Endocrinology and Bloom Syndrome

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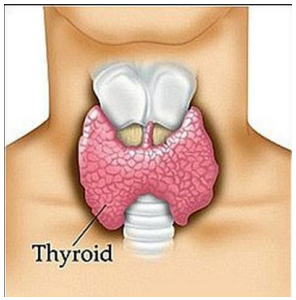
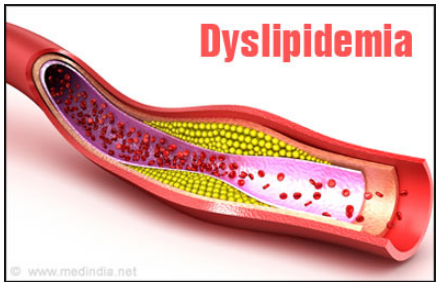
Endocrinology

What does endocrine entail?

Puberty

Thyroid





Fertility

Osteoporosis/
Fractures

Growth in Bloom's Syndrome

Small birth weight

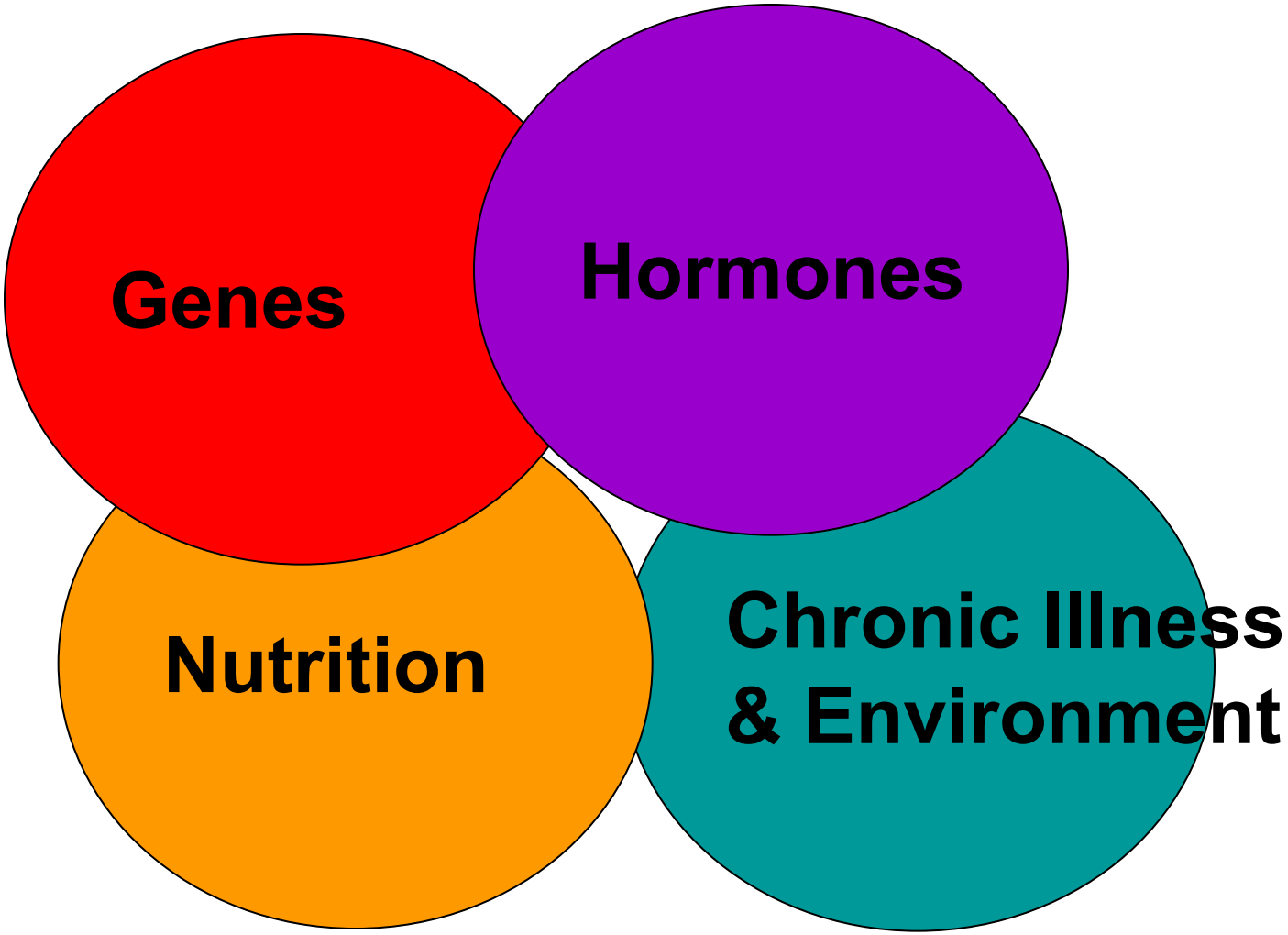
- About 1-1.5 kg(2.2 – 3.3lbs) smaller than general population

Stay small for age throughout childhood

Short adult height

- Men
  Height: 148.5cm (58.4")- Weight: 41.3kg (91 lbs)
- Woman
  Height: 141.5cm (55.7")- Weight: 36.6kg (80lbs)

Major Factors Involved in Growth



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

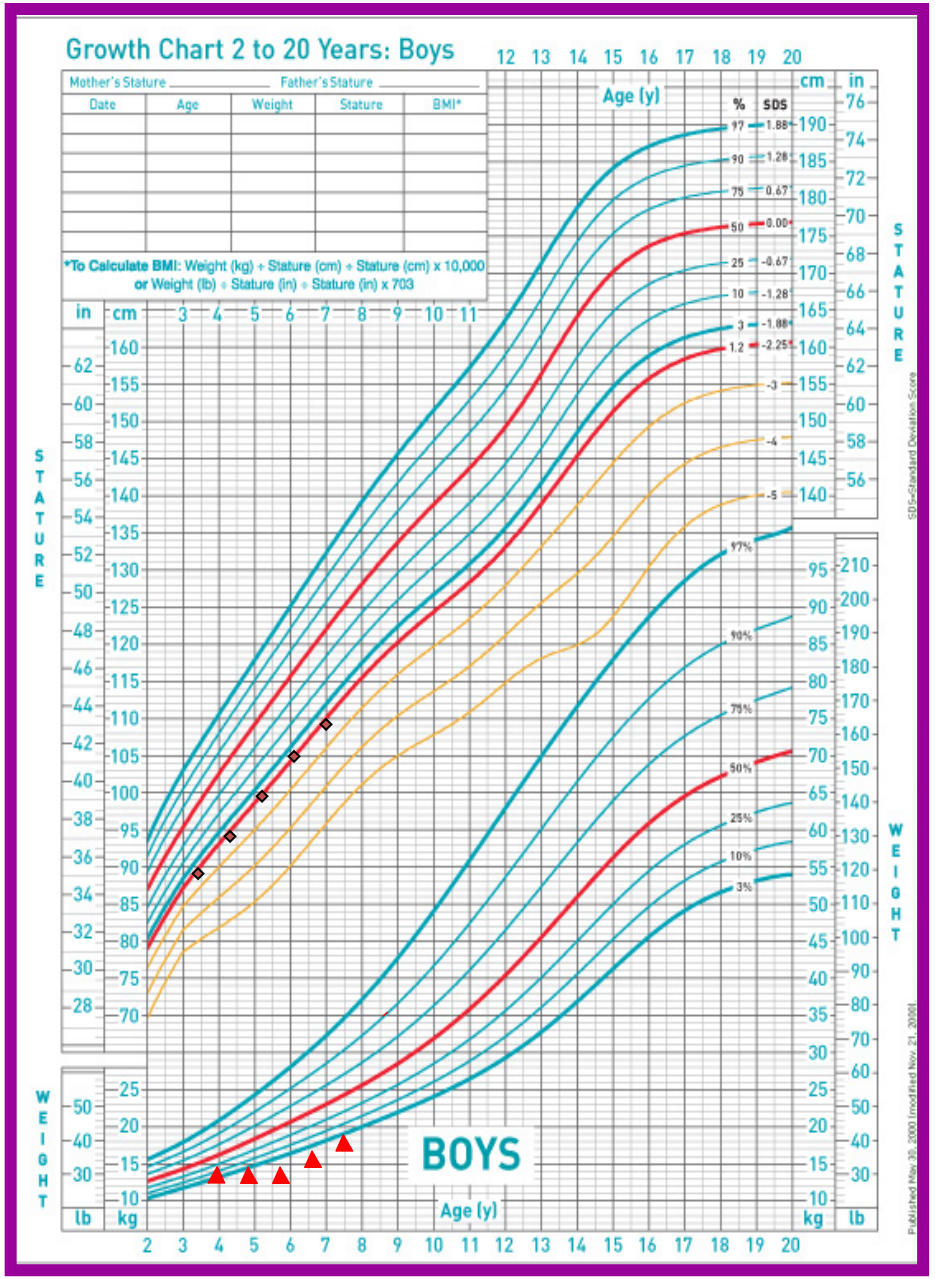
What about nutrition?

- Feeding difficulties in early life.
- Frequent vomiting and GE reflux.
- No evidence of malabsorption

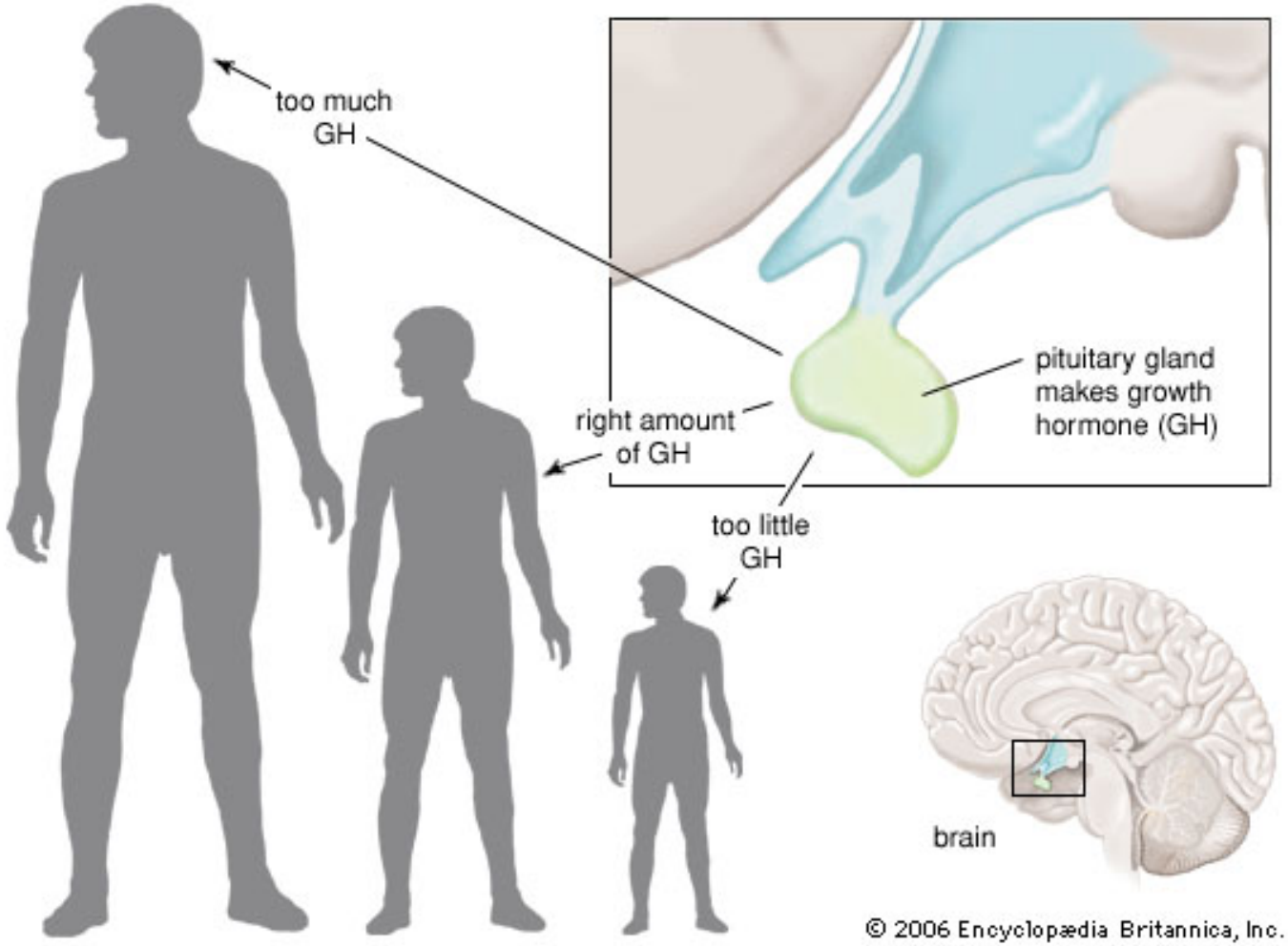
Normal weight for height as adults.

What can a parent do?

- Monitor growth
- Optimize calories
- Keep weight appropriate for body size



Growth hormone : stimulates linear growth



Growth hormone in Bloom Syndrome

Children with Bloom do NOT have growth hormone deficiency

Questions

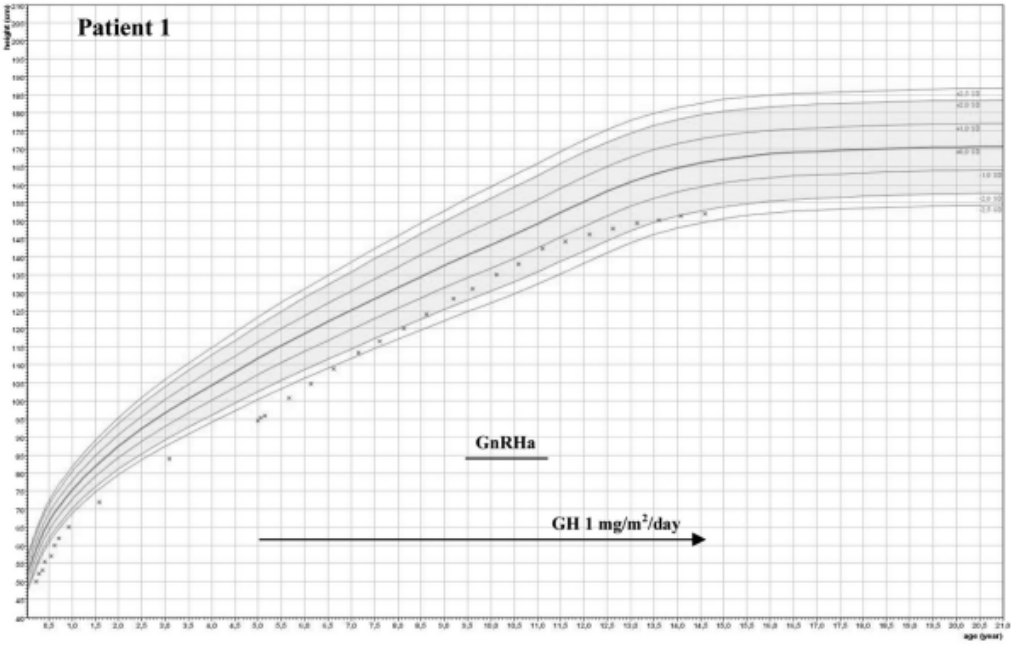
- Can be growth hormone still be considered?
- Are there any specific risks?

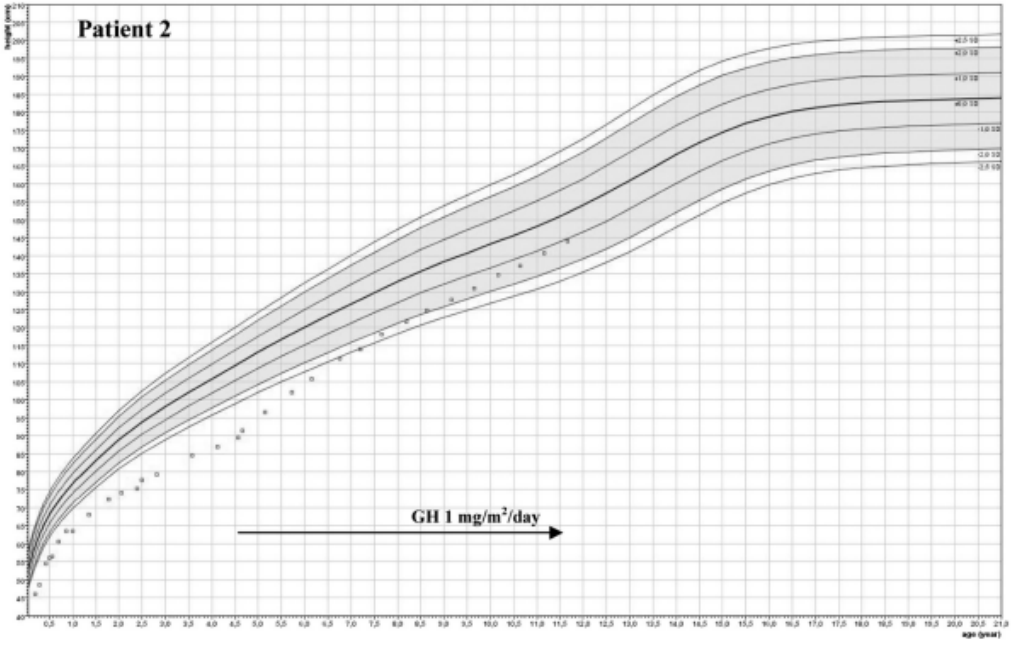
Are Children with Bloom's Syndrome candidates for growth hormone therapy?

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

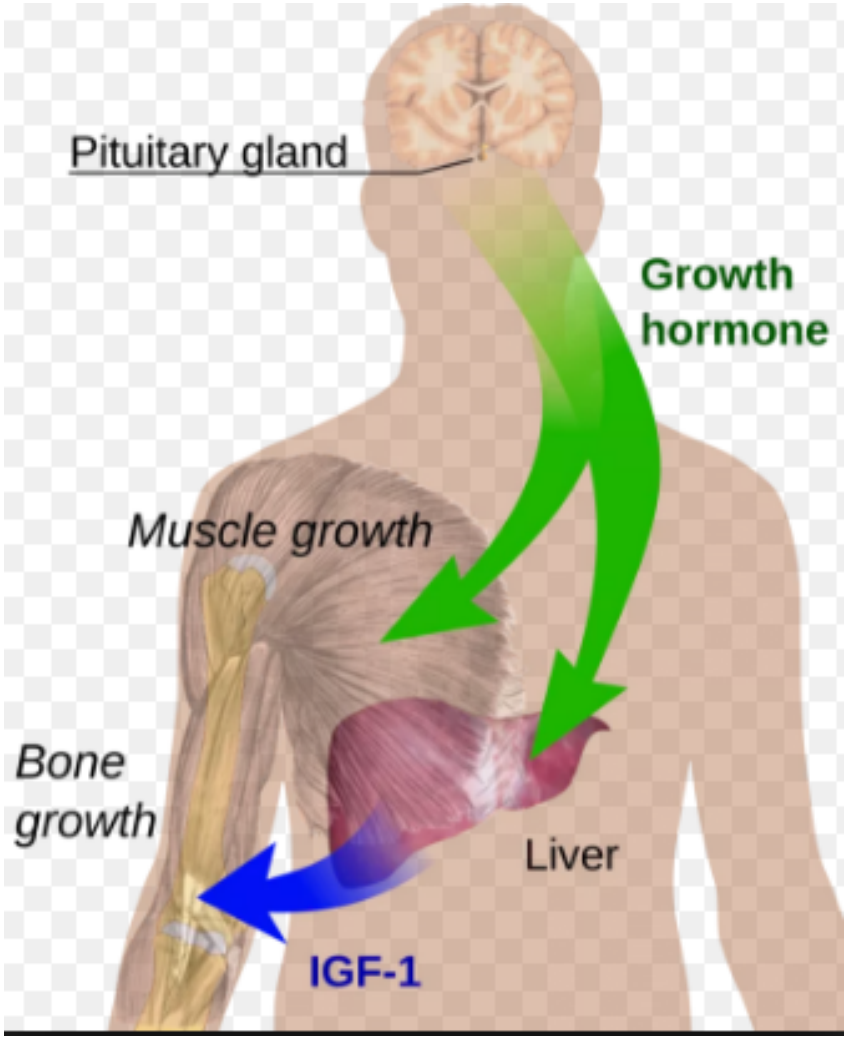
Limited experience in Bloom Syndrome

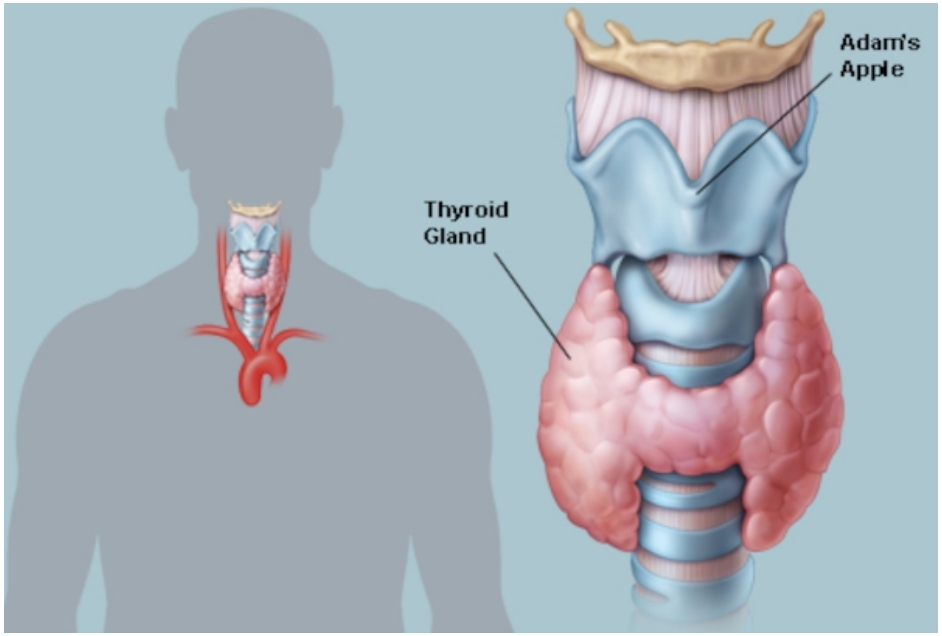
Growth hormone may increase an individual's risk for cancer: a topic of some debate.


Patient 1
GnRHa
GH 1 mg/m²/day


Patient 2
GH 1 mg/m²/day

Monitoring of IGF1 and IGFBP3 levels during growth hormone therapy is suggested.





Thyroid abnormalities in Bloom's syndrome

HYPOthyroidism

Children: Poor growth
Developmental delays

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

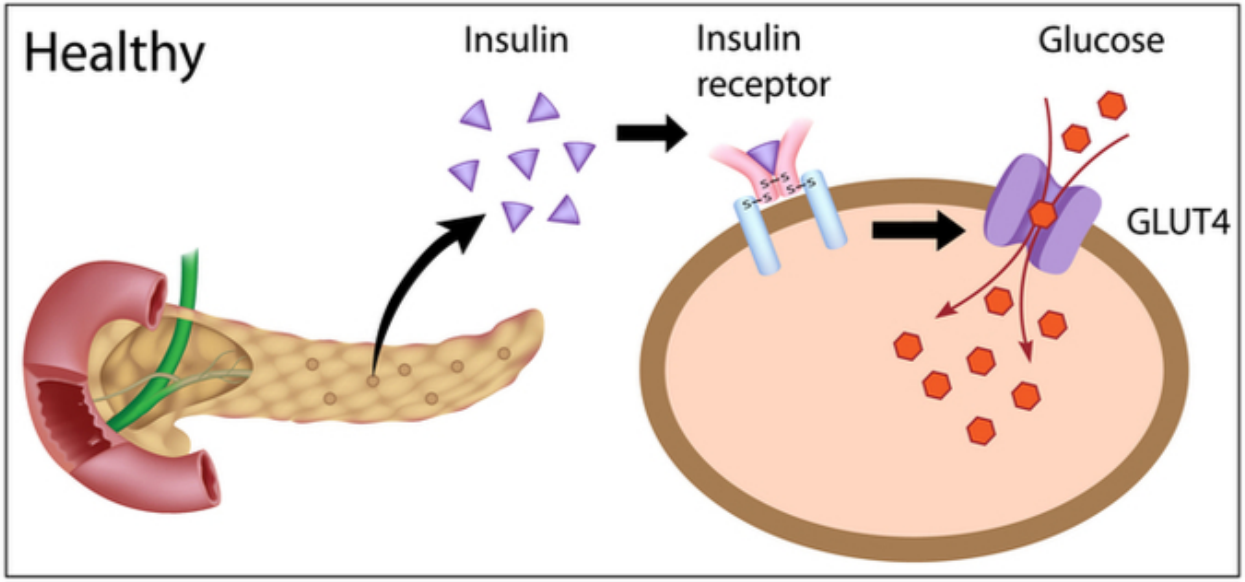
Symptoms can be non-specific.
Annual thyroid testing is suggested after age 10 yrs.

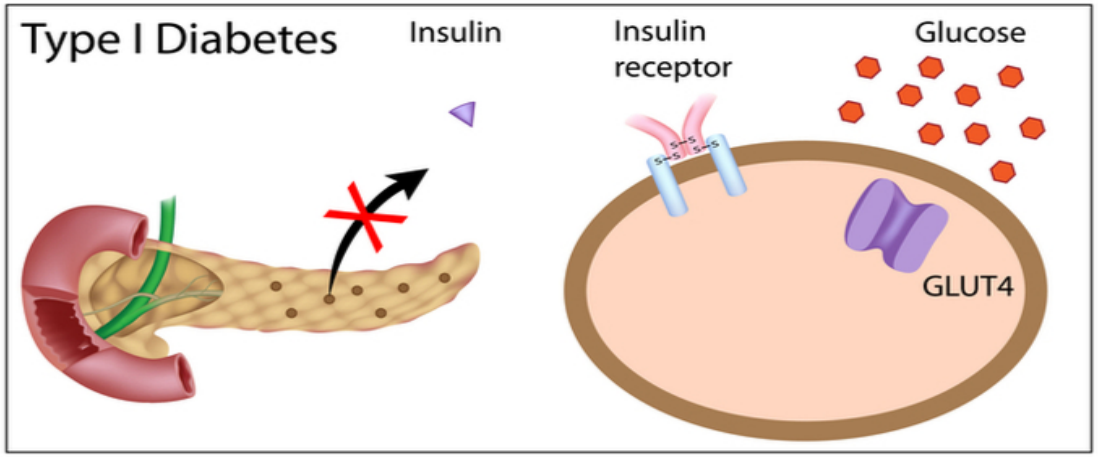

Diabetes

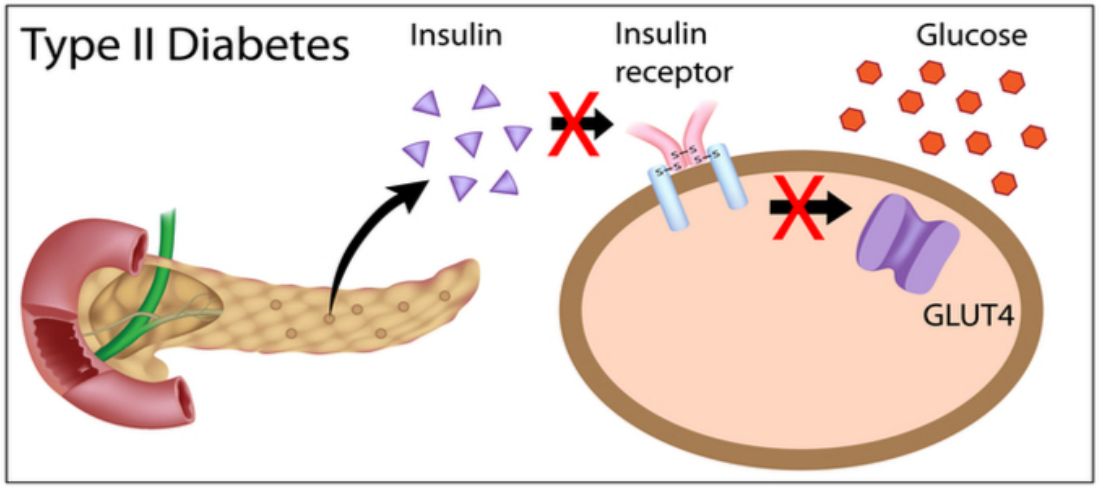
High rates of prediabetes and diabetes with Bloom Syndrome

- Prediabetes starts early in childhood.
- Risk of prediabetes and diabetes increase with age.
- People with prediabetes or diabetes may have no symptoms.

Bloom registry: 18% of people report diabetes - median age 26 years

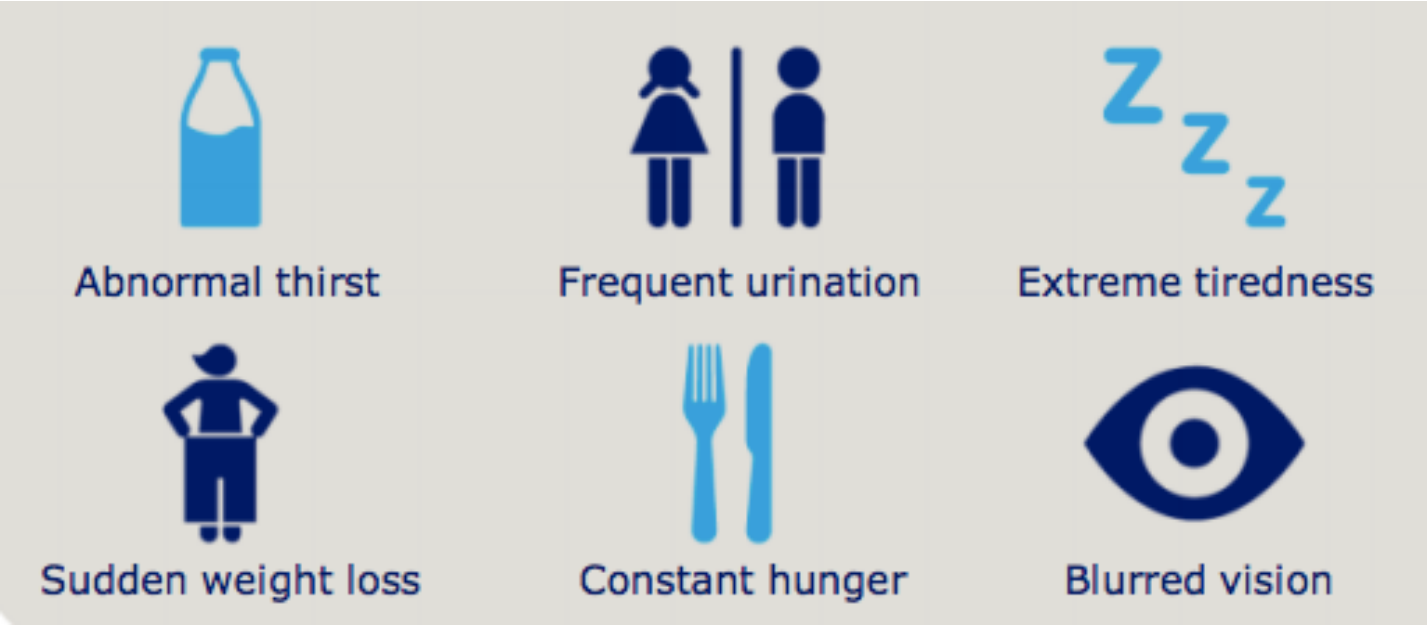

Healthy
Insulin
Insulin receptor
Glucose
GLUT4



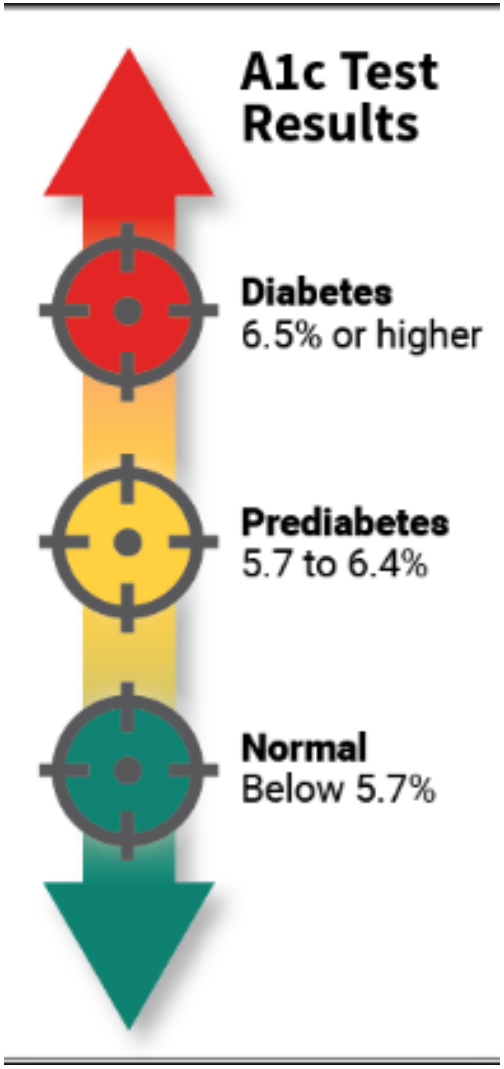


Individuals with Bloom's syndrome have type II Diabetes and insulin resistance

Common Signs of Diabetes




94


A1c Test Results
Diabetes
6.5% or higher
Prediabetes
5.7 to 6.4%
Normal
Below 5.7%


Monitoring
Treatment and Insulin
Regular Physical Activity
Diet Regimen

AND FERTILITY

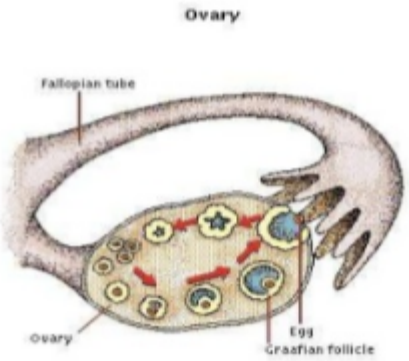
Puberty may be delayed but progresses normally.

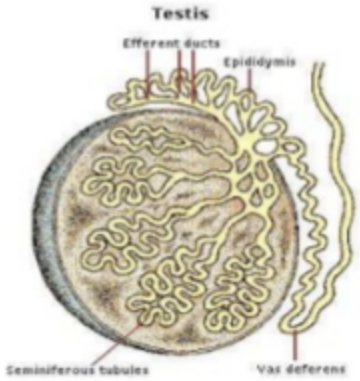
Concern of primary ovarian or testicular problem in Bloom.

- 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.





Research/ Future Directions

Women:
oocyte preservation

Men:
Semen analysis- cryopreservation
Testicular sperm extraction (TESE)

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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WILEY AMERICAN JOURNAL OF medical genetics PART A

ORIGINAL ARTICLE

Health supervision for people with Bloom syndrome

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