

Endocrinology

Evaluation and Treatment

Maria Vogiatzi, MD

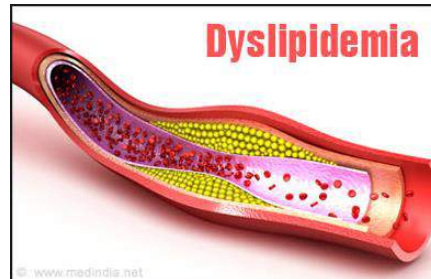
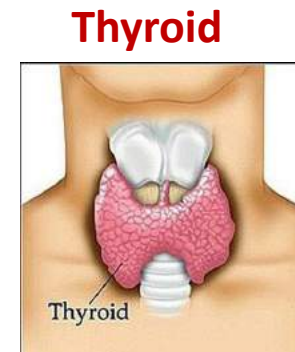
Pediatric Endocrinology

Children's Hospital of Philadelphia

Endocrinology



What does endocrine entail?



**Osteoporosis/
Fractures**



Evaluation of Short Stature, Carbohydrate Metabolism and Other Endocrinopathies in Bloom's Syndrome

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- Study was done from 1999 to 2003
- Included 11 participants
- 3 adults- 8 children; age 0.8 – 28.6 yrs
- 5 males & 6 females

Growth in Bloom's Syndrome

Small birth weight

In the Cornell Study

- average 1.85kg or 4.1 lbs for males
- average 1.51kg or 3.33lbs for females

Growth in Bloom's Syndrome

Individuals with Bloom's Syndrome remain small as children and reach a final height below normal.

Adult men:

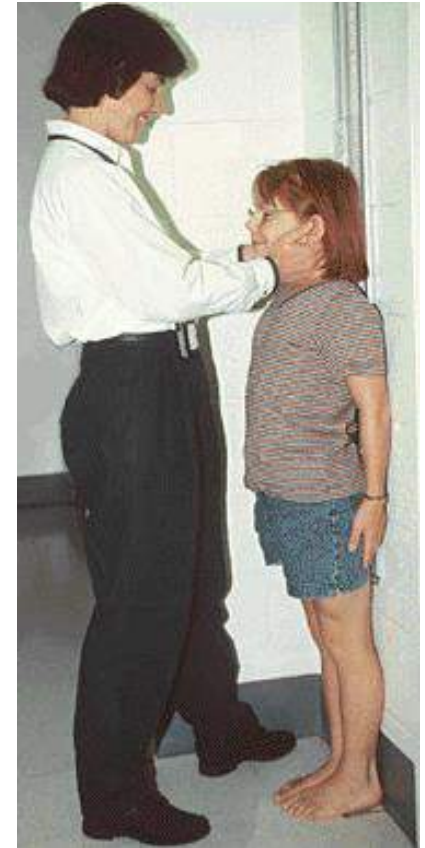
Average height: 148.5 cm or 58.4"

Average weight: 41.3 kg or 91 lbs

Adult woman:

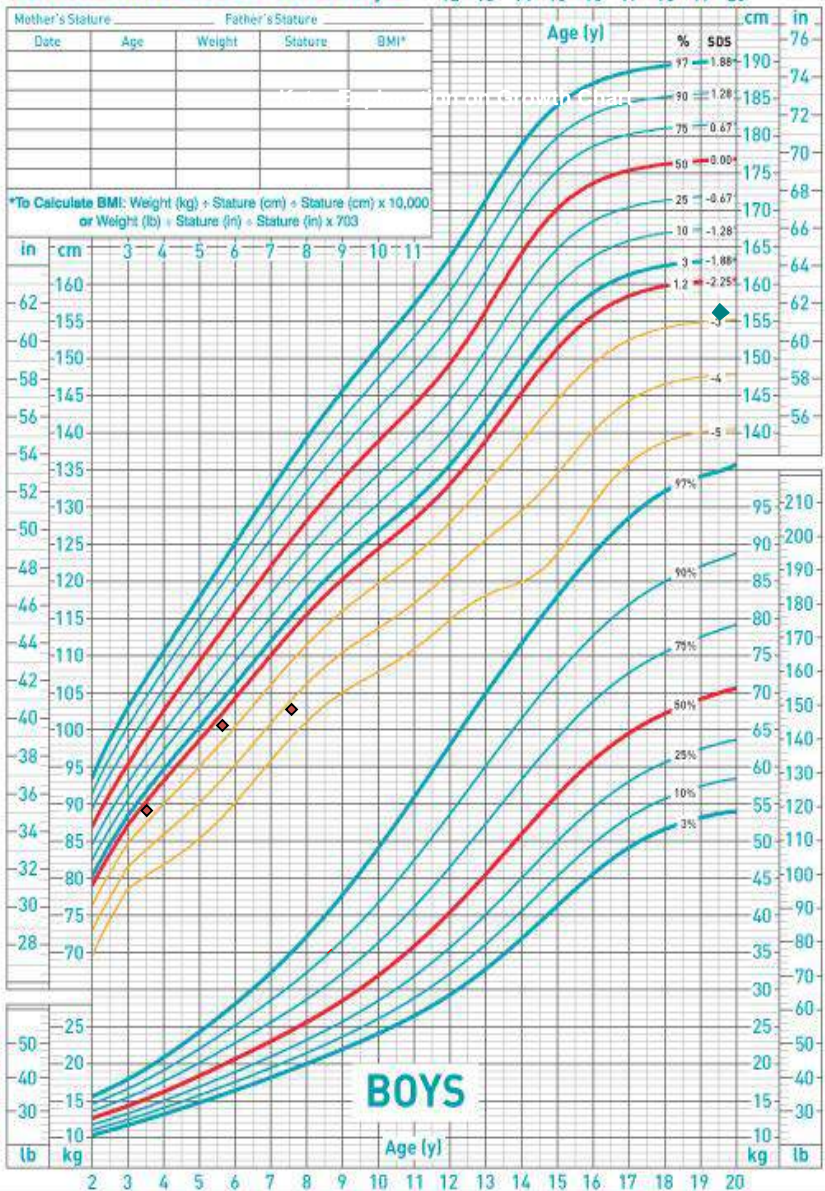
Average height: 141.5 cm or 55.7"

Average weight: 36.6 kg or 80.7 lbs



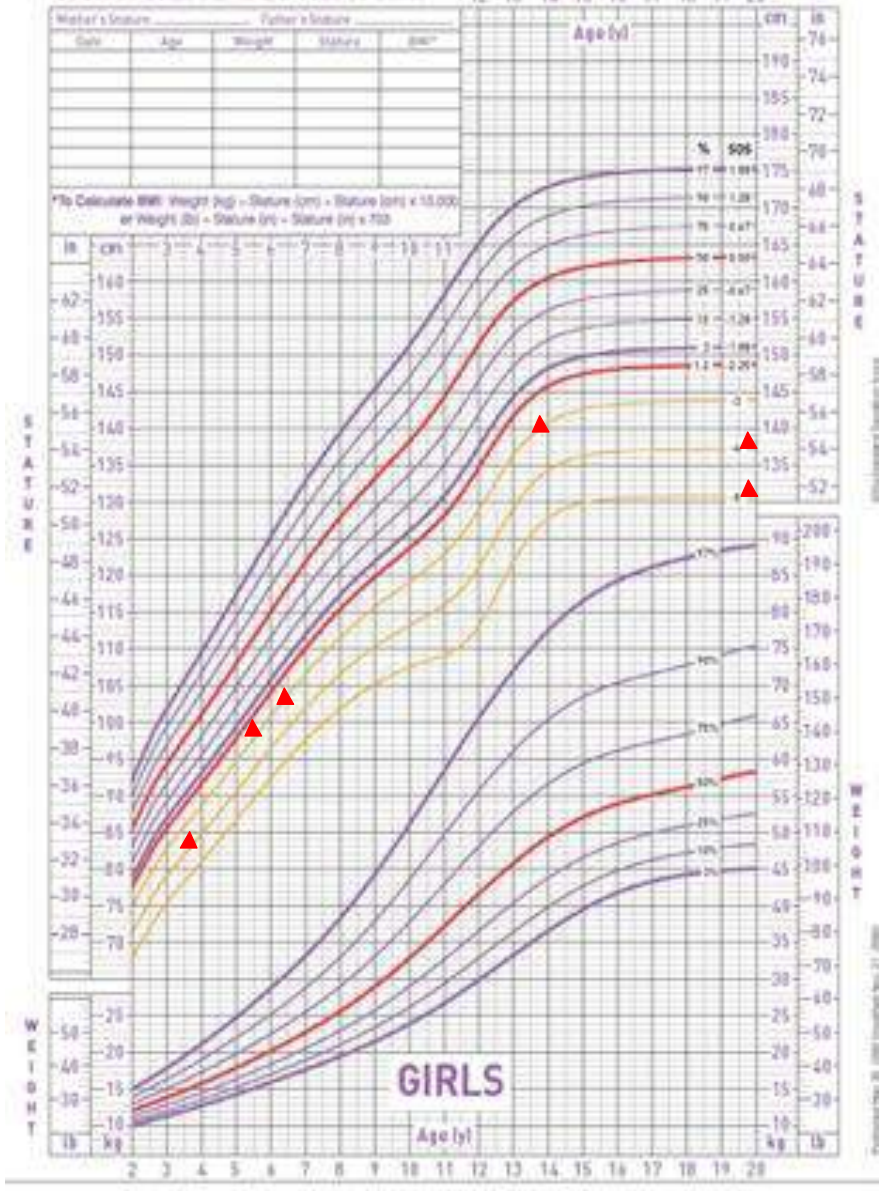
Growth Chart 2 to 20 Years: Boys

12 13 14 15 16 17 18 19 20



Growth Chart 2 to 20 Years: Girls

12 13 14 15 16 17 18 19 20



What can cause short stature in Bloom's Syndrome?

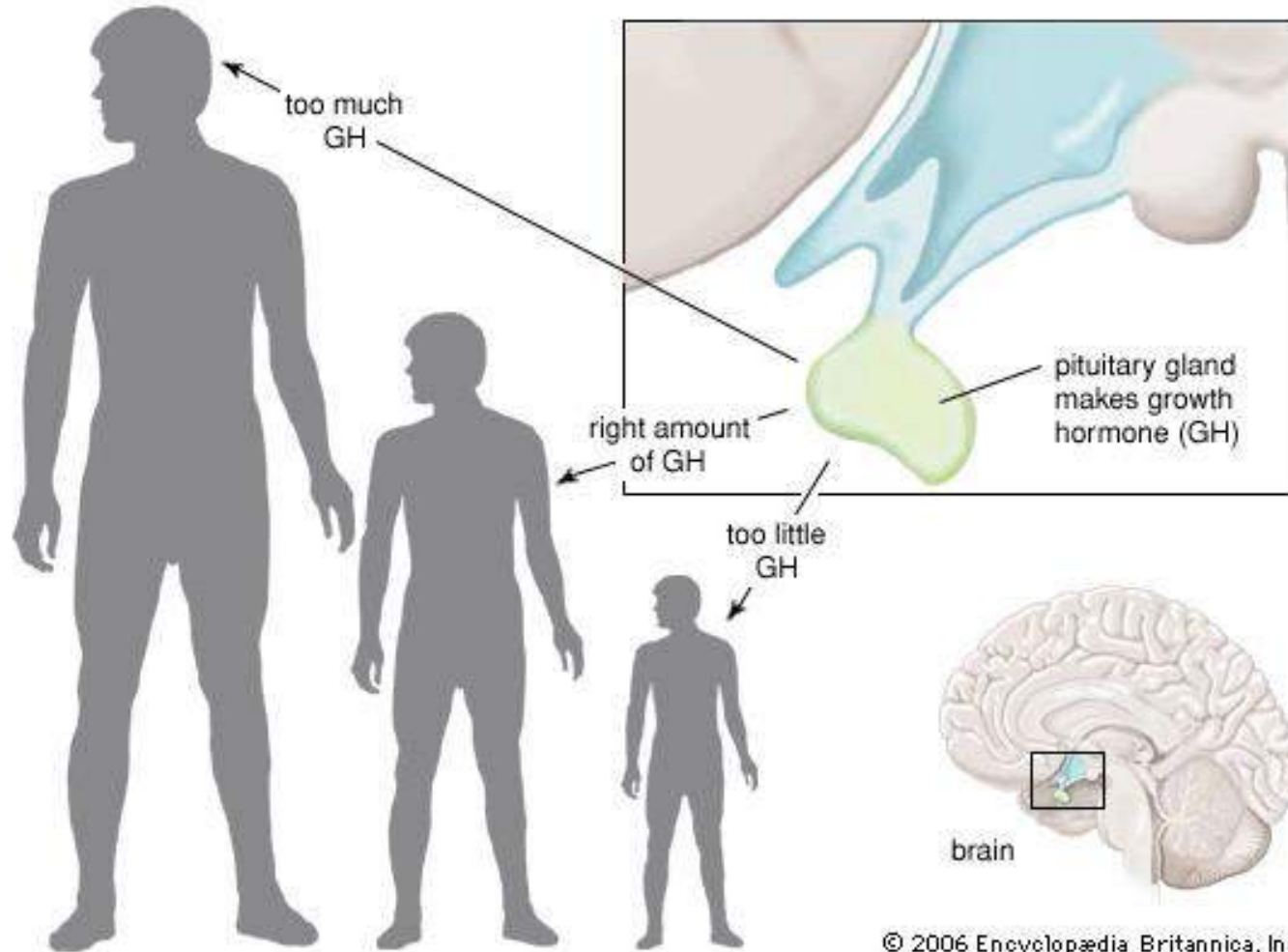
Genes: Bloom's Syndrome itself
Low birth weight is a frequent cause of short stature

Nutrition: slow weight gain

Hormonals: growth hormone
thyroid hormone

Chronic illness

Growth hormone: stimulates linear growth

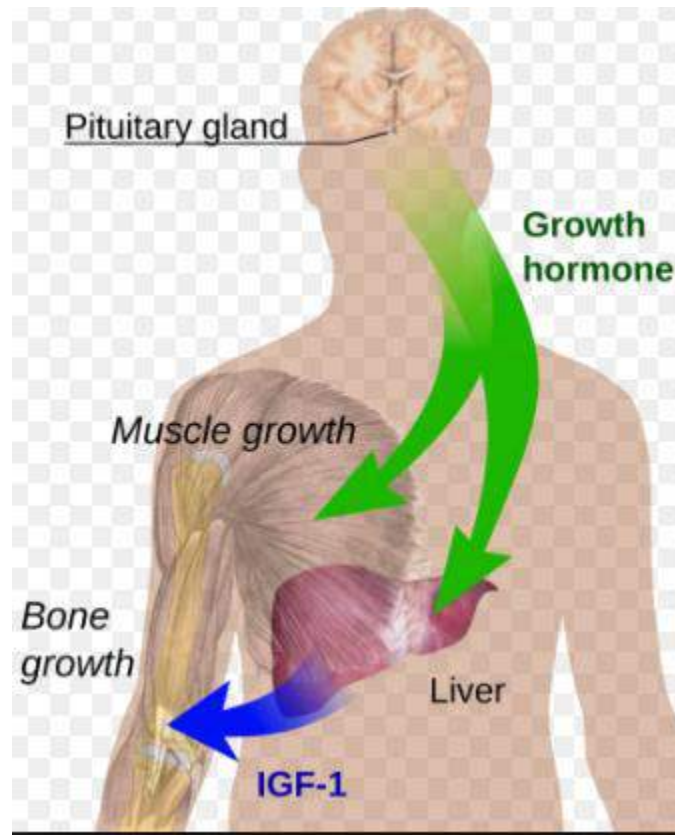


Children with growth hormone deficiency: short stature

16 yo boy with
growth hormone
deficiency



Children with Bloom's Syndrome do NOT have growth hormone deficiency



Are Children with Bloom's Syndrome candidates for growth hormone therapy?



Growth hormone improves final height in children born small and who grow poorly.

Limited experience in Bloom Syndrome

Growth hormone may increase an individual's risk for cancer: a topic of some debate.

What about nutrition?

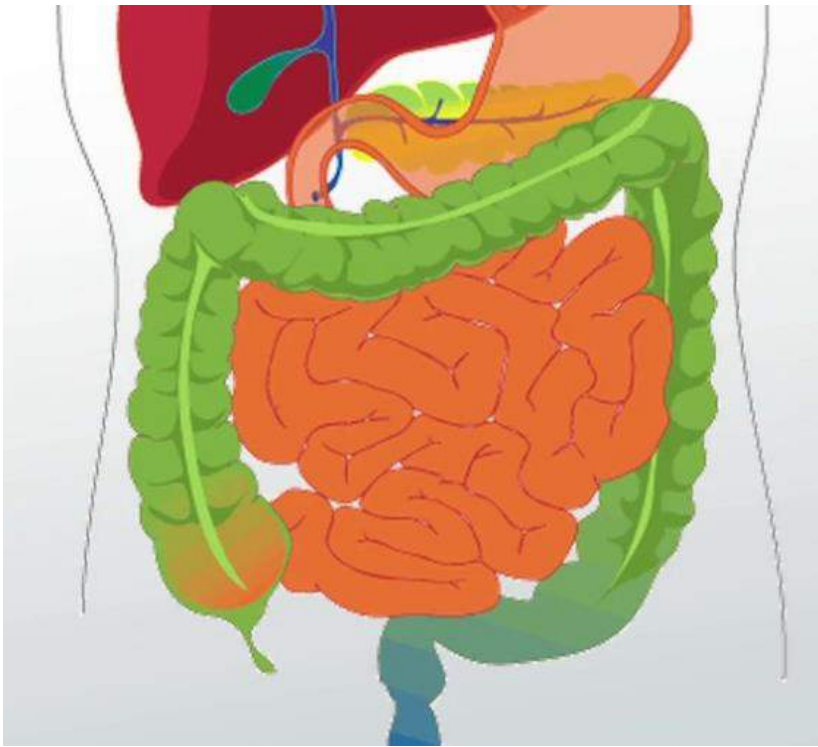


Feeding difficulties in early life.

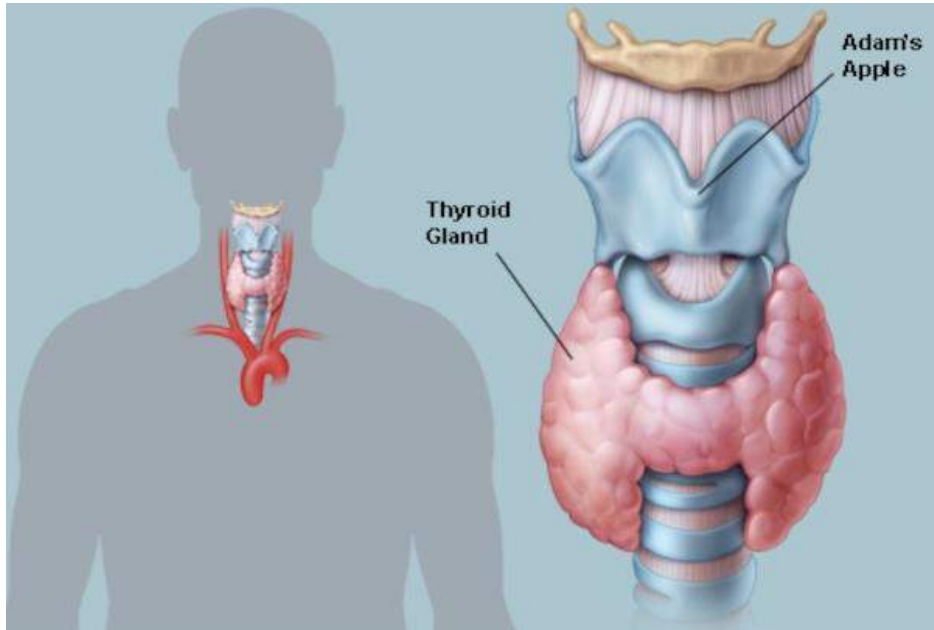
Frequent vomiting and GE reflux.

Normal weight for height as adults.

What about malnutrition?



Cornell Study:
no evidence of
malabsorption



Thyroid abnormalities in Bloom's syndrome

Cornell study: 18%
Bloom registry: 3%



HYPOTHYroidism

Children: Poor growth
Developmental delays

Children & Adults: Fatigue
Feeling cold
Overweight
Dry skin, hair loss
Constipation
Brain fog, Depression

Diabetes

The word "Diabetes" is displayed in a playful, multi-colored font. Each letter is held up by a hand of a different skin tone, representing diversity. The letters are: 'D' (blue), 'i' (purple), 'a' (red), 'b' (green), 'e' (yellow), 't' (pink), 'e' (light blue), and 's' (red). The hands are positioned below the letters, with the fingers gripping the bottom edges. The background is plain white.

High rates of prediabetes and diabetes Cornell Study

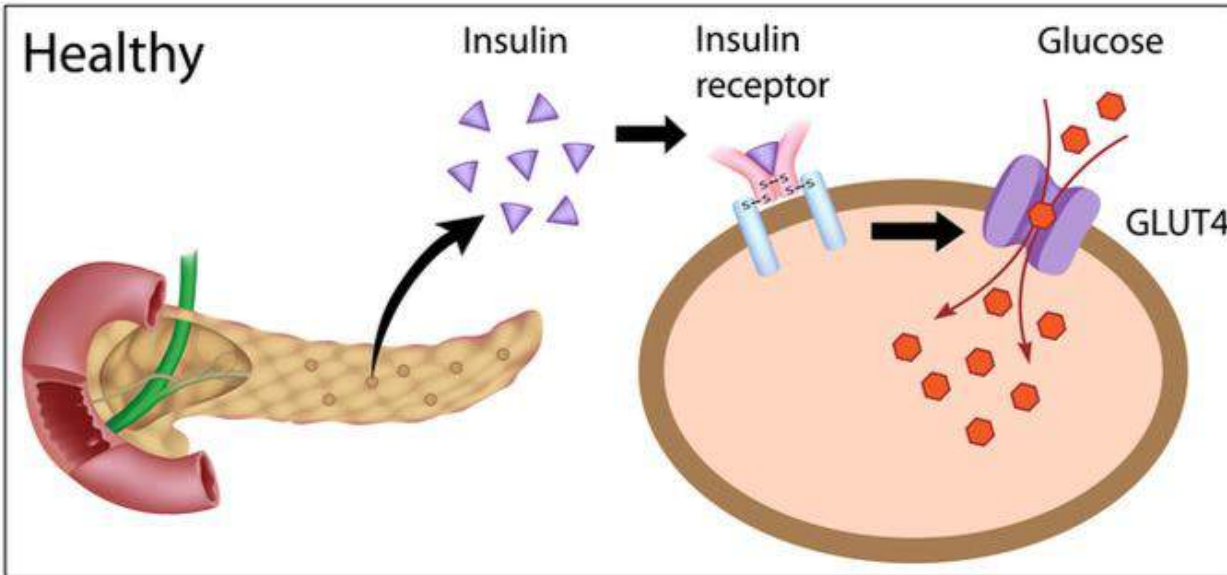
Oral Tolerance Glucose Test (OGTT)

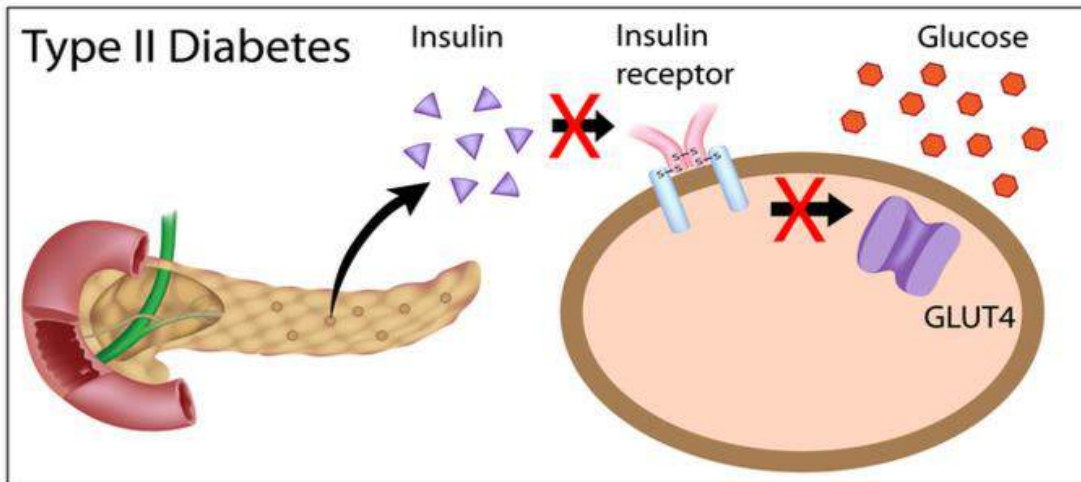
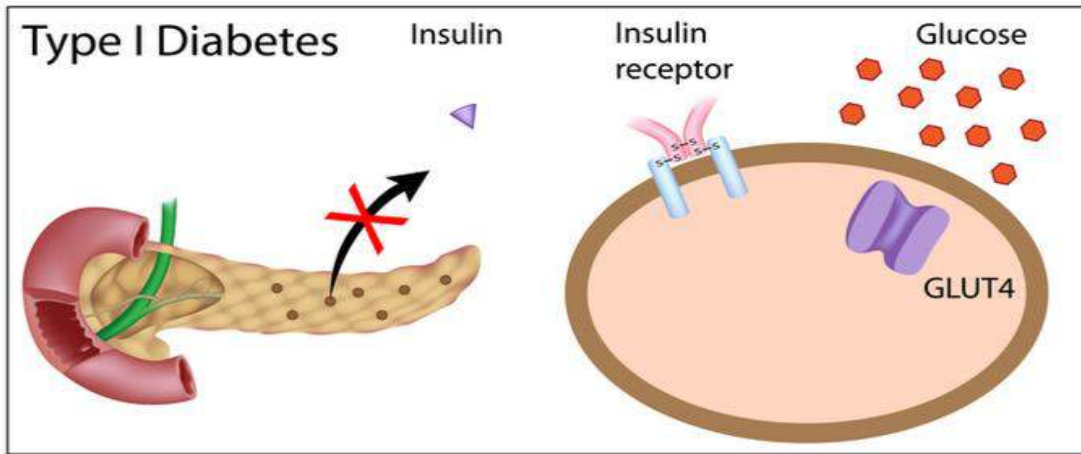
Participant	Sex	Age, years	Glucose, mg/dl	
			0 h	2 h
BSR205	M	0.8	99.0	92.0
BSR231	M	3.3	84.0	118.0
BSR234	F	3.4	97.0	124.0
BSR204	F	5.6	76.0	94.0
BSR230	F	5.7	77.0	167.0
BSR182	F	6.6	82.0	104.0
BSR214	M	7.8	93.0	116.0
BSR139	F	13.7	100.0	196.0
BSR92	F	23.0	136.0	371.0
BSR79	M	28.1	93.0	184.0
BSR53	F	28.6	117.0	195.0

High rates of prediabetes and diabetes Cornell Study

- Prediabetes starts early in childhood.
- Risk of prediabetes and diabetes increase with age.
- People with prediabetes or diabetes usually have normal fasting blood sugar.
- People with prediabetes or diabetes usually have no symptoms.

Bloom registry: 18% of people report diabetes





Individuals with Bloom's syndrome have type II Diabetes and insulin resistance

Common Signs of Diabetes



Abnormal thirst



Frequent urination



Extreme tiredness



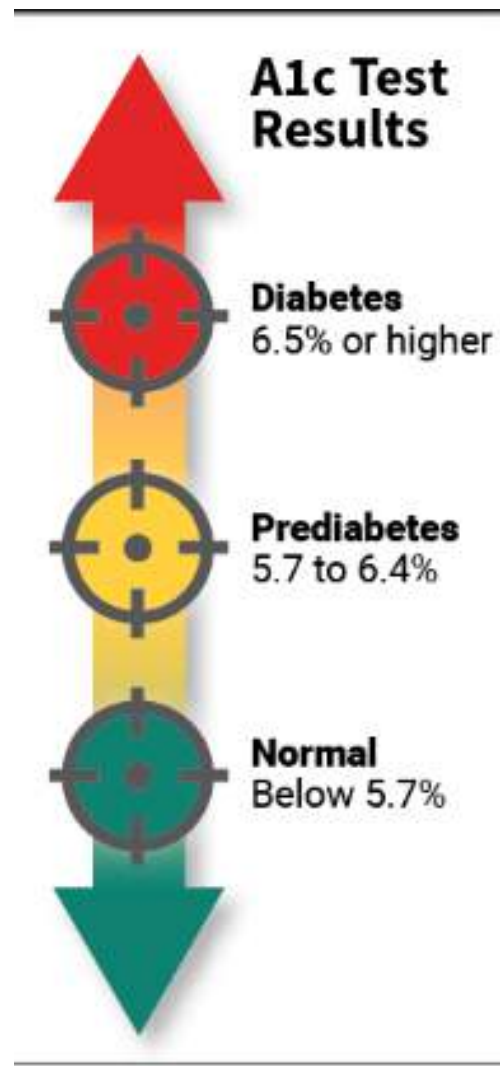
Sudden weight loss

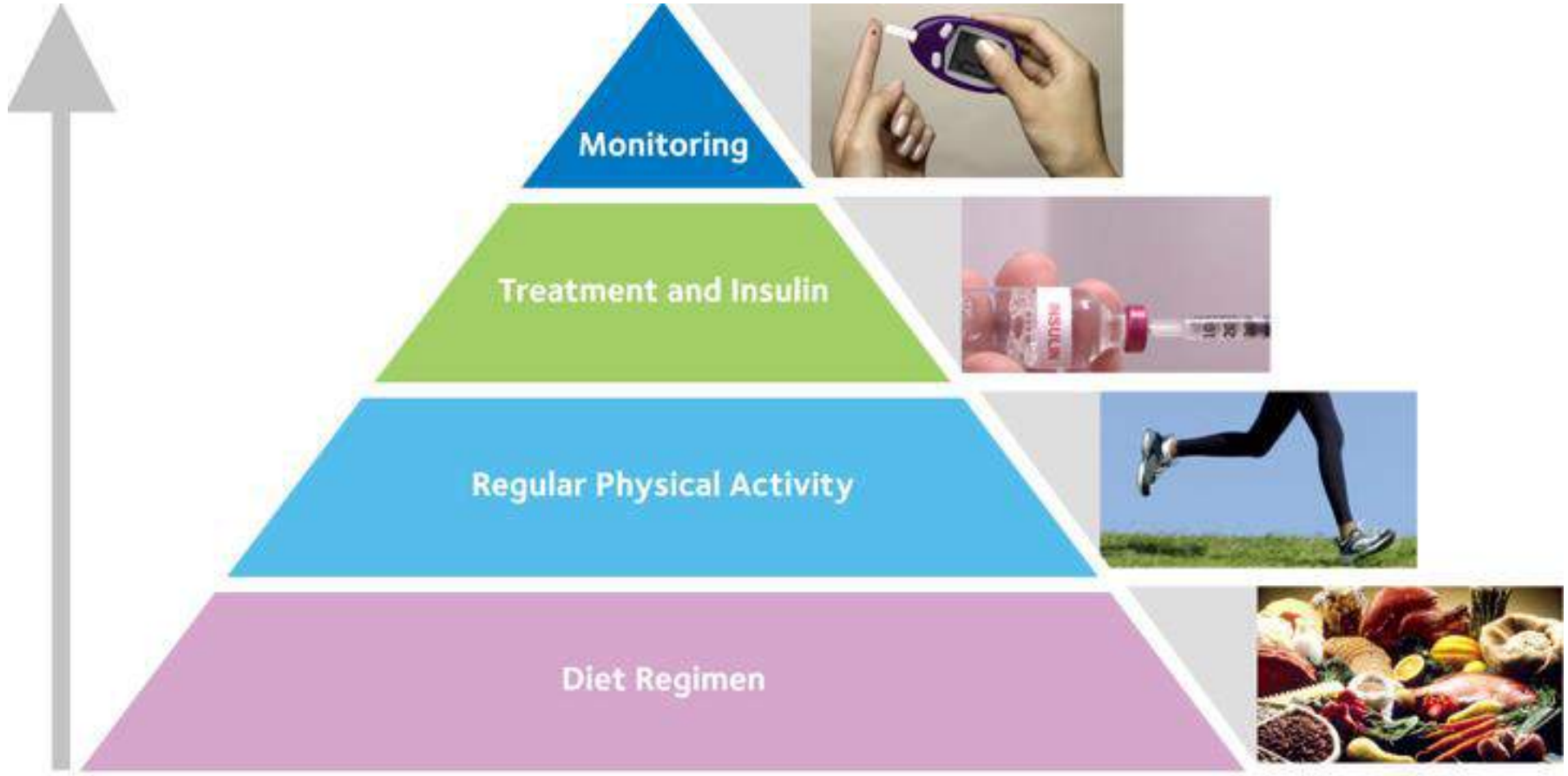


Constant hunger



Blurred vision





High rates of lipid abnormalities, especially among people with prediabetes or diabetes: Cornell Study

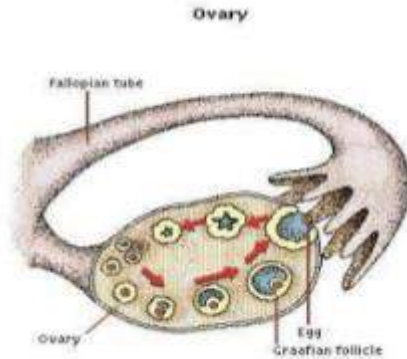
Participant	Sex	Age year	Cholesterol			
			total mg/dl	LDL mg/dl	HDL mg/dl	trig. mg/dl
BSR205	M	0.8	102			
BSR231	M	3.3	163			
BSR234	F	3.4	108	66	35	35
BSR204	M	5.6	169	104	40	94
BSR230	F	5.7	176			
BSR282	F	6.6	101			
BSR214	M	7.8	169			39
BSR139	F	13.7	123	50	58	75
BSR92	F	23.0	186	108	29	247
BSR79	M	28.1	219	132	53	169
BSR53	F	28.6	214	145	41	142

PUBERTY

A hand holding a black marker is shown underlining the word 'PUBERTY' which is written in large, black, hand-drawn capital letters on a white background. The hand is positioned at the bottom right of the word, with the marker tip touching the underline.

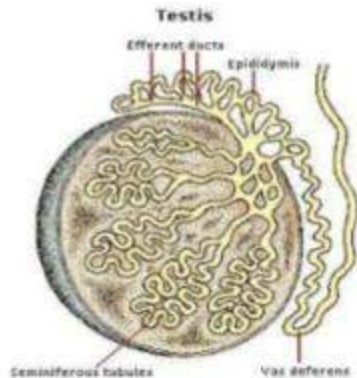
AND FERTILITY

Puberty may be delayed but progresses normally.



Concern of primary ovarian or testicular problem in Bloom.

- Women may have normal pregnancies and babies but may experience early menopause.



- Men usually have normal testosterone levels but may have sperm abnormalities.

Pregnancy and Bloom Syndrome



- Preconception and pregnancy screening for diabetes.
- Delivery difficulties because of small pelvis.
- Pregnancy outcome is usually good.

Conclusions

Children:

Monitor growth and weight gain.

Consult a feeding specialist; use of high calorie diet.

Use of Growth Hormone is controversial and needs to be closely monitored.

Cunniff C. Health Supervision for People with Bloom Syndrome. Journal: American Journal of Medical Genetics, 2018

Conclusions

Annual screening for diabetes as early as age 10 yrs.

Annual screening for lipid abnormalities as early as 10 years.

Screening for hypothyroidism.

Individuals with Bloom's syndrome should be aware of symptoms and signs of diabetes and hypothyroidism.

Cunniff C. Health Supervision for People with Bloom Syndrome. Journal: American Journal of Medical Genetics, 2018

Conclusions

Women with Bloom syndrome may have normal babies but experience early menopause.

Men with Bloom syndrome who wish to have children may need to have a semen analysis and consult a fertility specialist.

Cunniff C. Health Supervision for People with Bloom Syndrome. Journal: American Journal of Medical Genetics, 2018



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