

# Take It With A Grain of Salt – Understanding Common Lab Results

*Audience Participation!*  
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*Audience Participation!*  
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Global Down Syndrome Foundation Webinar

April 17, 2024

# Case Scenario

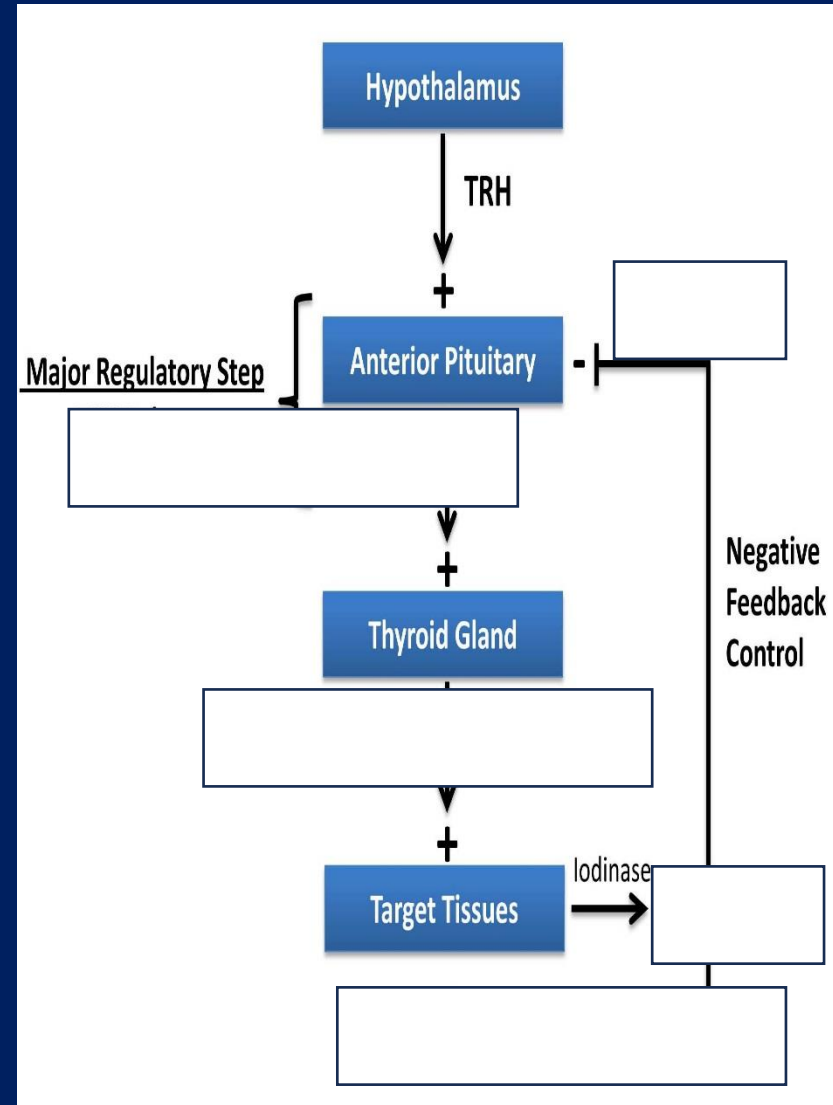
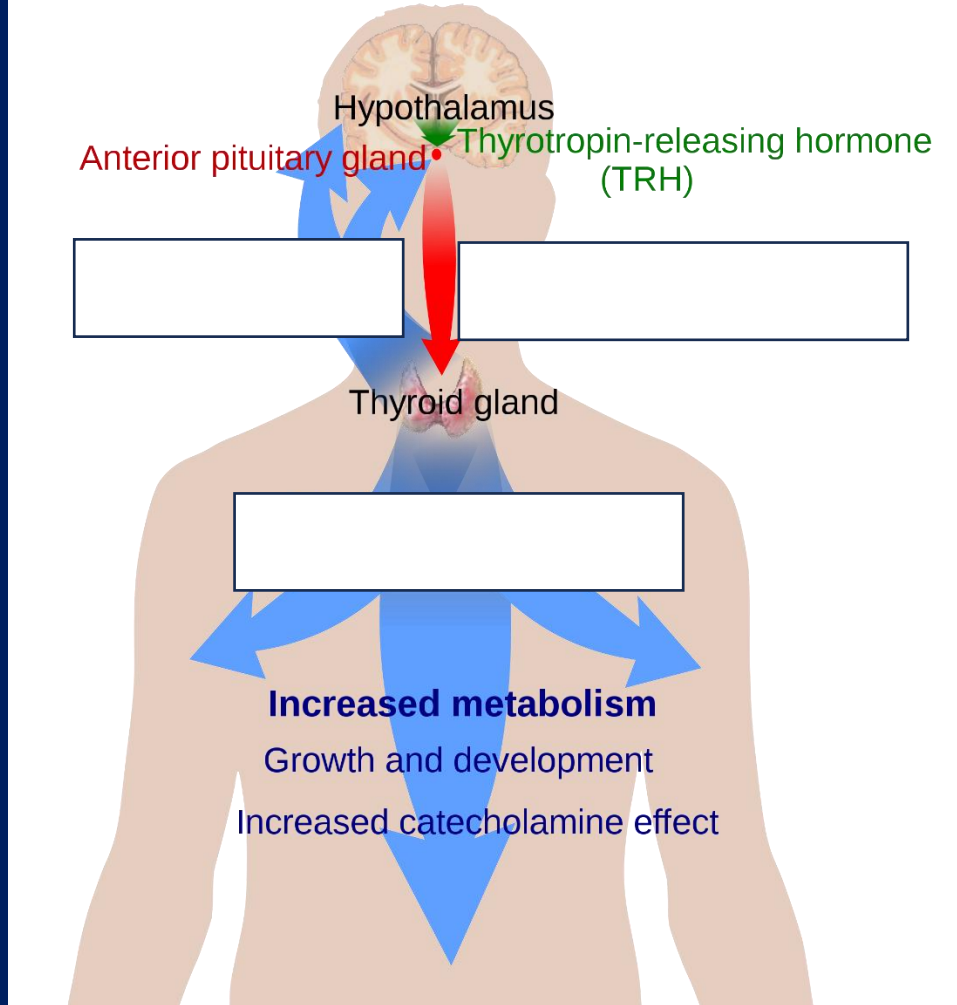
5-year-old male with Down syndrome who has not been seen in the Down Syndrome Center since an initial visit as a newborn. He has been seen by his PCP for annual visits with no concerns reported. When asked, the PCP is not entirely familiar with the AAP healthcare guidelines for children with Ds. The family made this appointment to see if there was anything that was recommended from a Ds screening perspective even if their child is asymptomatic.

# Thyroid Labs - Case Version 1

- Thyroid results were obtained:

TSH	Free T4
85.4 mU/L (nl 0.55-5.31)	0.5 ng/dL (nl 0.8-1.8)

# Thyroid system



# Hypothyroidism

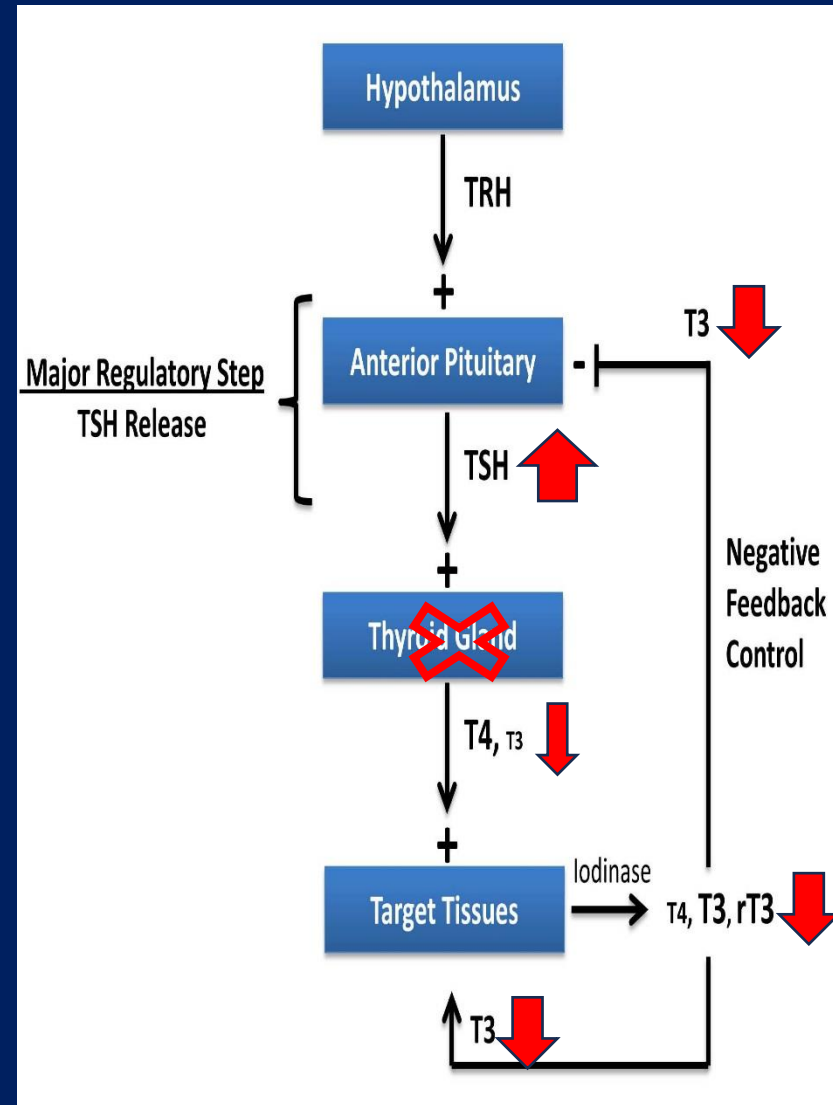
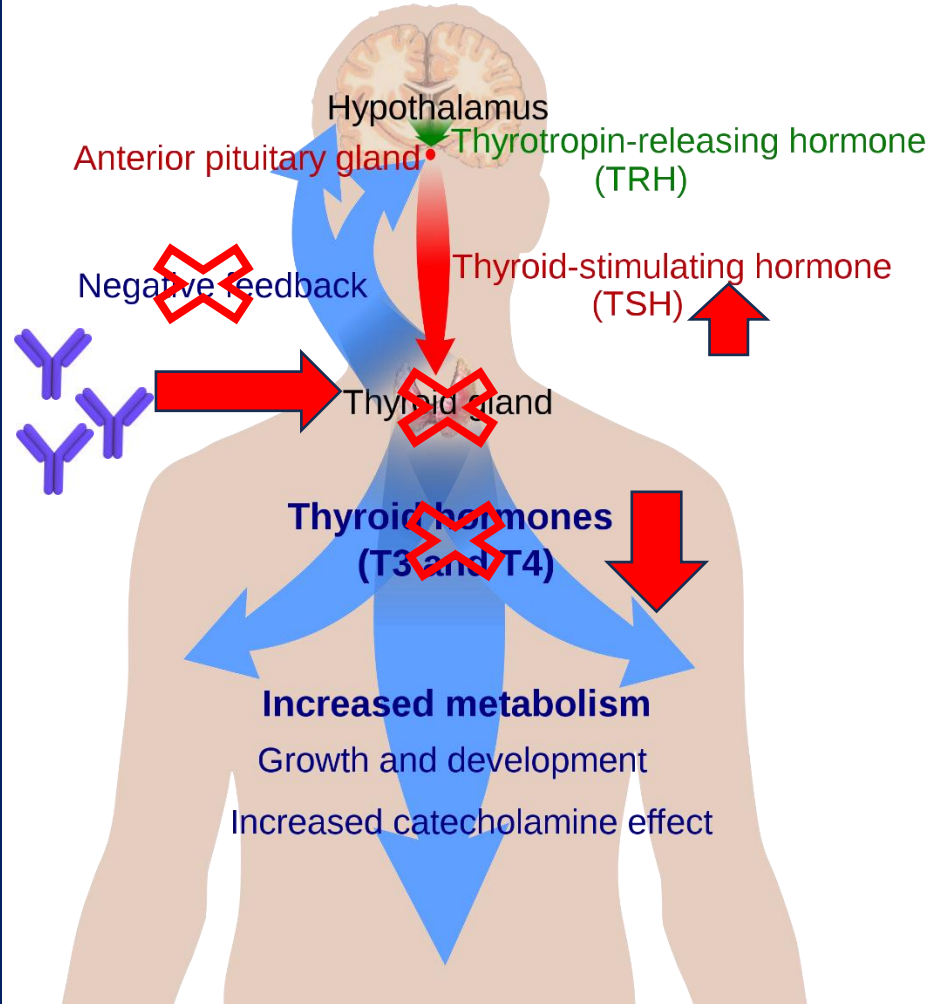
- Hypothyroidism
  - Congenital (1-1.5%)
    - 28-35X the rate for the typical population
  - Acquired/autoimmune (~13-34% of Ds with positive antibodies)
    - Hashimoto thyroiditis
    - Diagnosis confirmed by sending thyroid peroxidase (TPO) and thyroglobulin (TG) antibodies
- Best managed by Endocrinology
  - Patients can sometimes initially present with rapidly changing thyroid levels between hypothyroidism and hyperthyroidism

# Hypothyroidism Symptoms

- Constipation
- Dry skin
- Weight gain (slow weight gain in infants/younger children, rapid weight gain in older children)
- Fatigue
- Feeling cold frequently
- Dry, brittle hair
- Slow heart rate

# HASHIMOTO THYROIDITIS

## Thyroid system



# Thyroid Labs - Case Version 2

- Thyroid results were obtained:

TSH	Free T4
0.13 mU/L (0.55-5.31)	2.6 ng/dL (0.8-1.8)



# Hyperthyroidism

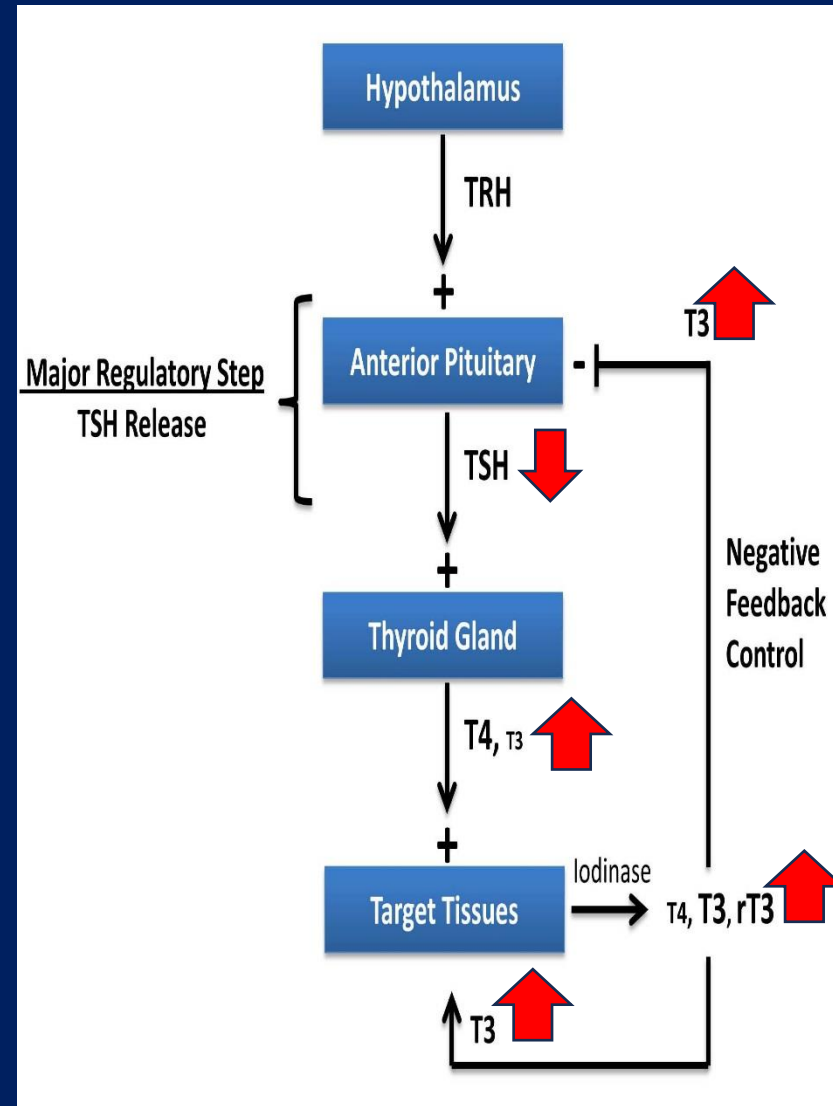
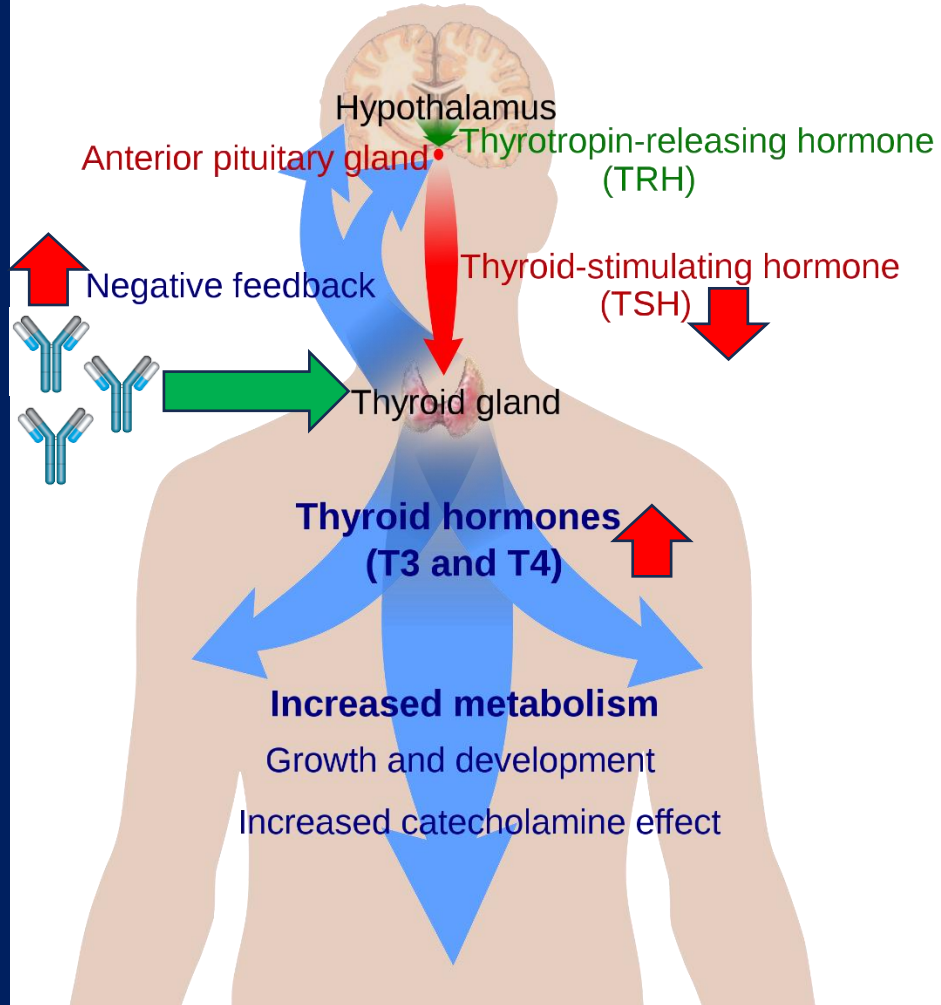
- Hyperthyroidism
  - Graves disease – autoimmune attack on thyroid gland that INCREASES the activity of the gland
  - 0.66% in Ds, 0.02% in general
- Diagnosis confirmed by sending TSH Receptor Antibody (TRAB)
- Requires Endocrinology management

# Hyperthyroidism Symptoms

- Diarrhea
- Clammy skin
- Poor growth, weight loss
- Fatigue or High Energy
- Feeling hot
- Hair loss
- High heart rate

# GRAVES DISEASE

## Thyroid system



# Thyroid Labs - Case Version 3

- Thyroid results were obtained:

TSH	Free T4
8.4 mU/L (0.55-5.31)	1.2 ng/dL (0.8-1.8)

# Subclinical/Compensated Hypothyroidism (Hyperthyrotropinemia)

- 7-40% of people with Ds (depends on definition used)
- Definition most commonly used:
  - High TSH (but  $<10$  mU/L)
  - Normal free T4
- Our approach
  - Repeat TSH, free T4, thyroid antibodies (TPO, TG) in 3 months
- Often returns to normal, but can later become hypothyroidism
- No evidence to start treatment with thyroid hormone unless becomes true hypothyroidism

# CBC – Case Version 1

LAB	RESULT	NORMAL
White Blood Cells (WBC)	3.7 x10 <sup>9</sup> /L	4.5– 14.5 x 10 <sup>9</sup> /L
Hemoglobin (Hb)	12.7 g/L	11.5 - 15.5 g/dL
Mean Corpuscular Volume (MCV)	91.8 fL	77 – 95.0 fL
Red Cell Distribution Width (RDW)	14%	11.8 – 15.2%
Platelet (Plt)	315	156– 369 x 10 <sup>9</sup> /L
Neutrophils (N)	45%	26 - 48%
Lymphocytes (L)	45%	35 - 65%
Monocytes (M)	6%	3 - 9%
Basophils (B)	2%	0 - 2%
Eosinophils (E)	2%	0 - 2%

# Leukopenia (low WBC count)

- Common in people with Down syndrome
- WBC count typically about 30-40% lower than general population
- Unless other findings on history or exam, isolated leukopenia is not a concern
- Theories
  - More frequent thymus removal (due to cardiac surgery)
  - Higher levels of inflammatory molecules

# CBC – Case Version 2

LAB	RESULT	NORMAL
White Blood Cells (WBC)	4.7 x10 <sup>9</sup> /L	4.5– 14.5 x 10 <sup>9</sup> /L
Hemoglobin (Hb)	12.7 g/L	11.5 - 15.5 g/dL
Mean Corpuscular Volume (MCV)	91.8 fL	77 – 95.0 fL
Red Cell Distribution Width (RDW)	14%	11.8 – 15.2%
Platelet (Plt)	315	156– 369 x 10 <sup>9</sup> /L
Neutrophils (N)	22%	26 - 48%
Lymphocytes (L)	65%	35 - 65%
Monocytes (M)	9%	3 - 9%
Basophils (B)	2%	0 - 2%
Eosinophils (E)	2%	0 - 2%
Absolute Neutrophil Count (ALC)	1.034 x10 <sup>9</sup> /L	1.5 – 8.0 x10 <sup>9</sup> /L



# Neutropenia (low neutrophil count)

- Common in people with Down syndrome (especially <6 years old)
- Levels in those under 6 about 30% less than general population
- Unless other findings on history or exam, typically not a concern

# CBC – Case Version 3

LAB	RESULT	NORMAL
White Blood Cells (WBC)	4.7 x10 <sup>9</sup> /L	4.5– 14.5 x 10 <sup>9</sup> /L
Hemoglobin (Hb)	12.7 g/L	11.5 - 15.5 g/dL
Mean Corpuscular Volume (MCV)	91.8 fL	77 – 95.0 fL
Red Cell Distribution Width (RDW)	14%	11.8 – 15.2%
Platelet (Plt)	315	156– 369 x 10 <sup>9</sup> /L
Neutrophils (N)	65%	26 - 48%
Lymphocytes (L)	22%	35 - 65%
Monocytes (M)	9%	3 - 9%
Basophils (B)	2%	0 - 2%
Eosinophils (E)	2%	0 - 2%
Absolute Lymph Count (ALC)	1.034 x10 <sup>9</sup> /L	1.5 – 8.0 x10 <sup>9</sup> /L

# Lymphopenia (low lymphocyte count)

- Common in people with Down syndrome
- Levels can average 50% less than general population
- Unless other findings on history or exam, typically not a concern
- Theories
  - More frequent thymus removal (due to cardiac surgery)
  - Higher levels of inflammatory molecules

# CBC – Case Version 4

LAB	RESULT	NORMAL
White Blood Cells (WBC)	4.7 x10 <sup>9</sup> /L	4.5– 14.5 x 10 <sup>9</sup> /L
Hemoglobin (Hb)	11.7 g/L	11.5 - 15.5 g/dL
Mean Corpuscular Volume (MCV)	78 fL	77 – 95.0 fL
Red Cell Distribution Width (RDW)	16.5 %	11.8 – 15.2%
Platelet (Plt)	315	156– 369 x 10 <sup>9</sup> /L
Neutrophils (N)	45%	26 - 48%
Lymphocytes (L)	45%	35 - 65%
Monocytes (M)	6%	3 - 9%
Basophils (B)	2%	0 - 2%
Eosinophils (E)	2%	0 - 2%

# Iron Studies – Case Version 4

LAB	RESULT	NORMAL
Ferritin	6 ng/mL	10 – 60 ng/mL
Total Iron Binding Capacity	470 ug/dL	250 – 400 ug/dL
Transferrin	380 mg/dL	120 – 315 mg/dL

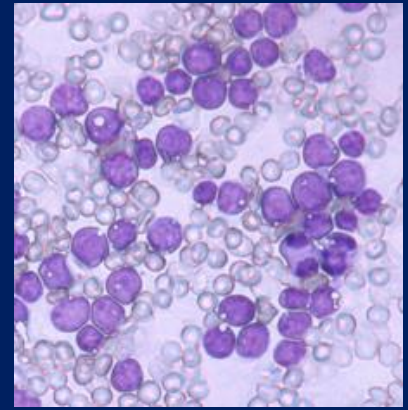
# Iron Deficiency Anemia

- More common in Down syndrome?
  - Unclear
  - Picky eating, texture aversions → limited iron intake
  - Celiac disease → poor iron absorption
- Difficulty in diagnosis
  - Lab values that are typically used to diagnose in other children are not reliable in children with Ds

# CBC – Case Version 5

LAB	RESULT	NORMAL
White Blood Cells (WBC)	1.7 x10 <sup>9</sup> /L	4.5– 14.5 x 10 <sup>9</sup> /L
Hemoglobin (Hb)	8.7 g/L	11.5 - 15.5 g/dL
Mean Corpuscular Volume (MCV)	85 fL	77 – 95.0 fL
Red Cell Distribution Width (RDW)	12.5 %	11.8 – 15.2%
Platelet (Plt)	75	156– 369 x 10 <sup>9</sup> /L
Neutrophils (N)	15%	26 - 48%
Lymphocytes (L)	65%	35 - 65%
Monocytes (M)	6%	3 - 9%
Basophils (B)	2%	0 - 2%
Eosinophils (E)	2%	0 - 2%
Other/Blasts	15%	0%

# Leukemia



- Acute myeloid leukemia (AML)
  - Most often, a specific type of leukemia called myeloid leukemia of Down syndrome (ML DS)
  - Younger age than most children with AML
  - Very sensitive to lower dose chemotherapy specifically adapted for Ds
    - 80% long-term survival
- Acute lymphoblastic leukemia (ALL)
  - Ds leads to less favorable prognosis than in other children and higher toxic side effects to chemotherapy
    - 80% long-term survival



# CBC – Case Version 6

LAB	RESULT	NORMAL
White Blood Cells (WBC)	6.5 x10 <sup>9</sup> /L	4.5– 14.5 x 10 <sup>9</sup> /L
Hemoglobin (Hb)	17.1 g/L	11.5 - 15.5 g/dL
Mean Corpuscular Volume (MCV)	85 fL	77 – 95.0 fL
Red Cell Distribution Width (RDW)	12.5 %	11.8 – 15.2%
Platelet (Plt)	275	156– 369 x 10 <sup>9</sup> /L
Neutrophils (N)	29%	26 - 48%
Lymphocytes (L)	61%	35 - 65%
Monocytes (M)	6%	3 - 9%
Basophils (B)	2%	0 - 2%
Eosinophils (E)	2%	0 - 2%

# Polycythemia (High hemoglobin level) in Ds

## Self-resolving

- Dehydration at time of lab draw

## Low Oxygen

- High altitude
- Unrepaired cardiac defect
- Obstructive sleep apnea

# Case Scenario (Update Version 1)

The 5-year-old child's parent calls because she noticed that you did not send a celiac screening so now the PCP needs to draw blood again based on the guideline recommendation to check celiac labs. You review the chart and find that the child was growing well with no concerning GI symptoms.

# Case Scenario (Update Version 2)

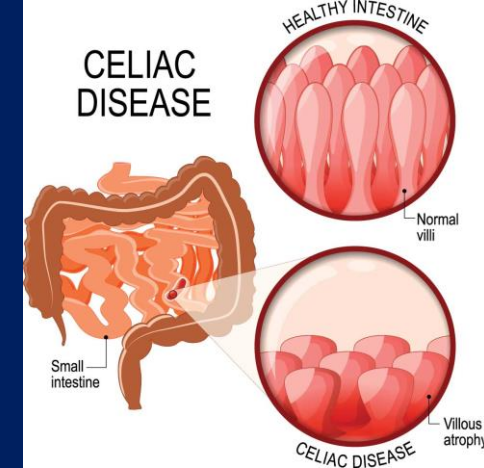
The 5-year-old's labs have come back normal but over the past few weeks he has developed loose stools, several times daily with abdominal bloating and decreased appetite. Review of the records reveals that he had a celiac screening at 2 years old that was normal.

# Celiac Studies – Update Version 2

LAB	RESULT	NORMAL
IgA	76 mg/dL	15 – 144 mg/mL
Tissue Transglutaminase IgA (tTG IgA)	150	<15

# Celiac Disease

- Sensitivity to gluten resulting in antibody attack on intestinal cells
- IgA = type of antibody
- Tissue Transglutaminase = enzyme which helps in protein digestion
  - IgA antibody against this enzyme is found 98% of people with celiac disease
    - Rarely can have celiac disease even if test is negative
    - GI scope with biopsies required for definitive diagnosis
- Up to 15% of people with Ds will have a positive screening
  - Most will be asymptomatic with no known benefit from a restricted diet
  - Can change from negative to positive test over time and exposure



# Down Syndrome Center of Western PA

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## HELPFUL WEBSITES:

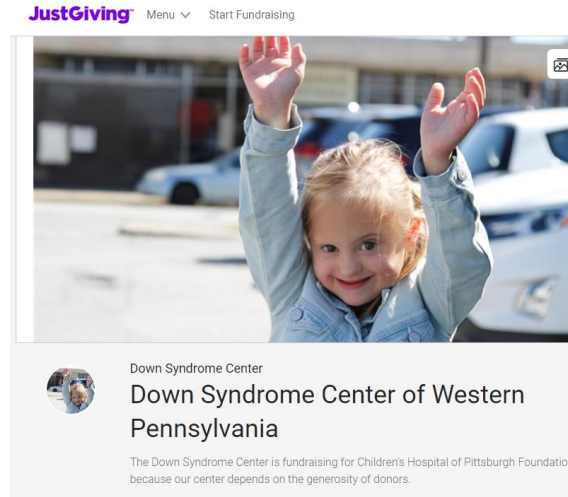
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