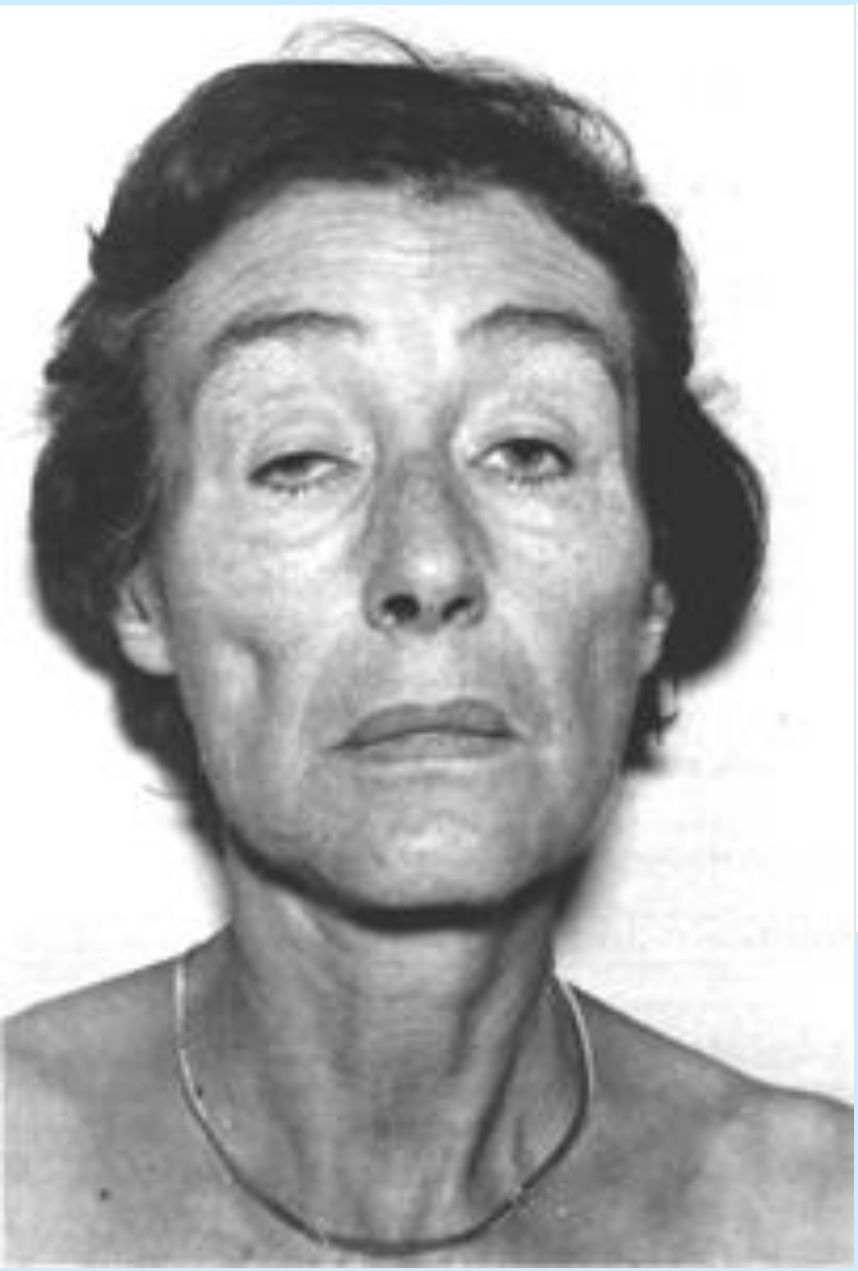
Fig. (A,B) Patients at the age of 9 and 4, respectively.
Note the enlargement of the calf muscle and, to a lesser extent, of the quadriceps muscle.
A considerable atrophy of the muscles of the scapular region is also evident.

Oculopharyngeal muscular dystrophy

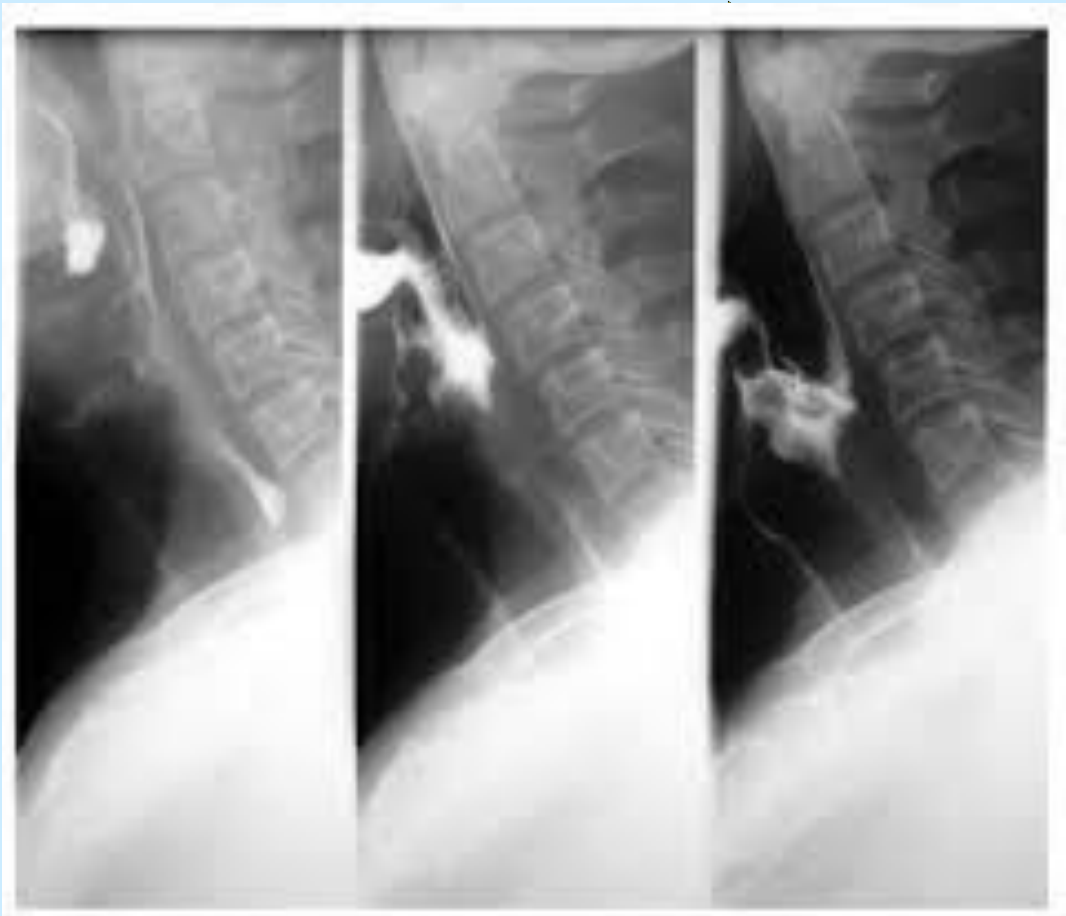


= usually starts around the 50s or 60s.
= It causes a weakness in the eye and throat muscles.
= The first symptoms are droopy eyelids and difficulty swallowing.
= Later on, after many years, mild limb weakness around the shoulders and hips may also develop.

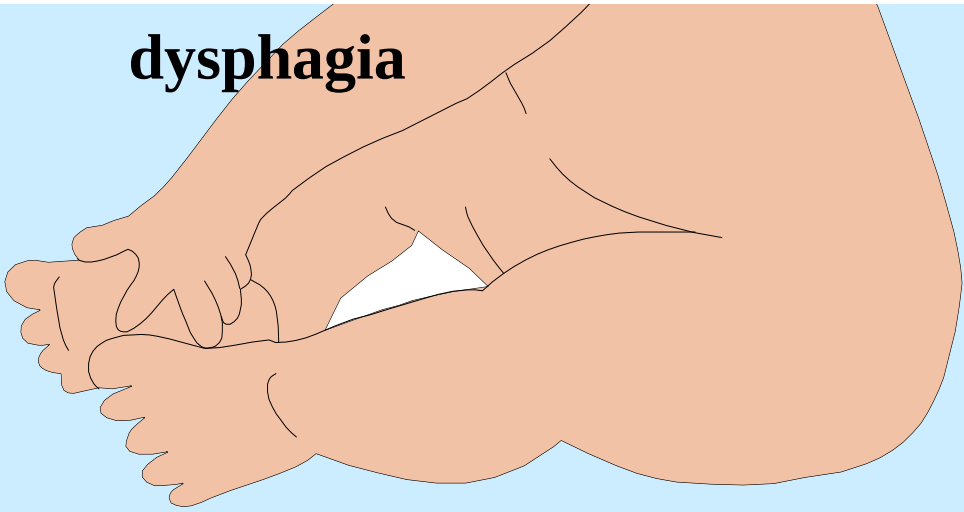
There are various types of treatment that can help with the eyelid and swallowing problems.



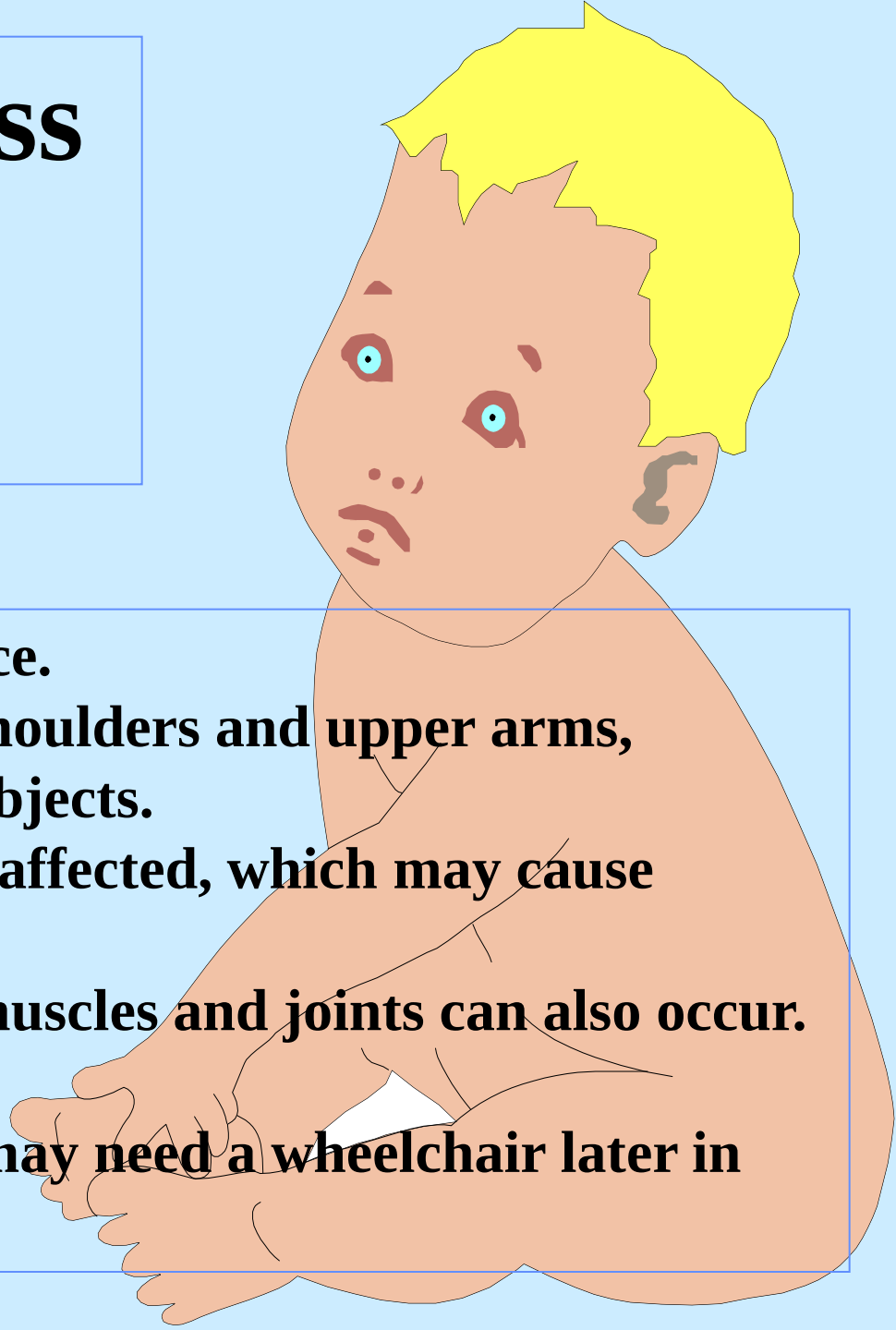
bilateral ptosis



dysphagia



Emery-Dreifuss muscular dystrophy

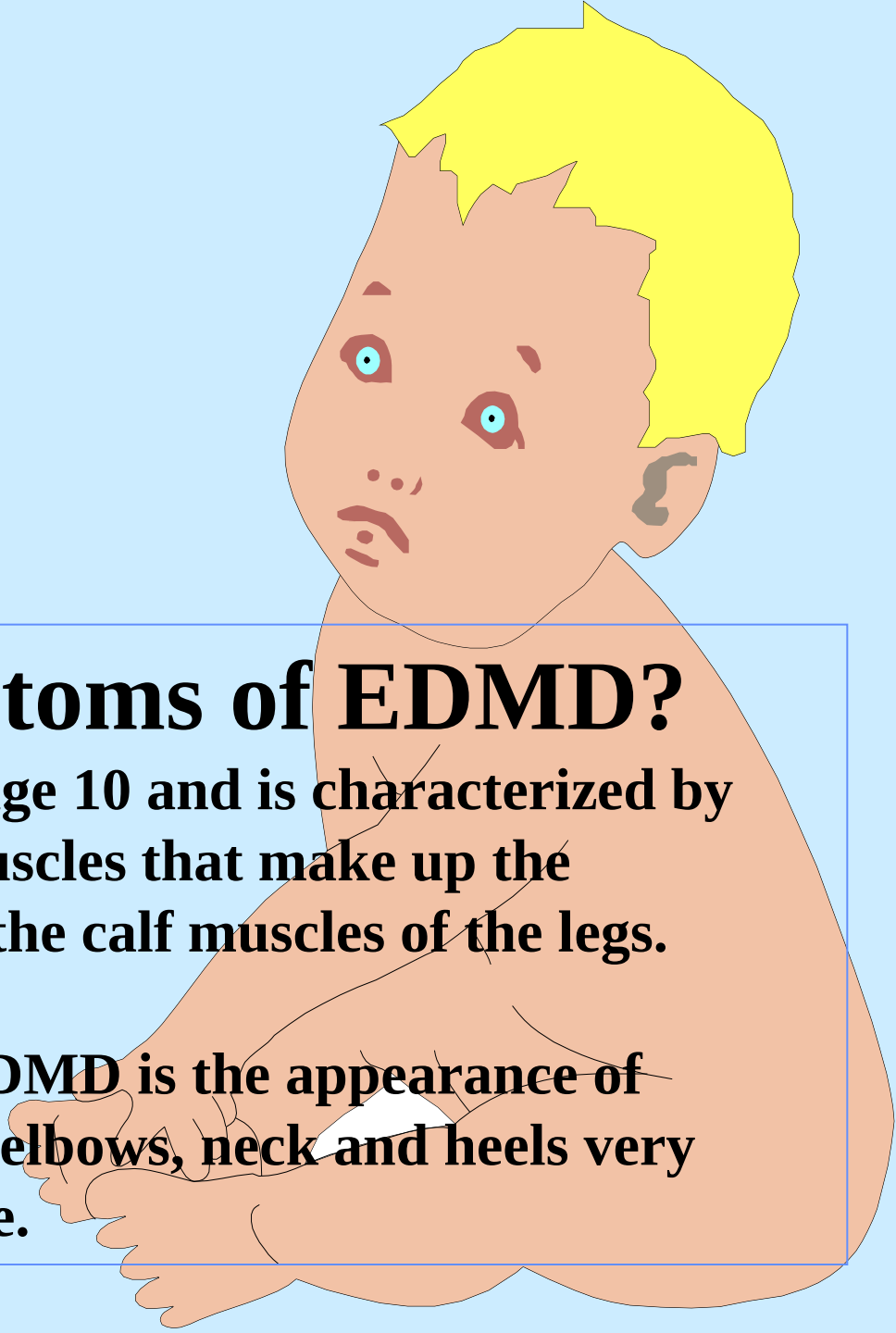


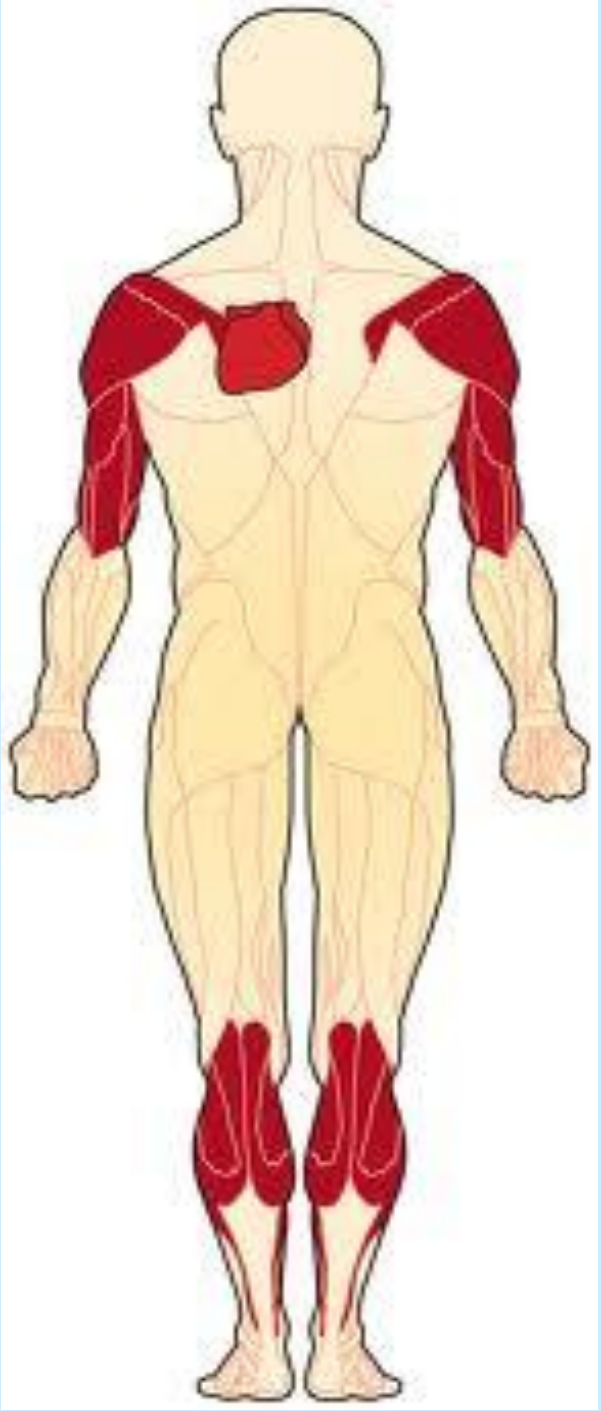
Starts in childhood or adolescence.
It can affect the muscles of the shoulders and upper arms, making it difficult to lift heavy objects.
The muscles in the lower leg are affected, which may cause tripping over when walking.
Contractures (tightness) of the muscles and joints can also occur.
Usually progresses very slowly.
People who have this condition may need a wheelchair later in life.

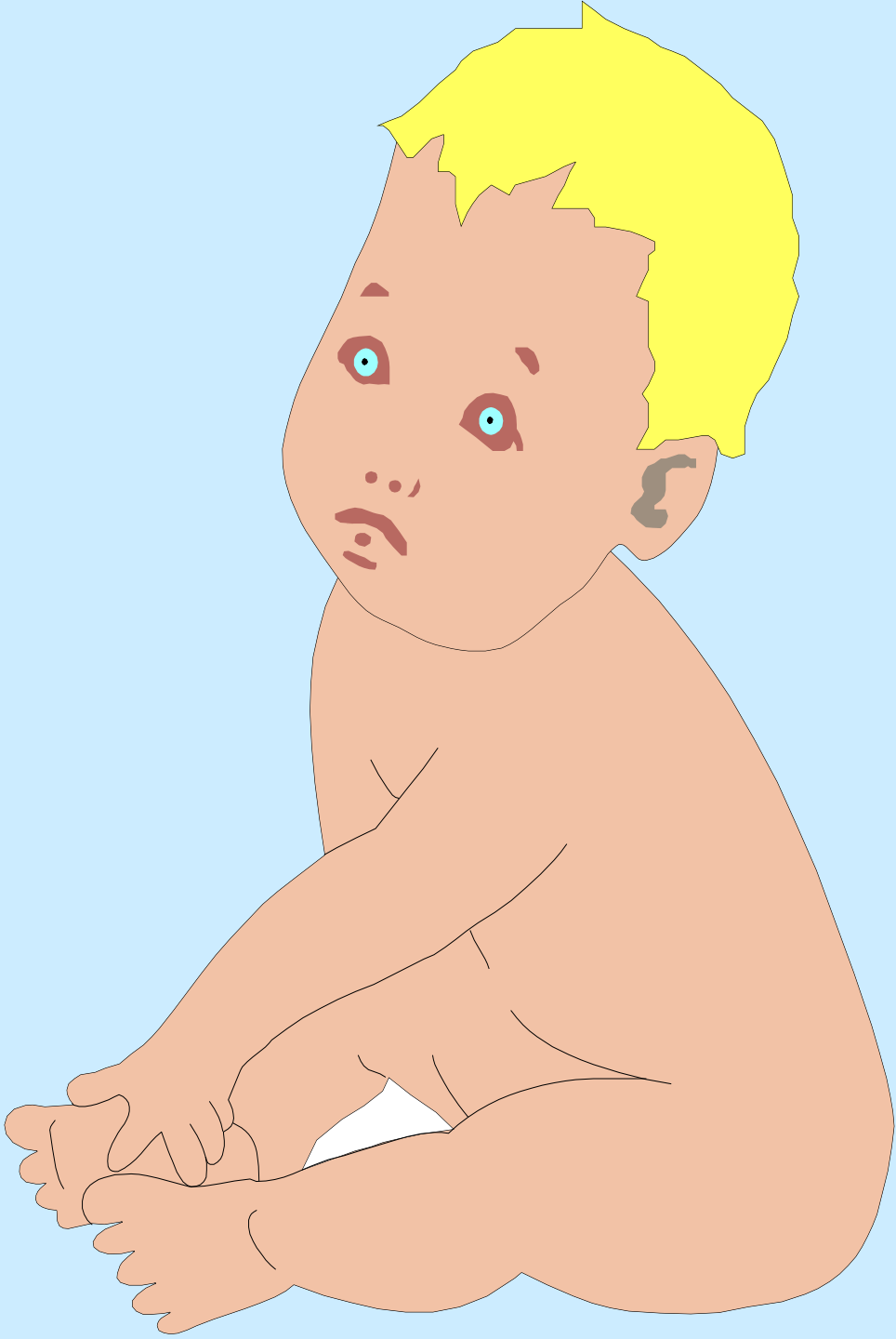
What are the symptoms of EDMD?

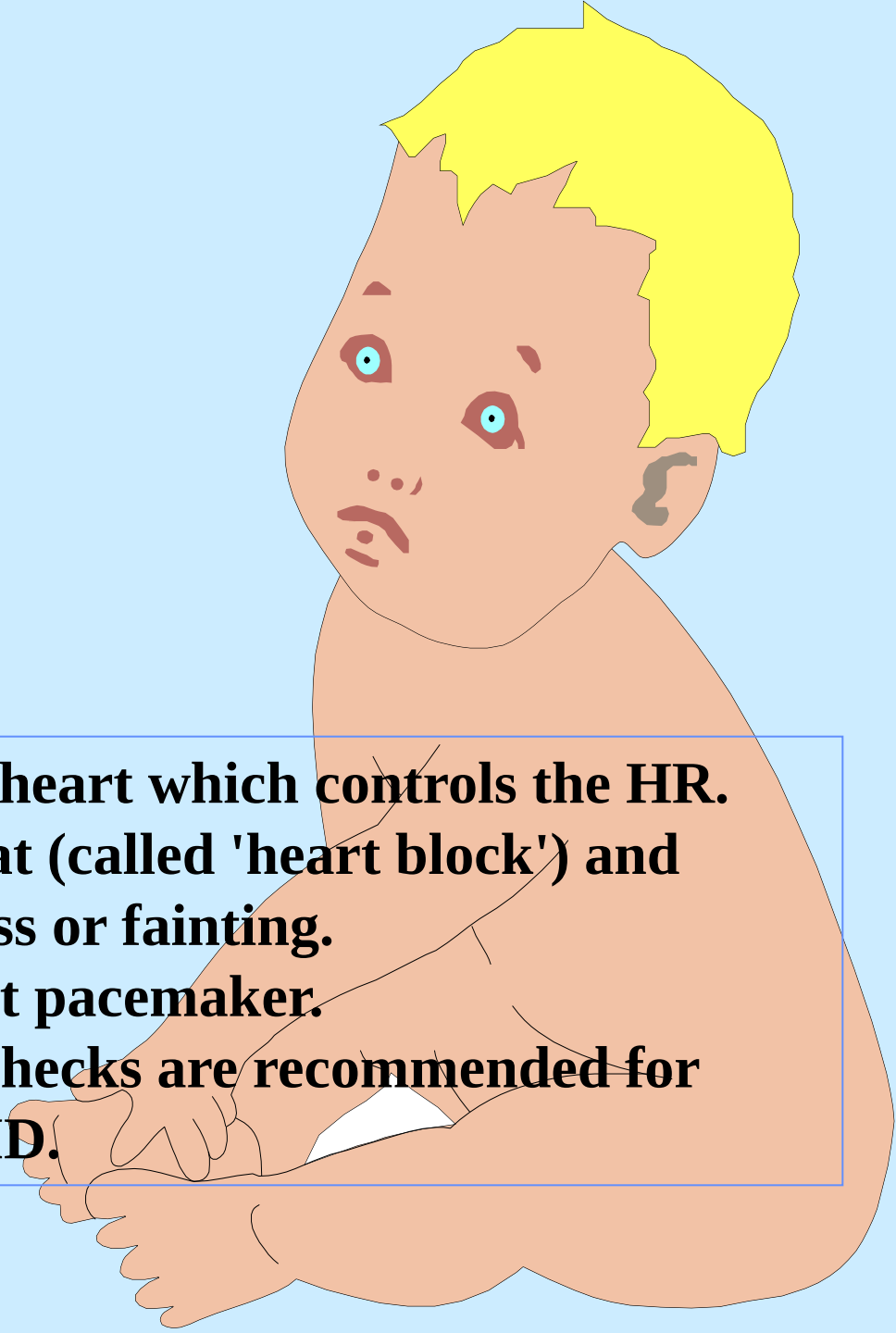
EDMD usually shows itself by age 10 and is characterized by wasting and weakness of the muscles that make up the shoulders and upper arms and the calf muscles of the legs.

Another prominent aspect of EDMD is the appearance of contractures (stiff joints) in the elbows, neck and heels very early in the course of the disease.









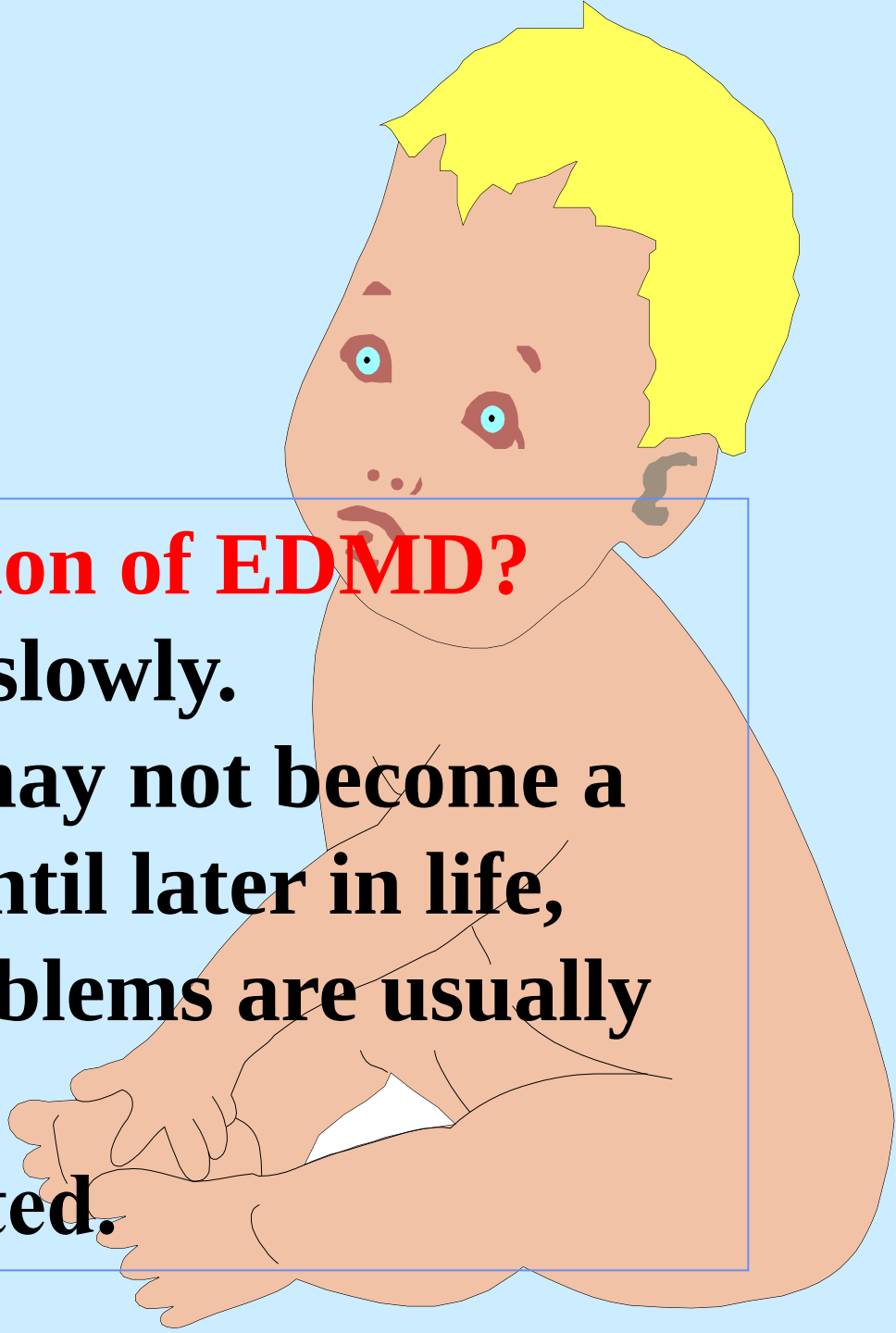
= sometimes affects a part of the heart which controls the HR.
= This may cause a slow heartbeat (called 'heart block') and symptoms of tiredness, giddiness or fainting.
= This can be treated with a heart pacemaker.
= For this reason, regular heart checks are recommended for people with Emery-Dreifuss MD.

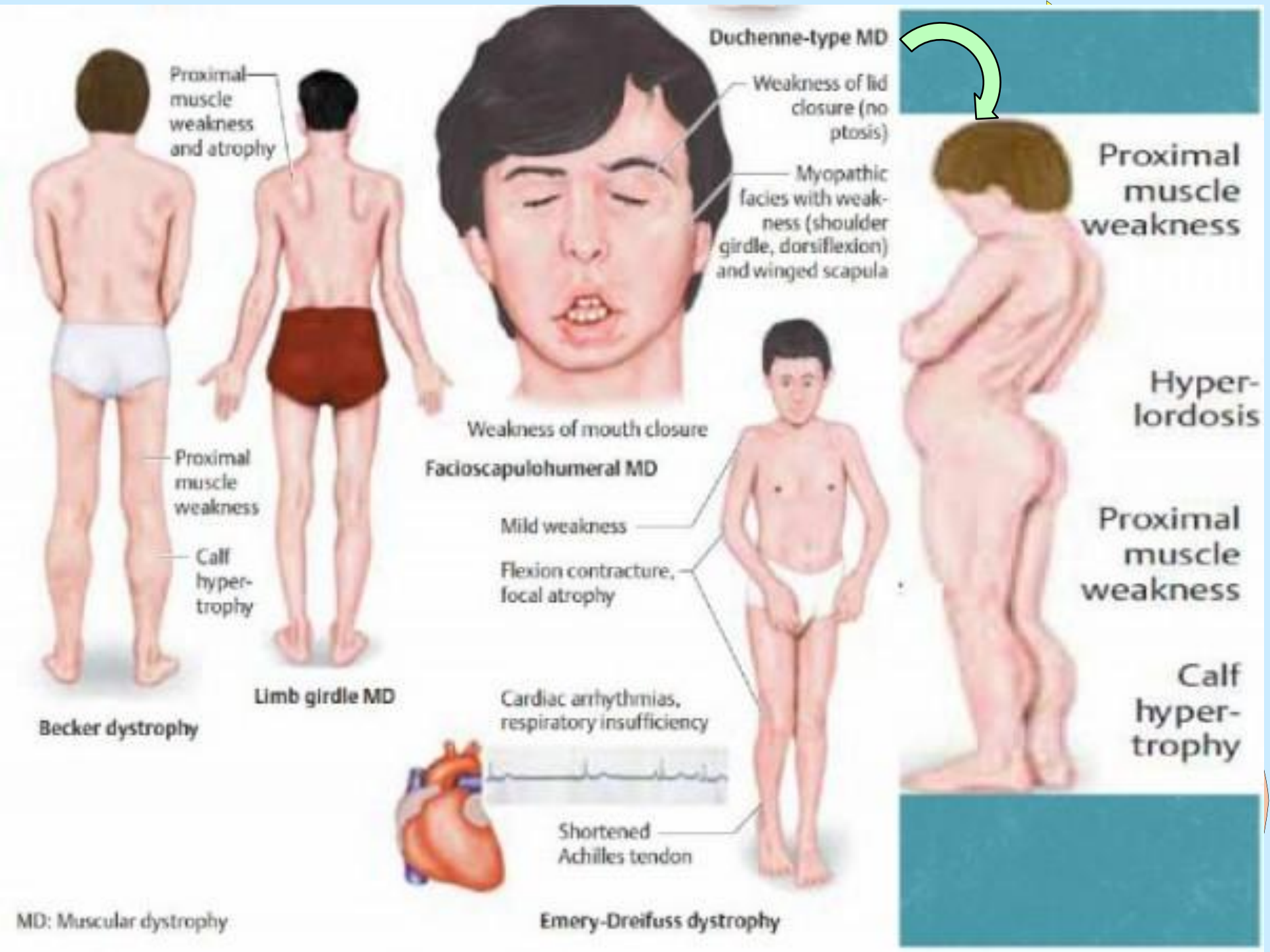
What is the progression of EDMD?

= EDMD progresses slowly.

= Muscle weakness may not become a source of difficulty until later in life, although cardiac problems are usually detectable by age 20.

= Intellect isn’t affected.





MD: Muscular dystrophy

Thank you