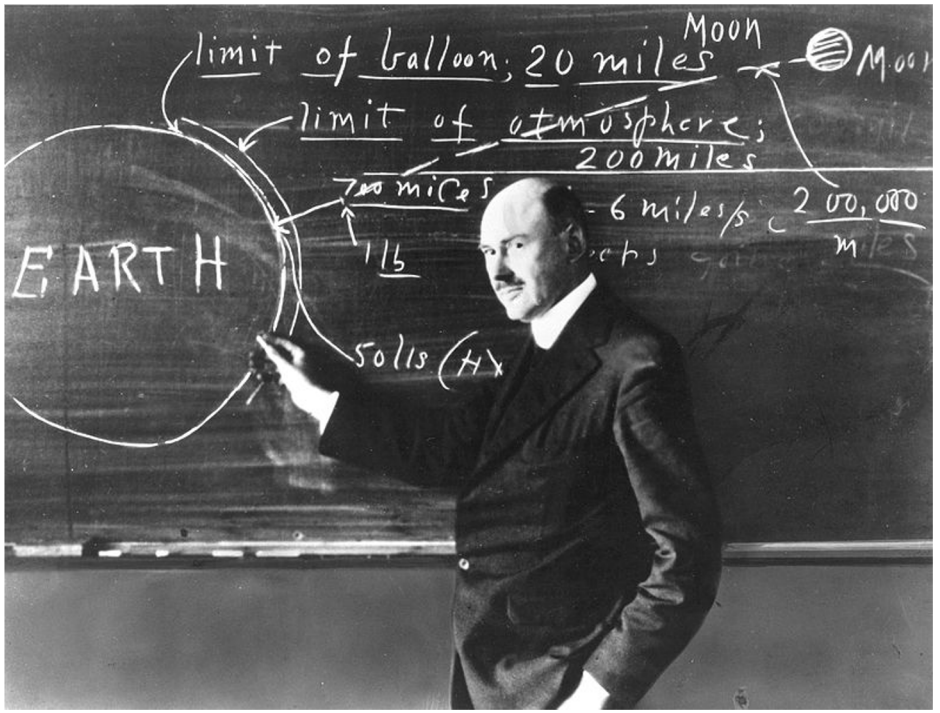
Genetic Testing:
It's Not Rocket Science



Daryl Scott MD/PhD
Baylor College of Medicine

One Important Question

Evaluation

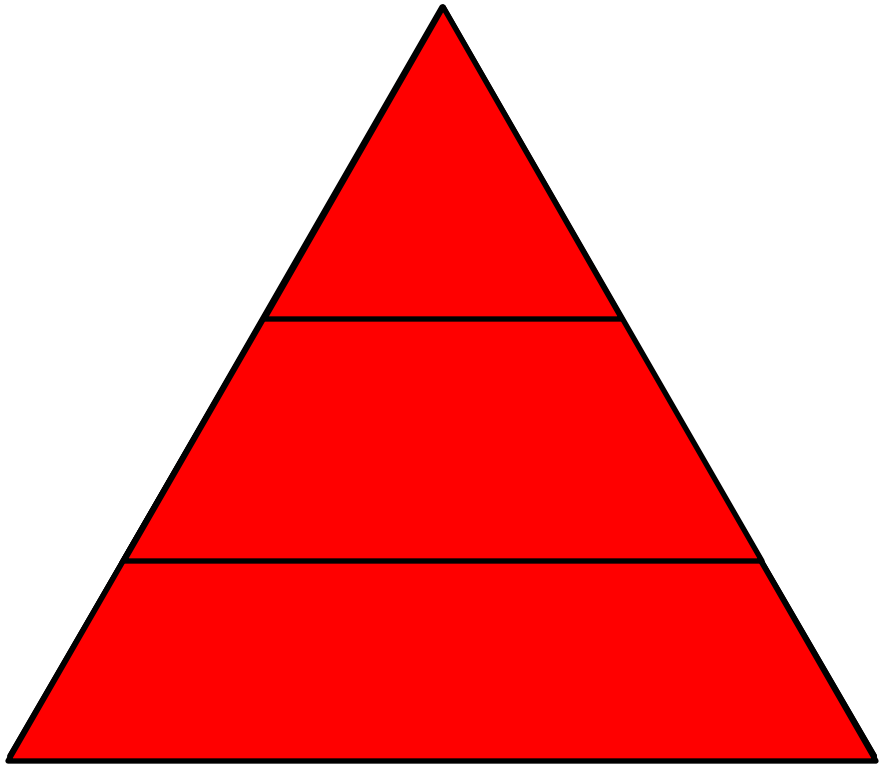
- Medical history
- Family history
- Physical exam

Do I know the diagnosis?

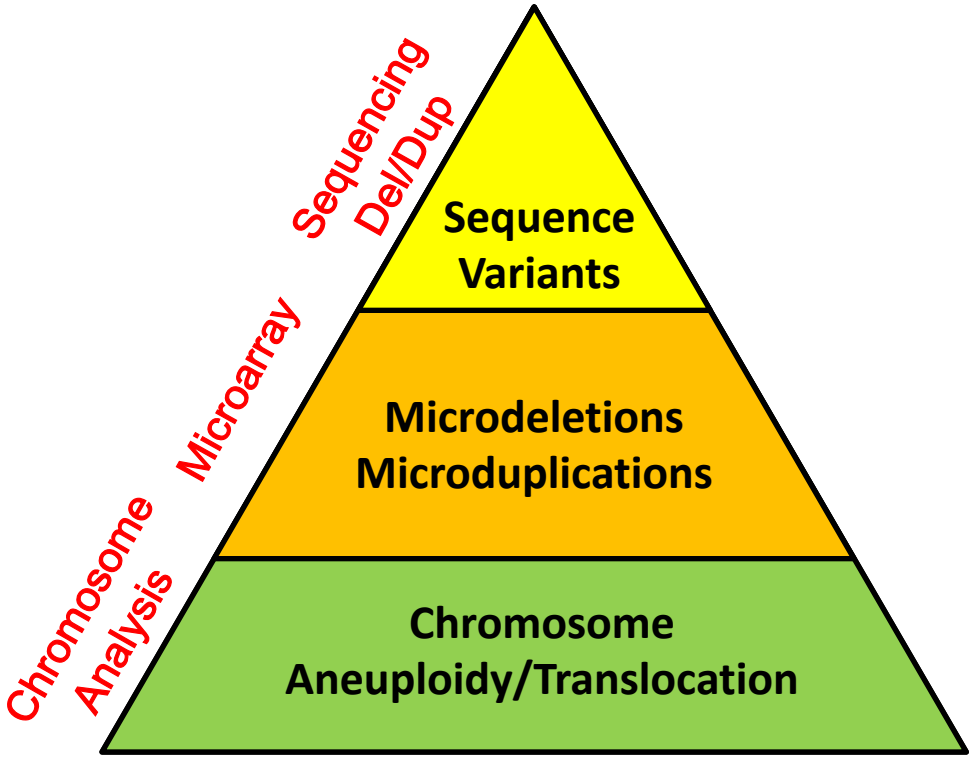
Yes—Triangle Circle

No—3Gs

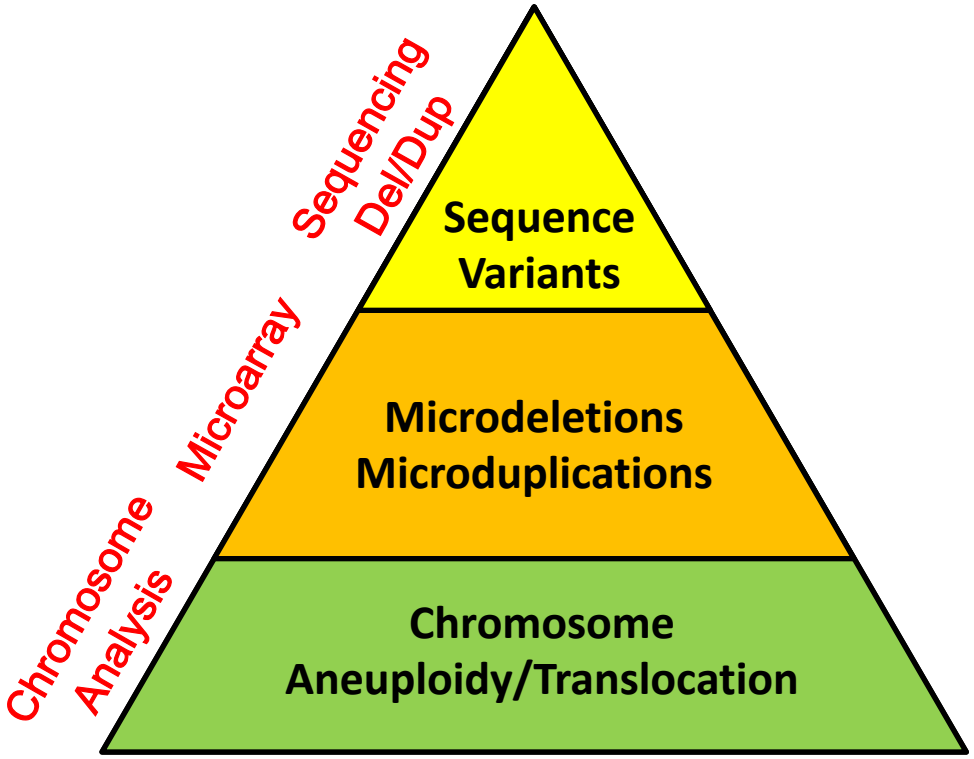
I Know the Diagnosis



I Know the Diagnosis

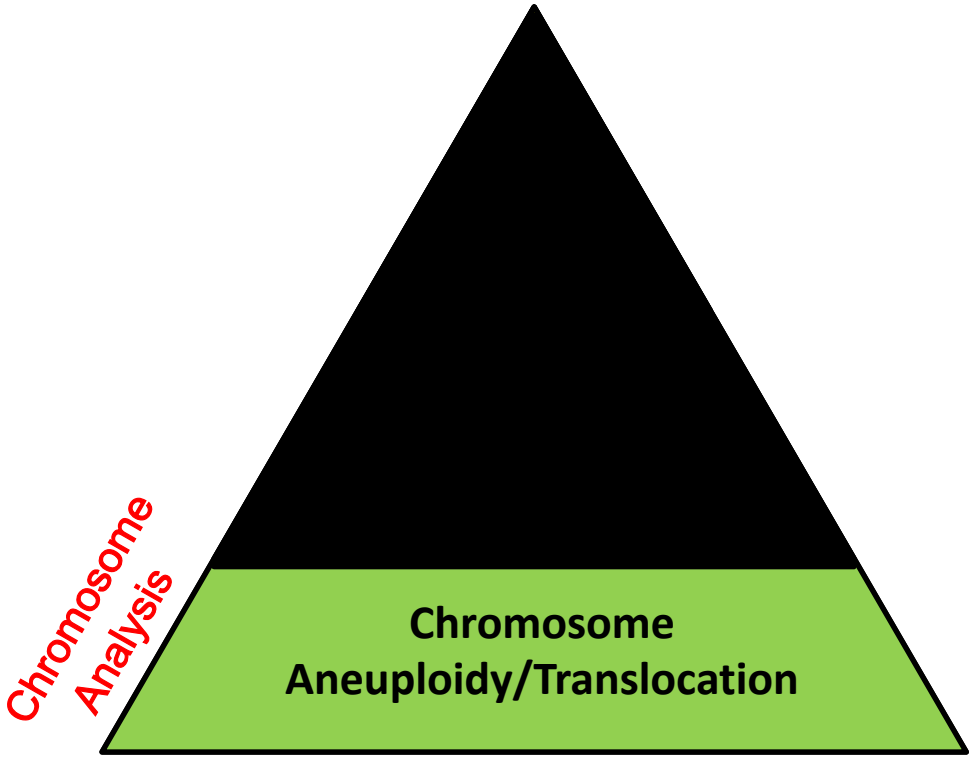


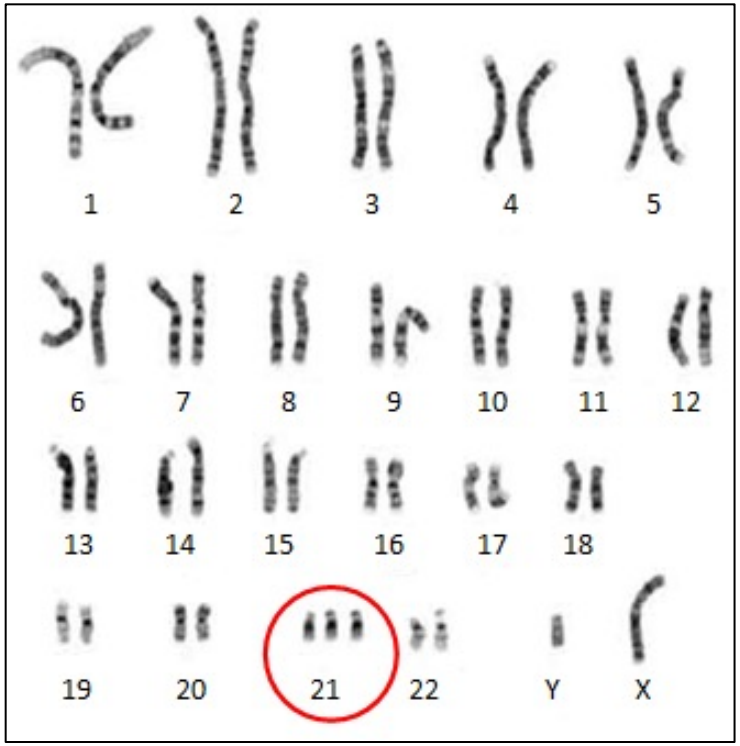
I Know the Diagnosis



Down syndrome

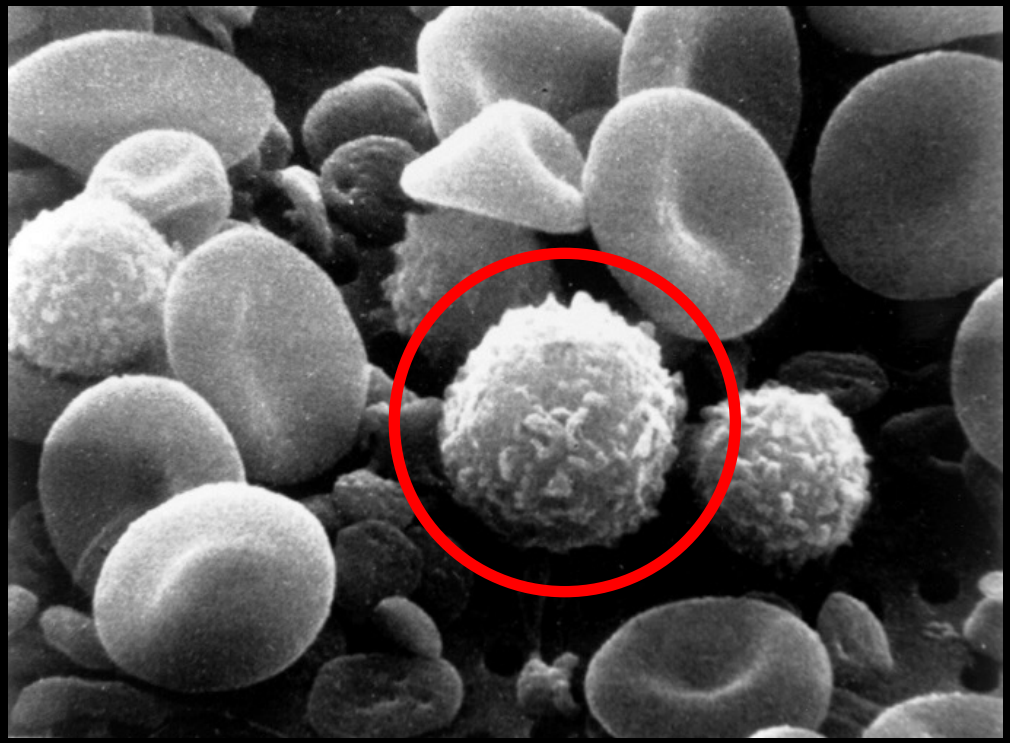
I Know the Diagnosis





Down syndrome

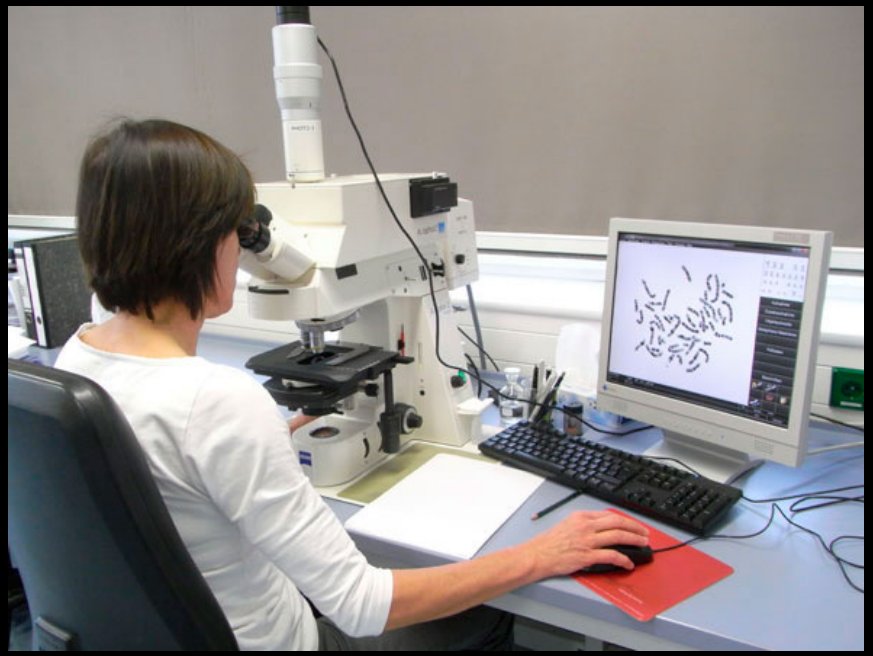
Chromosome Analysis

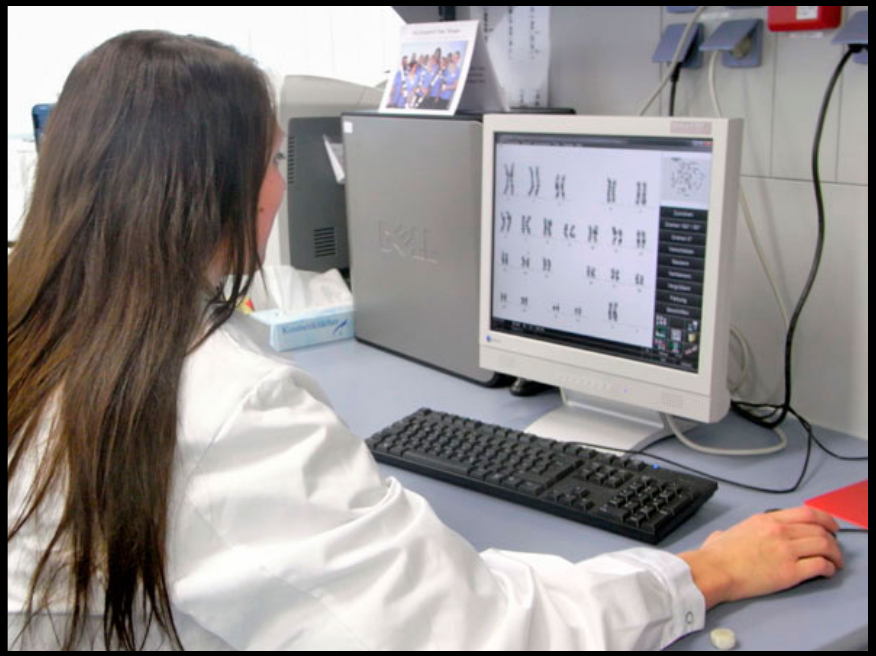


Blood sample
White blood cells
Grow up in culture

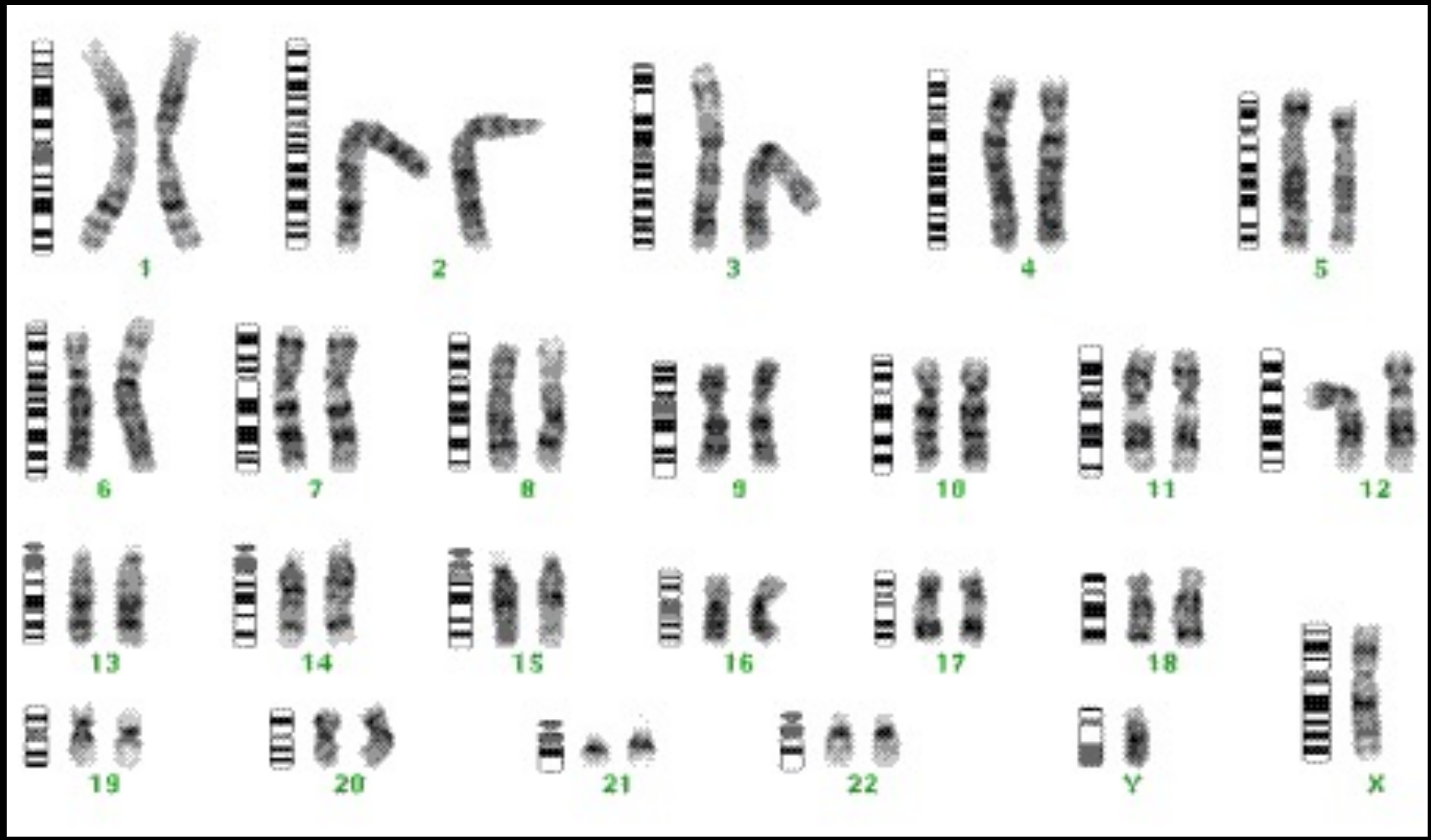
Chromosome Analysis

Chromosome Analysis



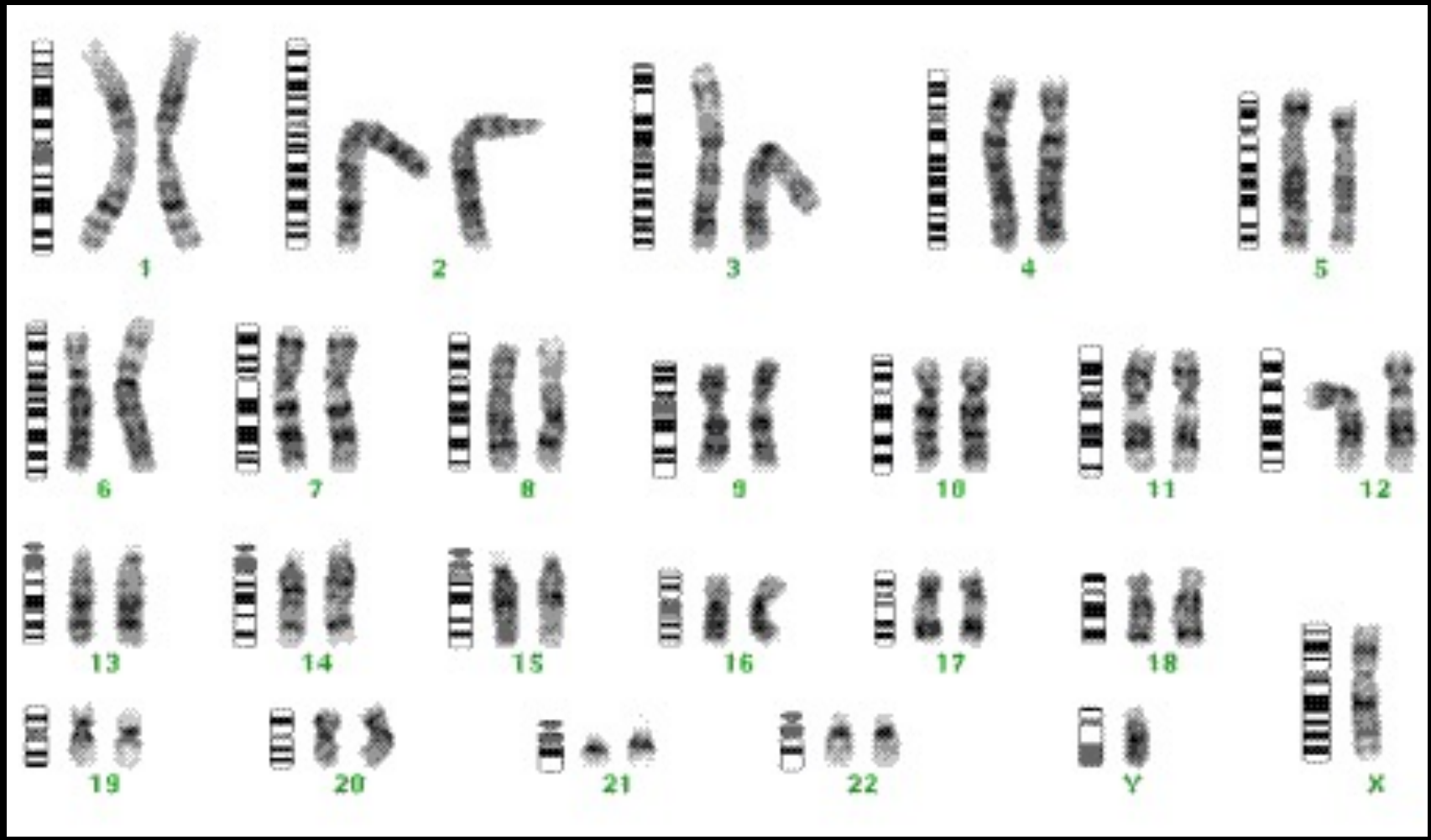


Chromosome Analysis



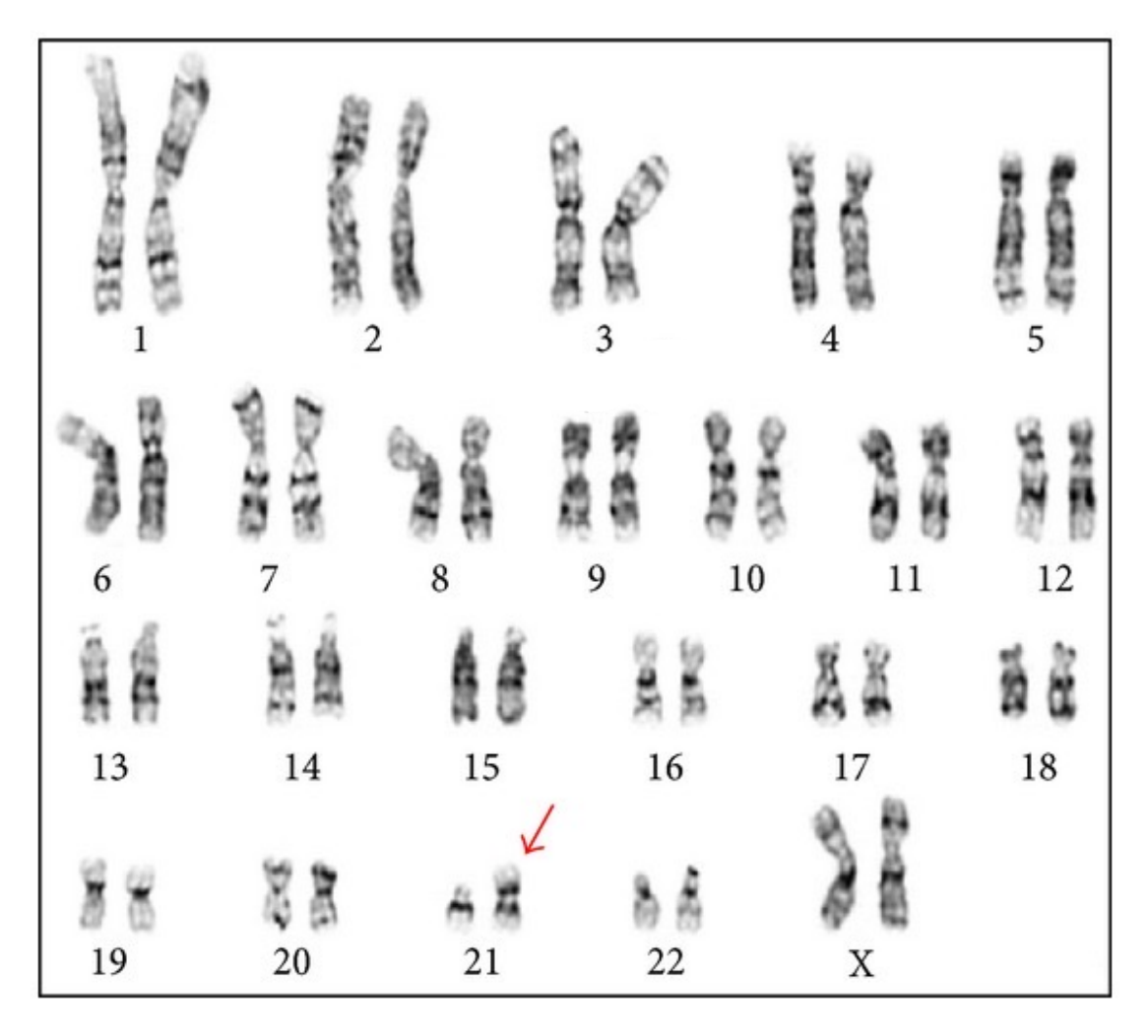
Karyotype = The Picture

Chromosome Analysis



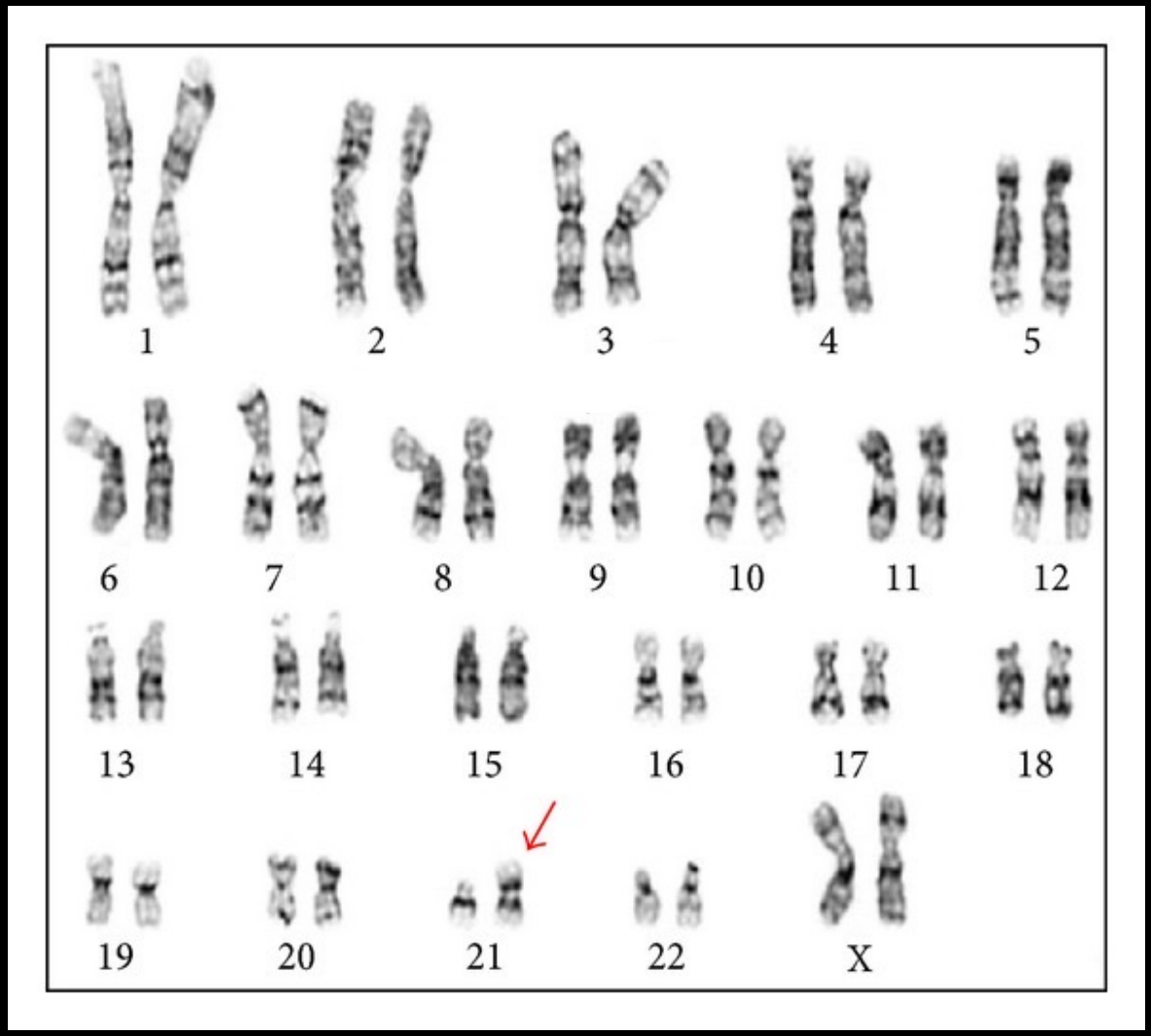
Resolution = 5 Million Bases (5Mb)

Chromosome Analysis



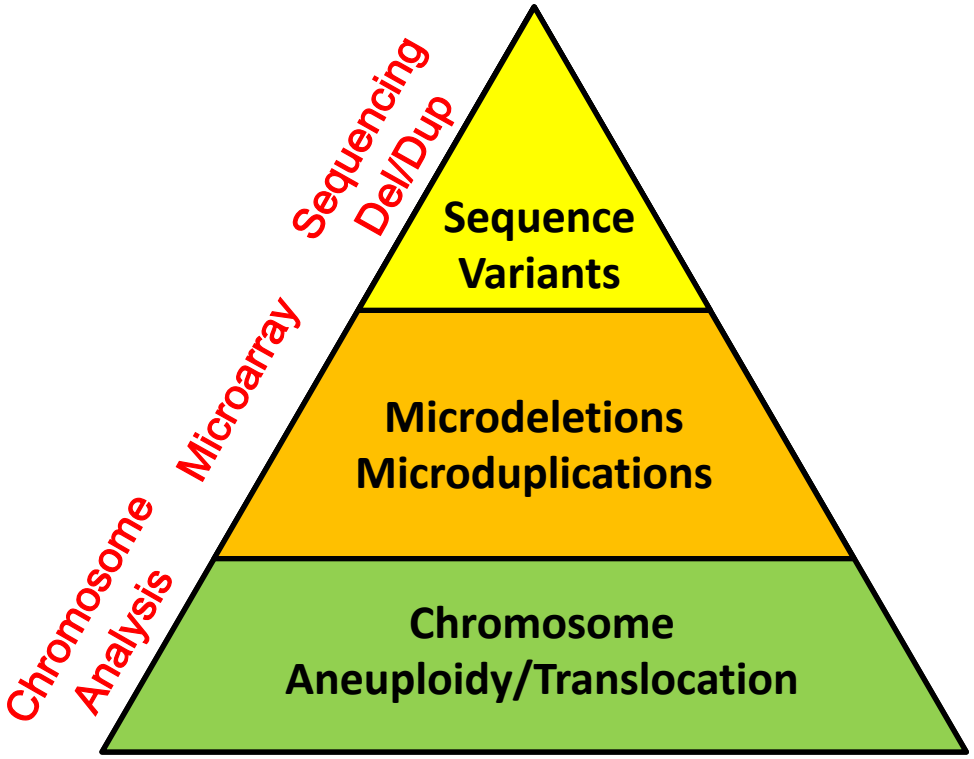
Chromosomal rearrangements

Chromosome Analysis



Up to 100% recurrence risk!

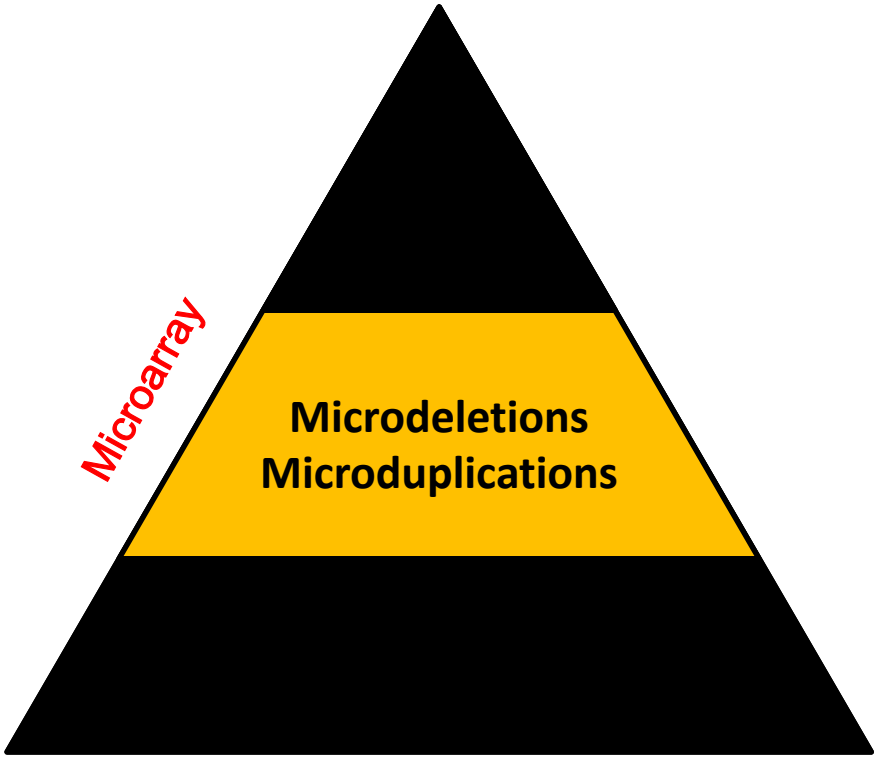
I Know the Diagnosis

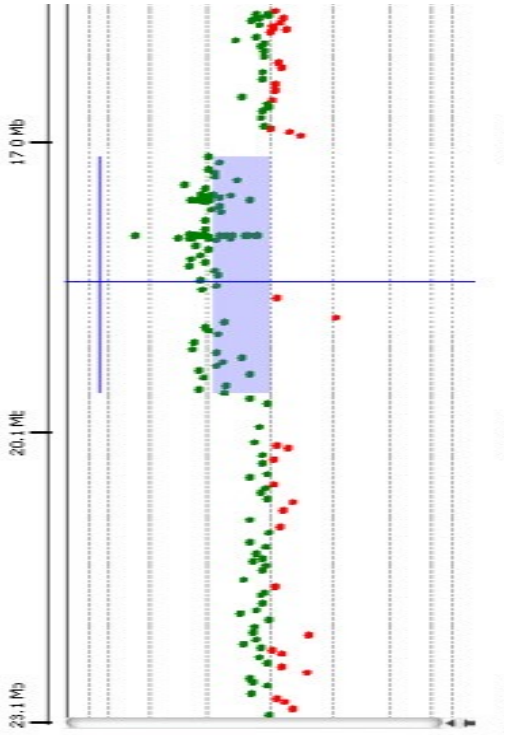


Boy: Tetralogy of Fallot, hypocalcemia, nasal voice, intellectual disability

DiGeorge syndrome

I Know the Diagnosis





Deletion
Chromosome 22q11.2

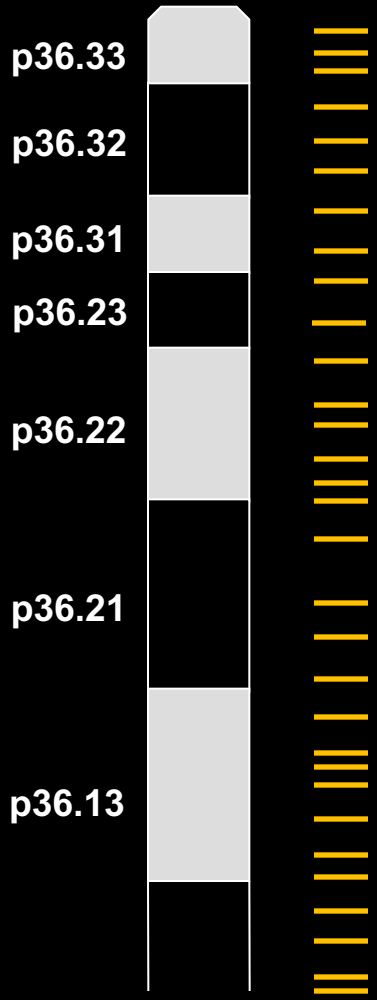
DiGeorge syndrome

Chromosomal Microarray

DNA microarray with thousands of probes

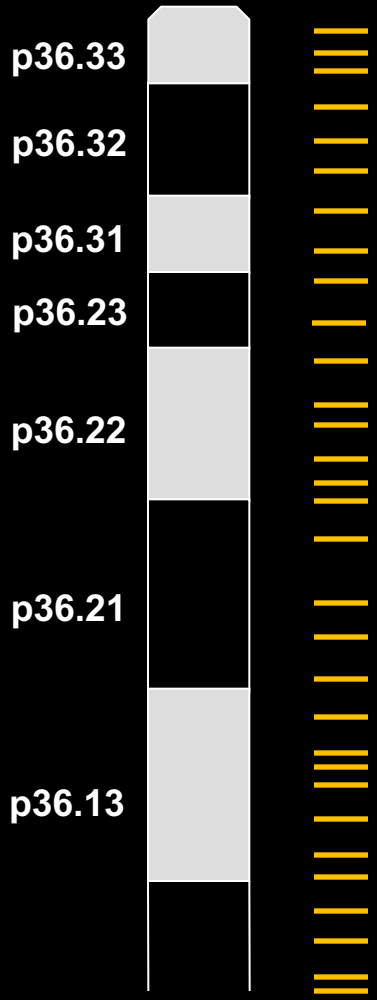
Chromosomal Microarray

DNA microarray with thousands of probes

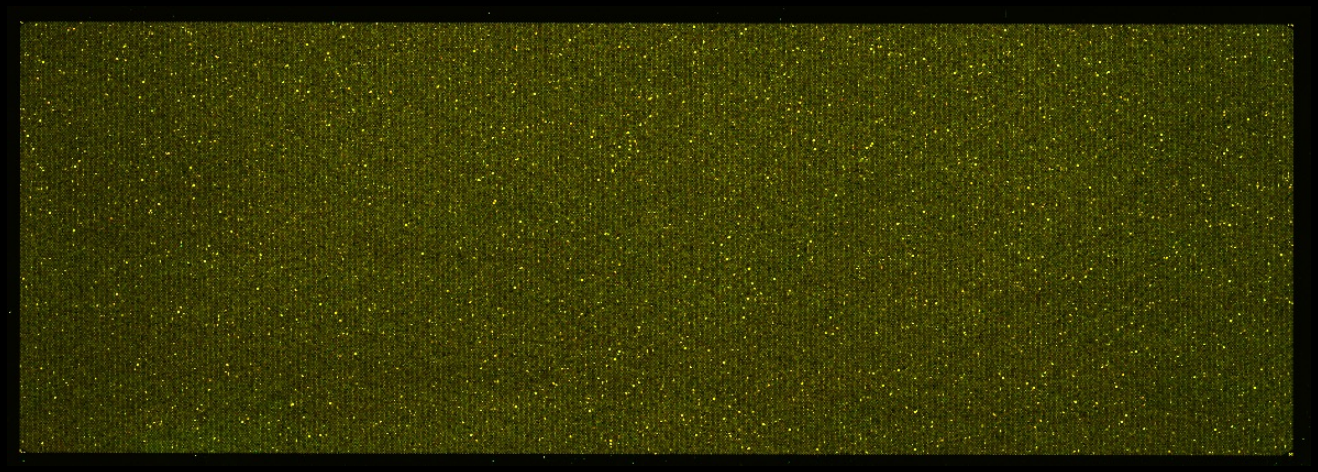


Chromosomal Microarray

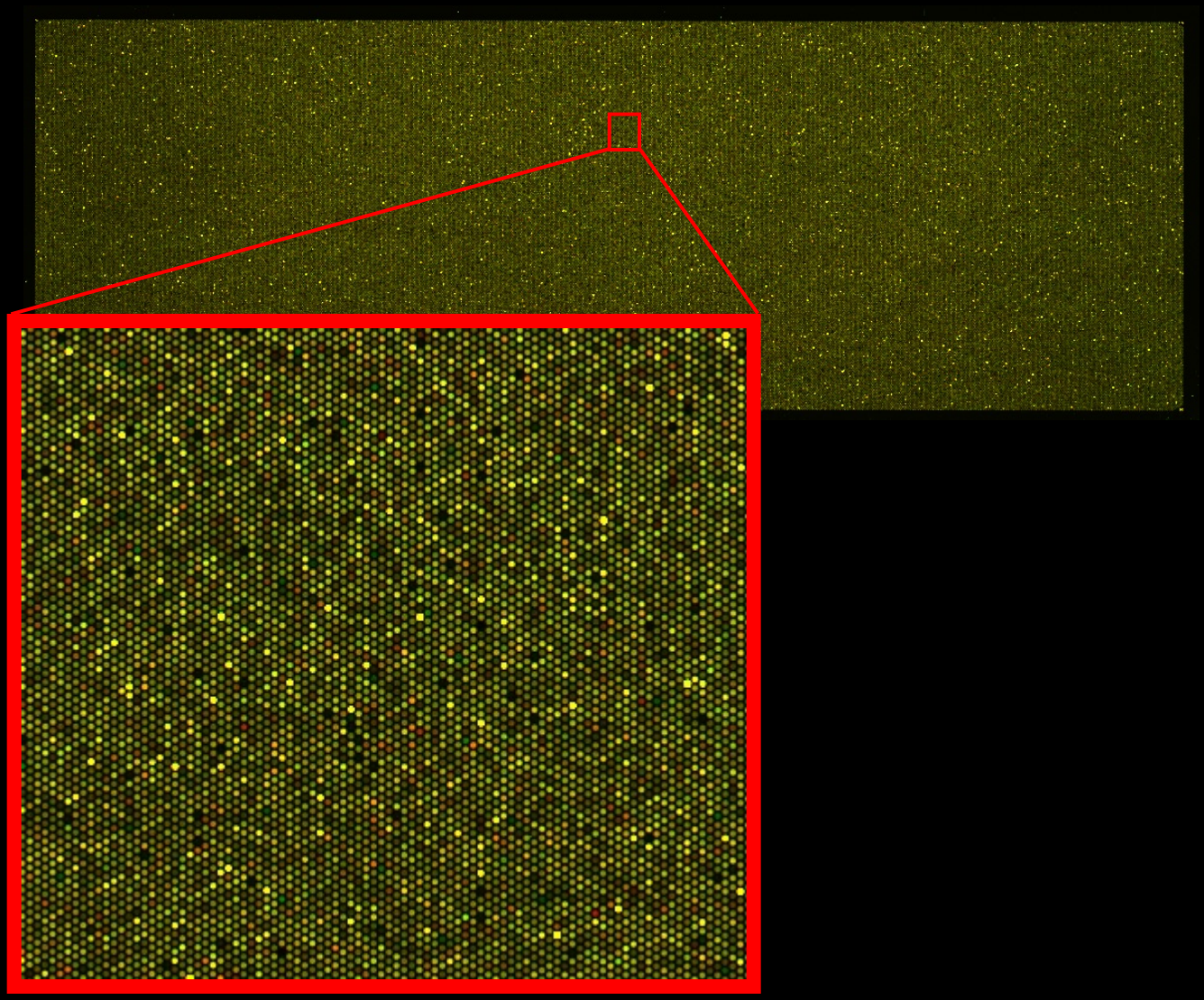
Laser scanner



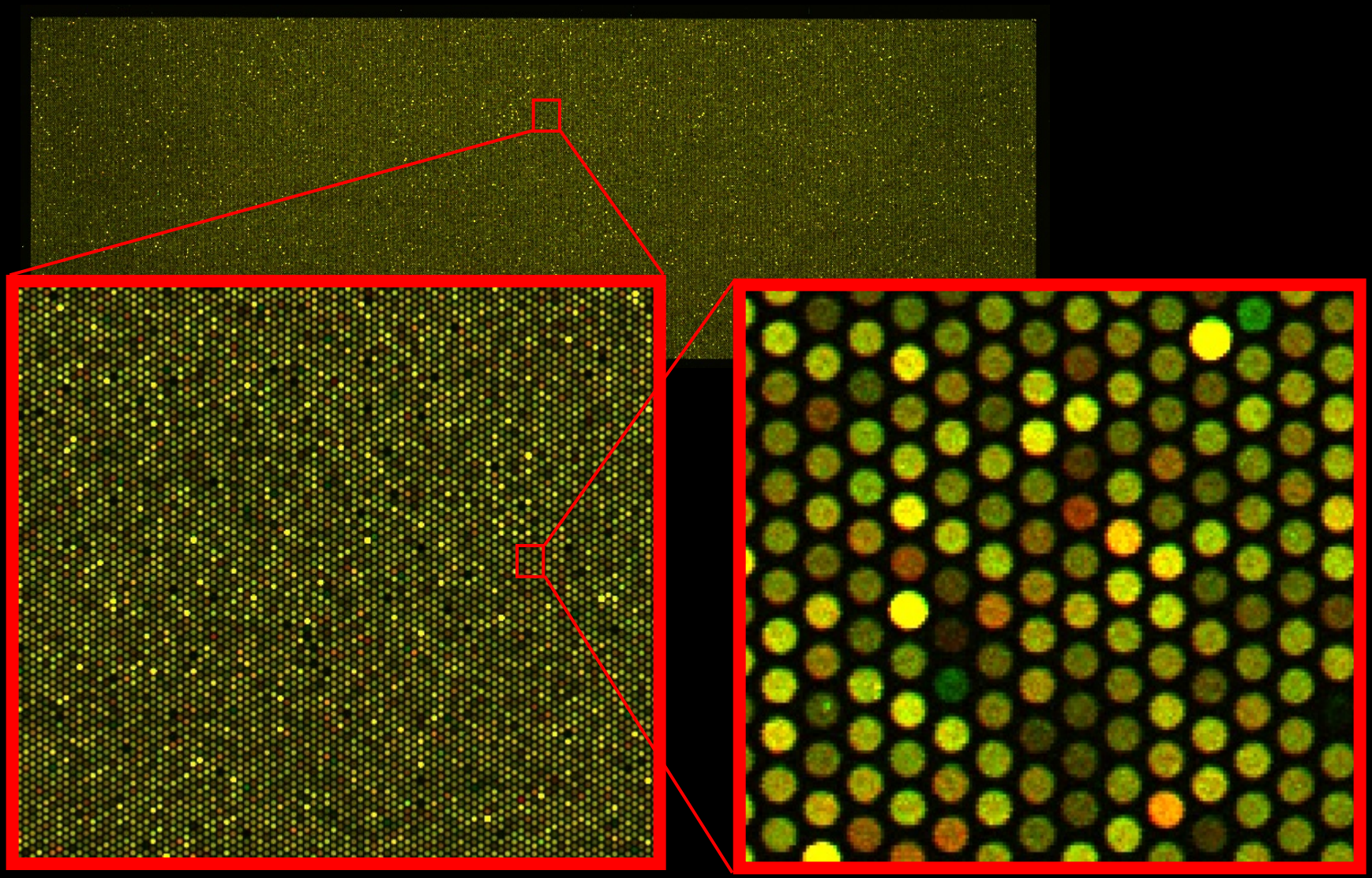
Chromosomal Microarray



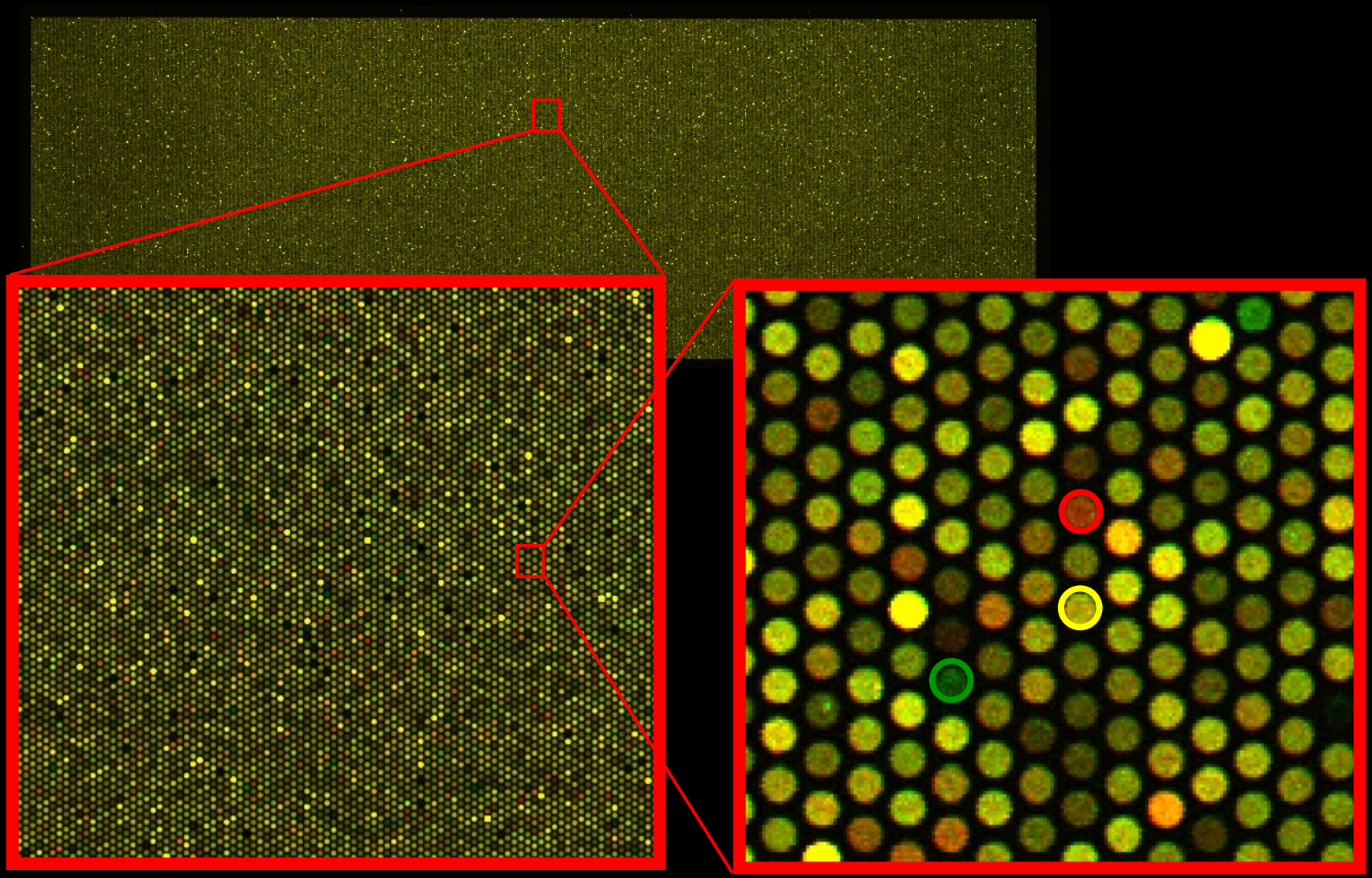
Chromosomal Microarray



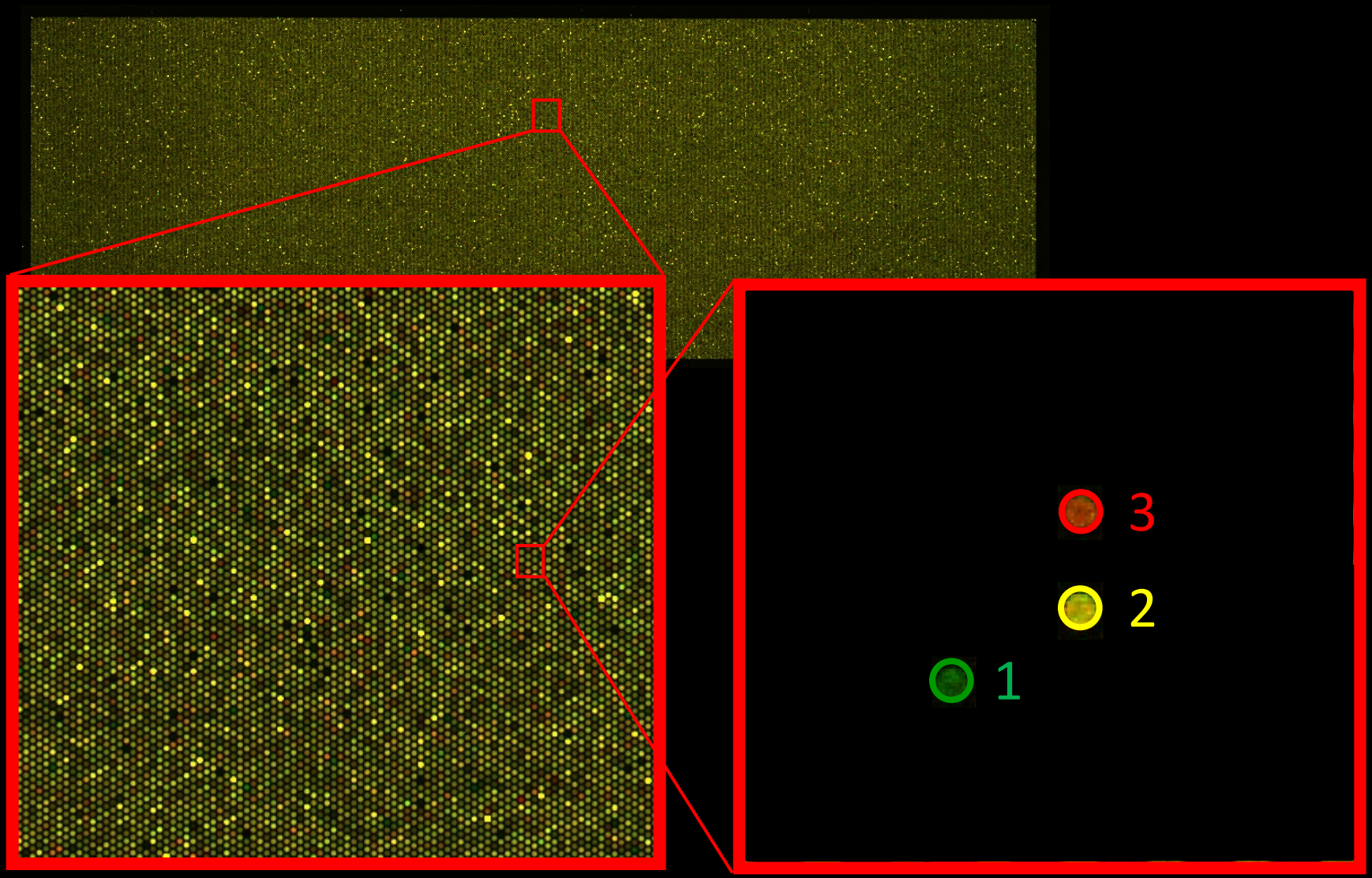
Chromosomal Microarray



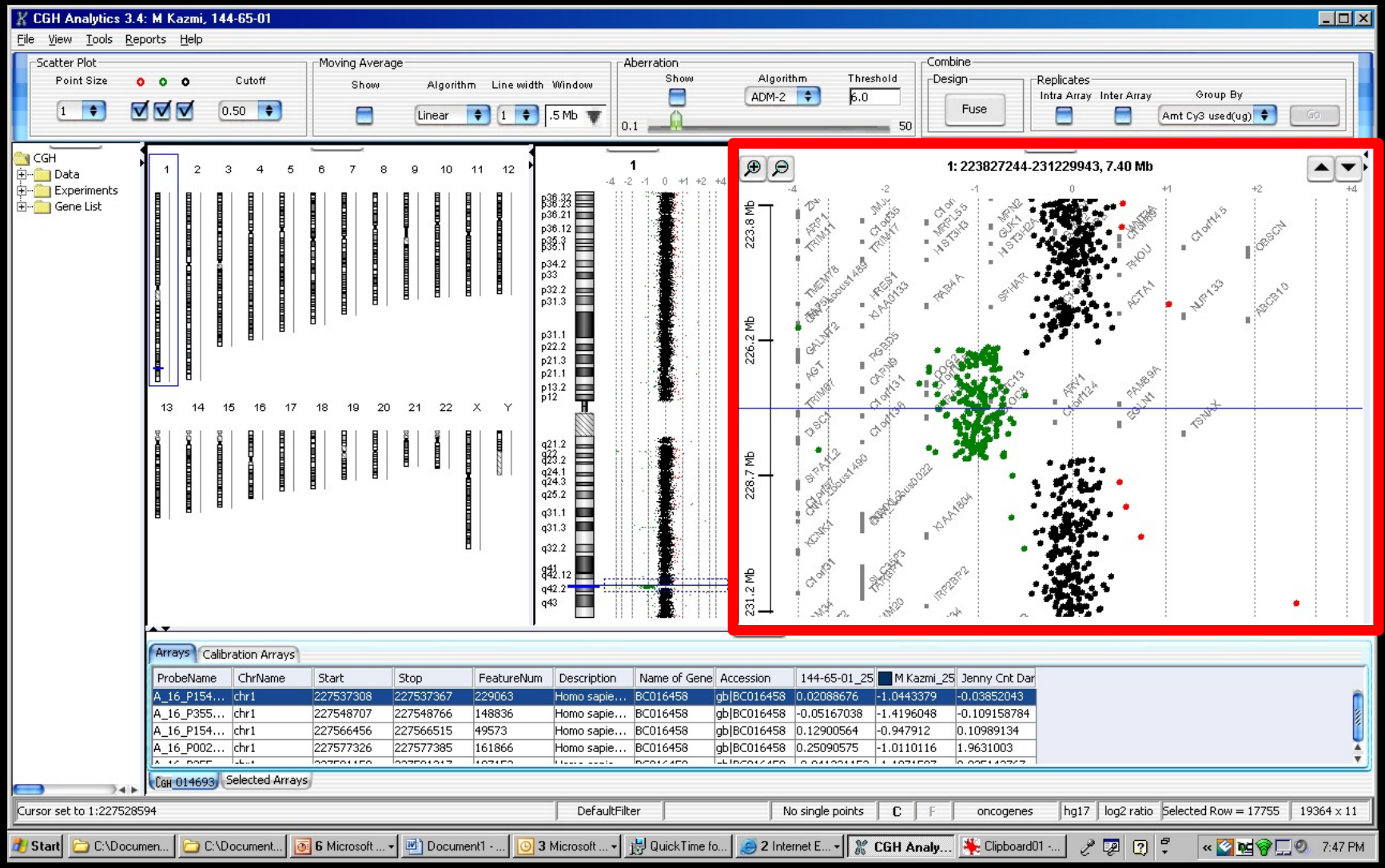
Chromosomal Microarray



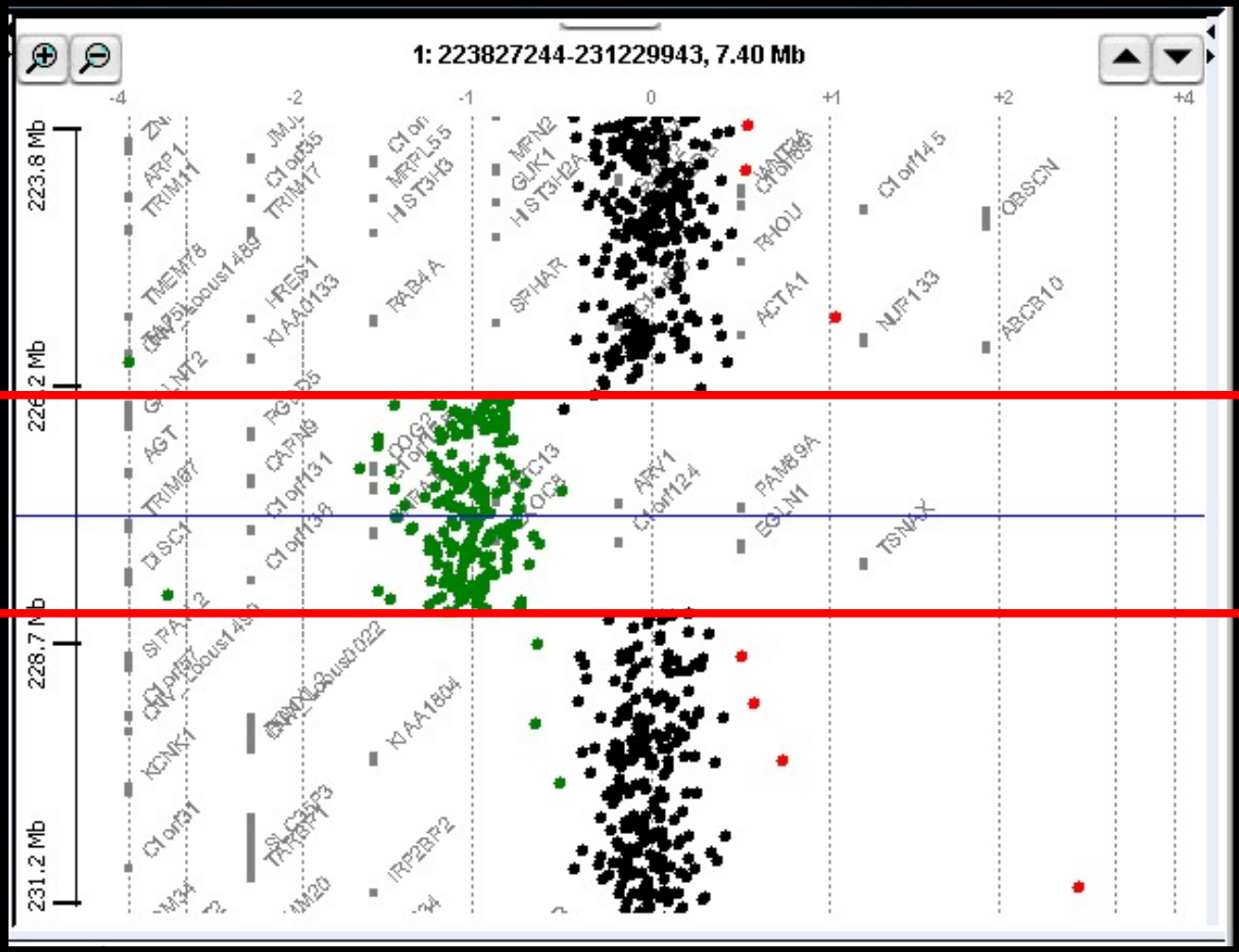
Chromosomal Microarray



Chromosomal Microarray



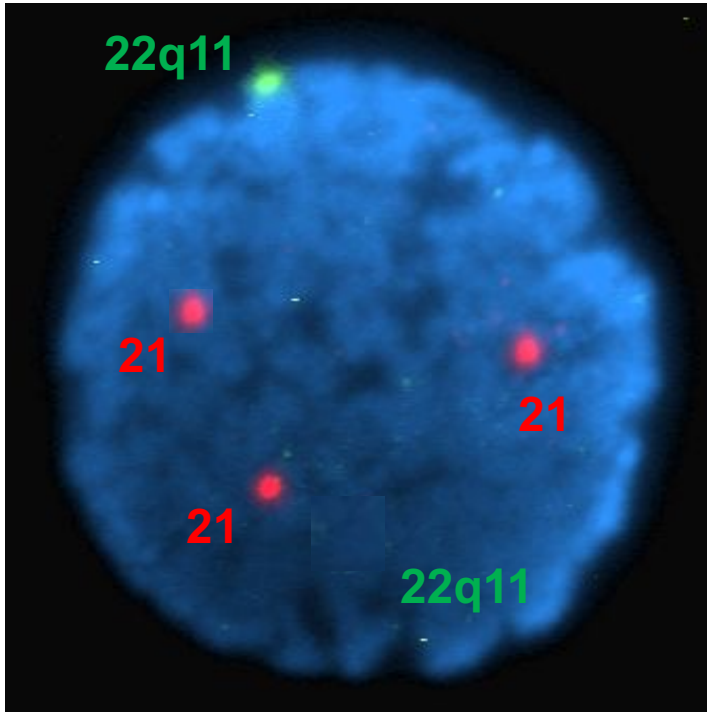
Chromosomal Microarray



Can’t see chromosomal rearrangements

Fluorescence In Situ Hybridization (FISH)?

Down syndrome



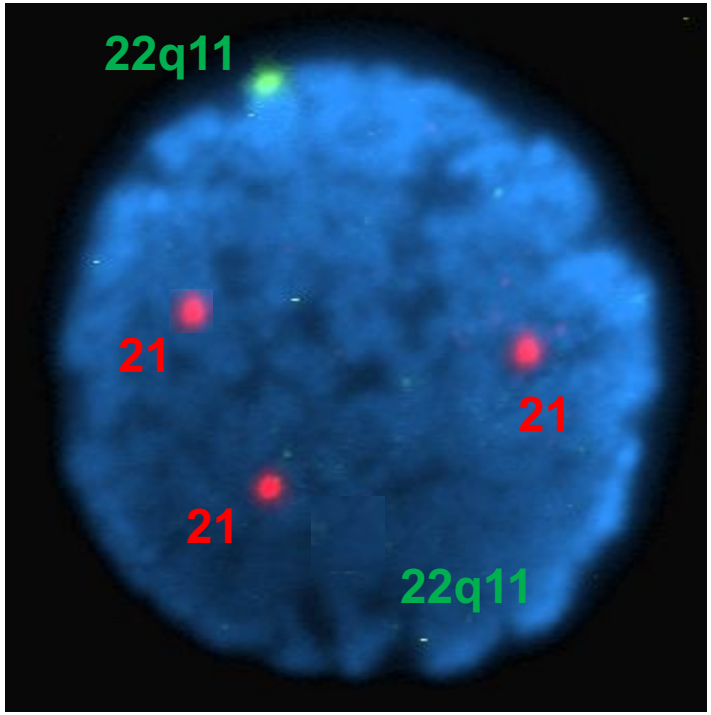
DiGeorge syndrome

Can't tell size

Can't tell orientation

Fluorescence In Situ Hybridization (FISH)?

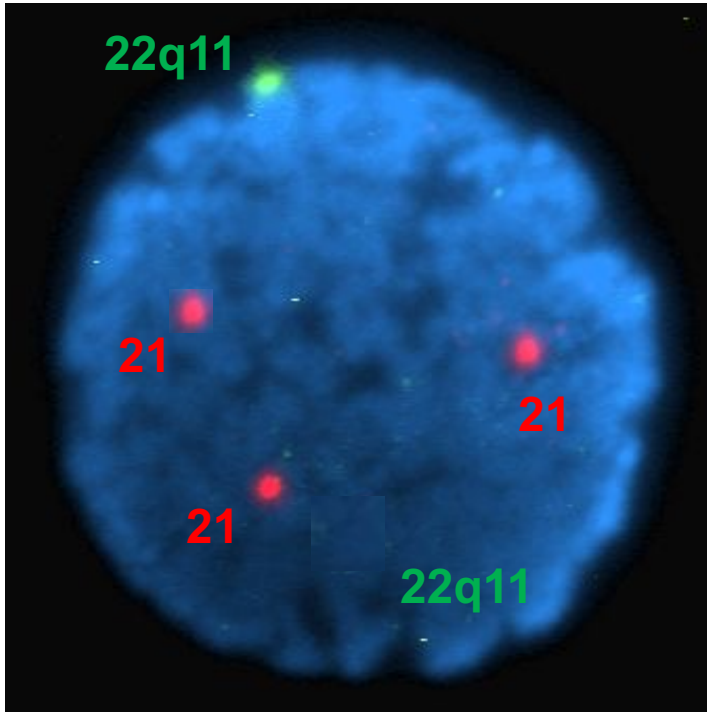
Down syndrome
Chromosome
Analysis



Speed
Preliminary Answer

DiGeorge syndrome
Chromosomal
Microarray

Fluorescence In Situ Hybridization (FISH)?



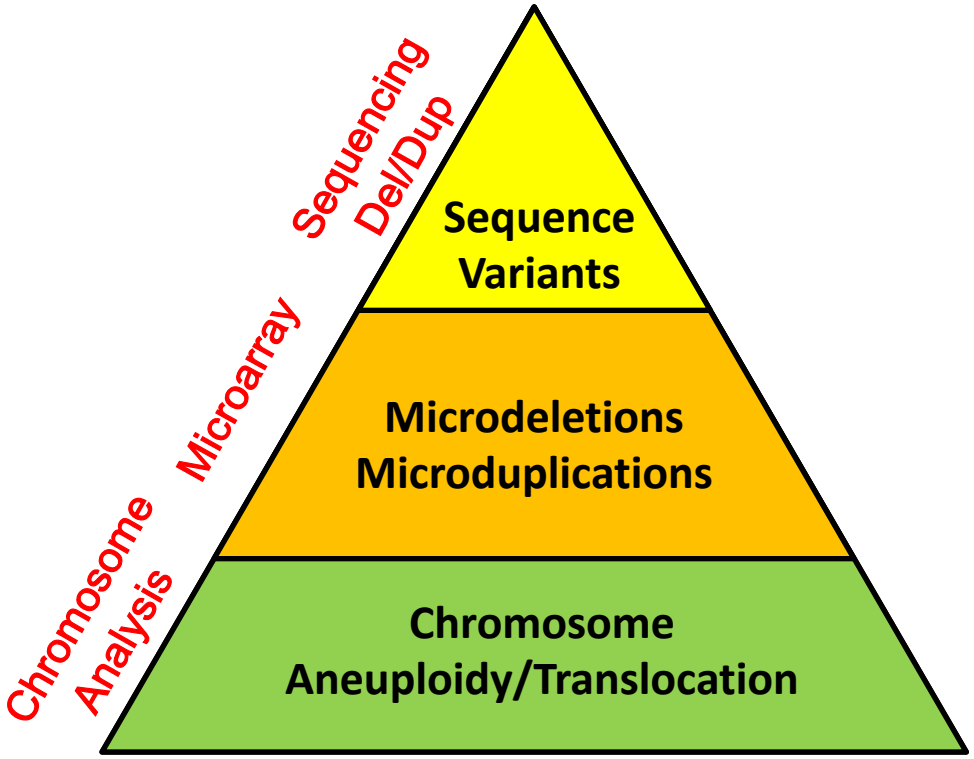
Down syndrome
Chromosome
Analysis

DiGeorge syndrome
Chromosomal
Microarray

Confirm

Check Family Members

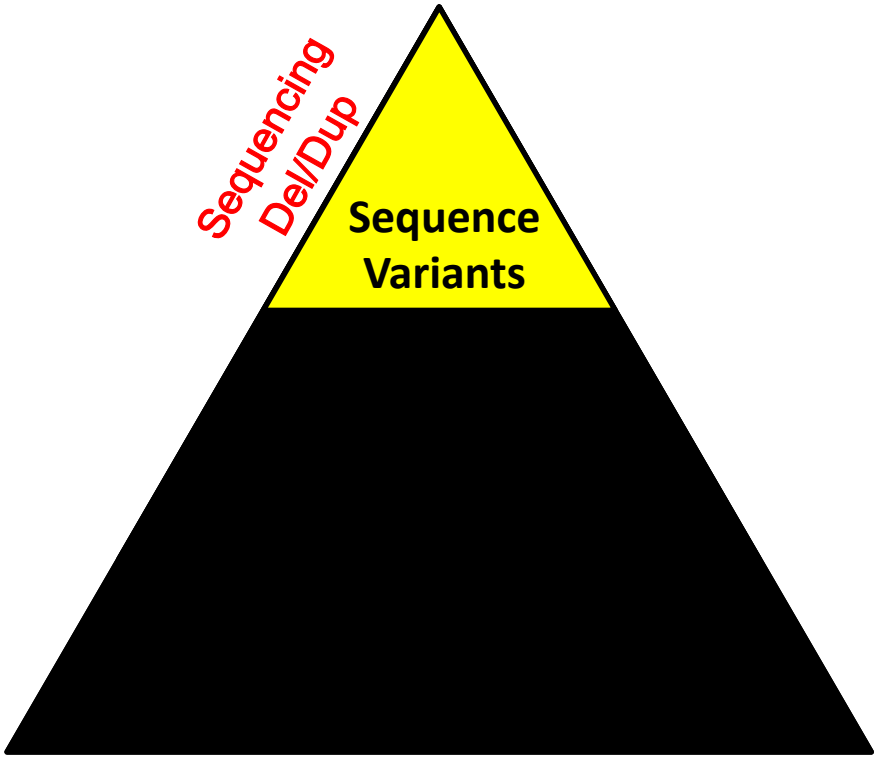
I Know the Diagnosis



Baby Girl: Meconium ileus
Brother: Chronic lung disease

Cystic Fibrosis

I Know the Diagnosis



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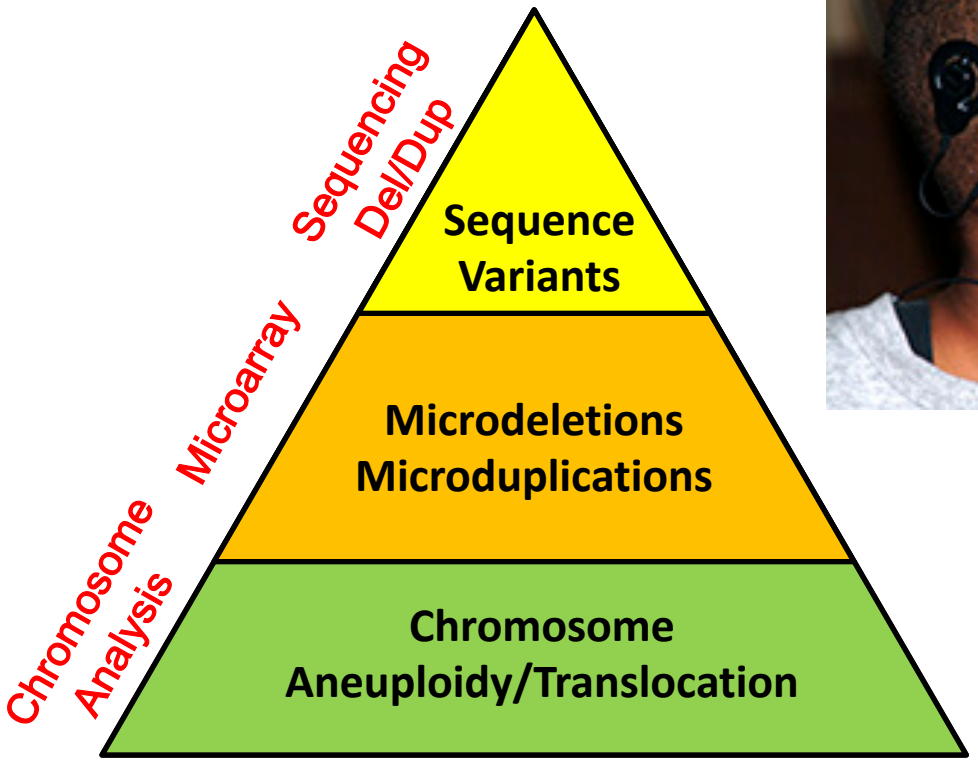
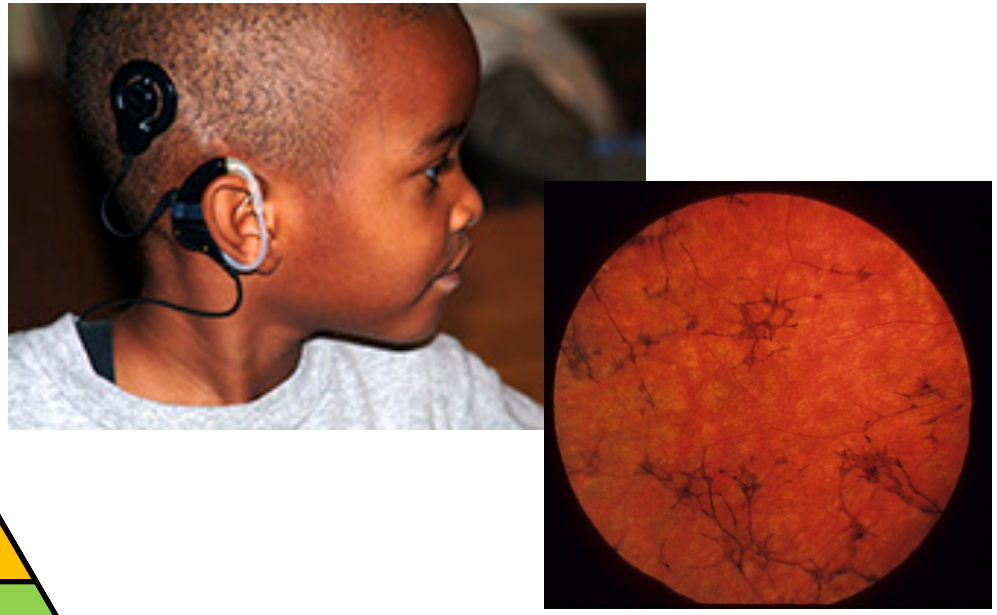
Brother: Chronic lung disease

Cystic Fibrosis

I Know the Diagnosis

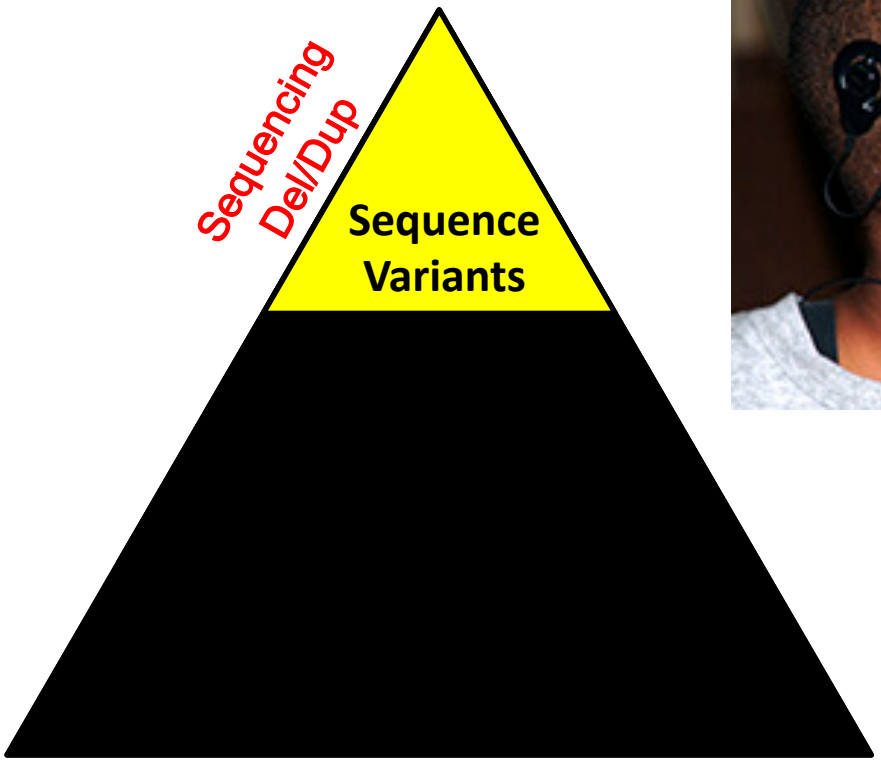


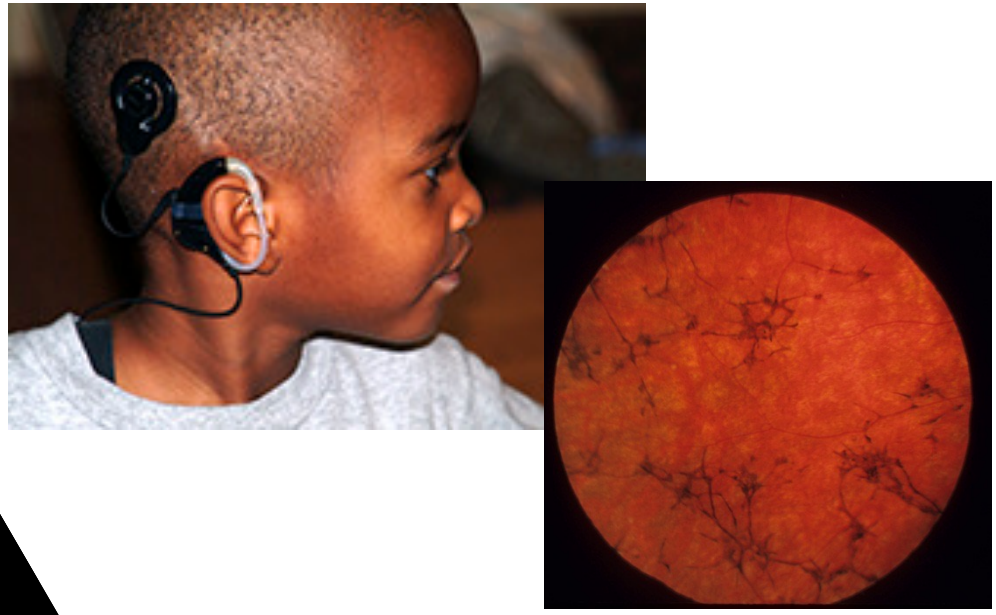


Boy: Profound hearing loss
Retinitis pigmentosa

Usher syndrome

I Know the Diagnosis



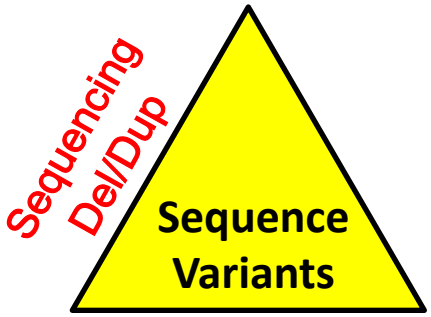


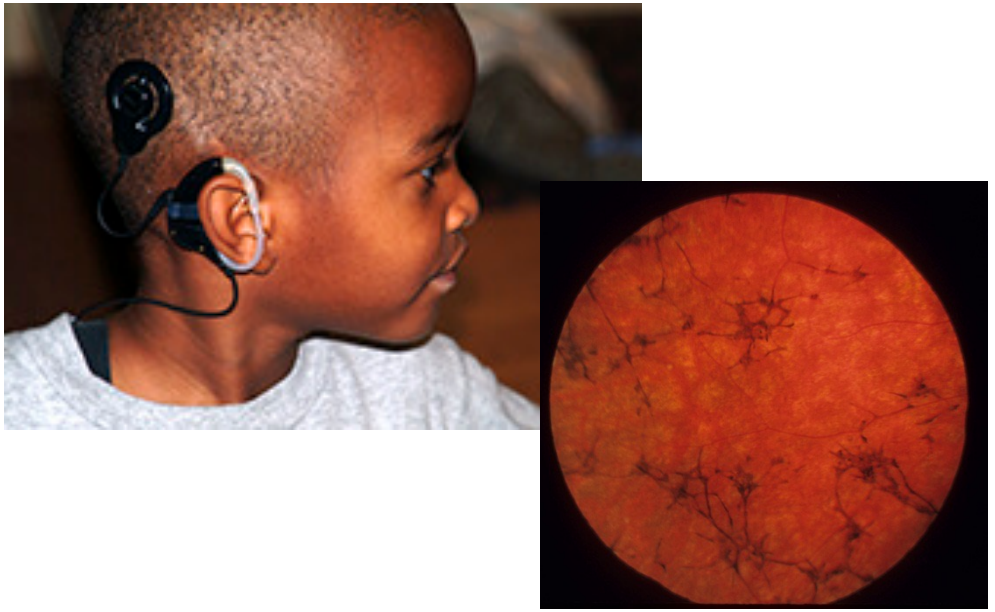
Boy: Profound hearing loss
Retinitis pigmentosa

Usher syndrome

***CDH23*, *CLRN1*, *GPR98*, *MYO7A*,**
PCDH15*, *USH1C*, *USH1G*, and *USH2A

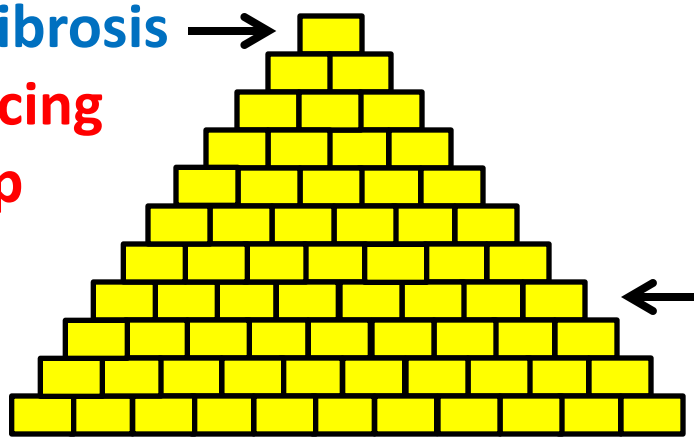
I Know the Diagnosis





Boy: Profound hearing loss
Retinitis pigmentosa

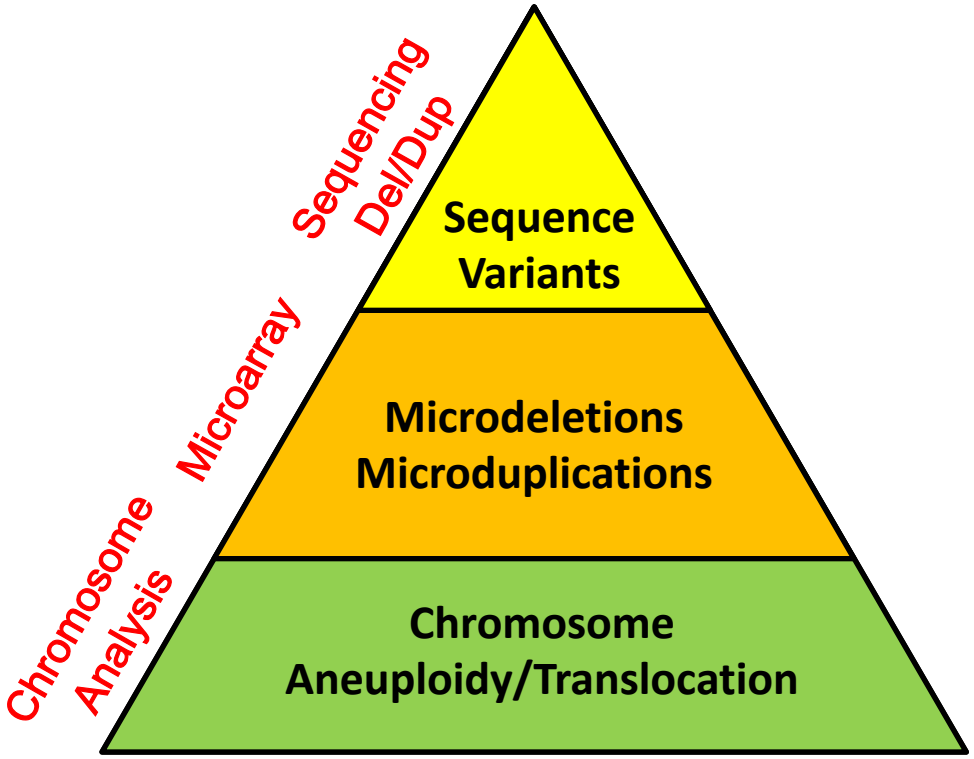
Cystic Fibrosis
Sequencing
Del/Dup



Usher syndrome Panel

CDH23, *CLRN1*, *GPR98*, *MYO7A*, *PCDH15*, *USH1C*, *USH1G*, and *USH2A*

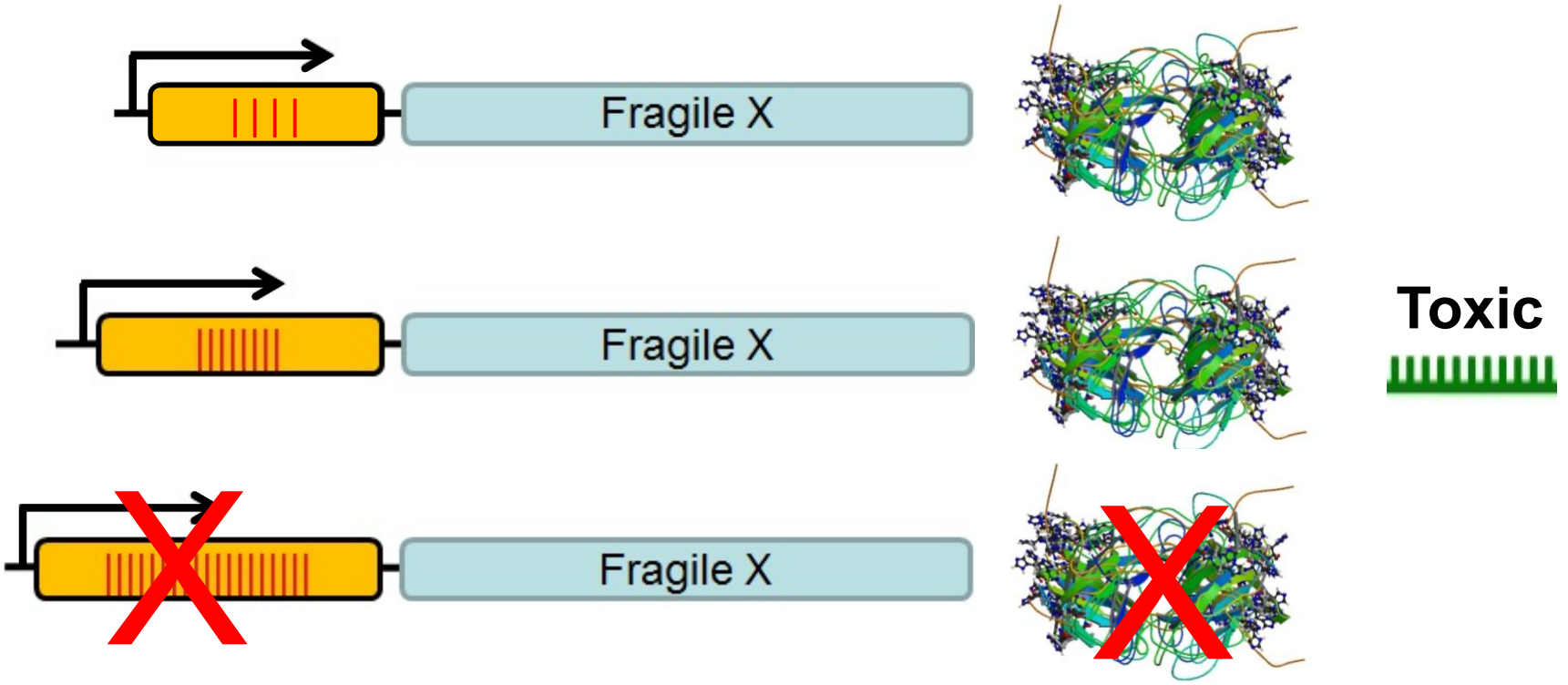
I Know the Diagnosis



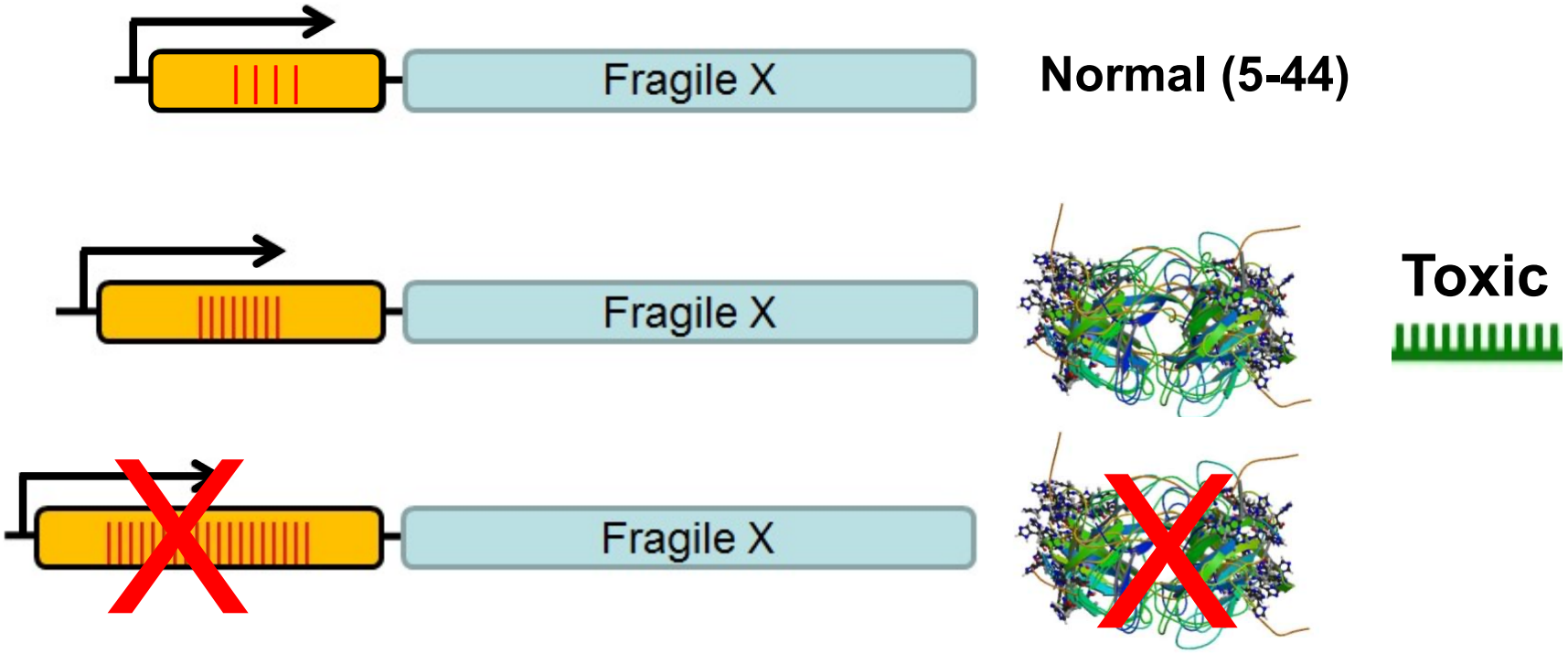
Boy: Cognitive delay
Mother: Ovarian failure
Grandfather: Ataxia/Tremors

Fragile X syndrome

Fragile X



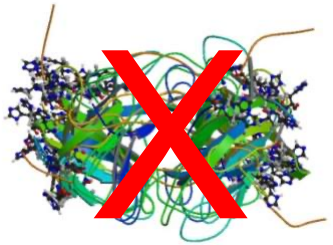
Fragile X



Fragile X

Normal (5-44)

Premutation (55-200)
Ovarian insufficiency
Tremors and ataxia



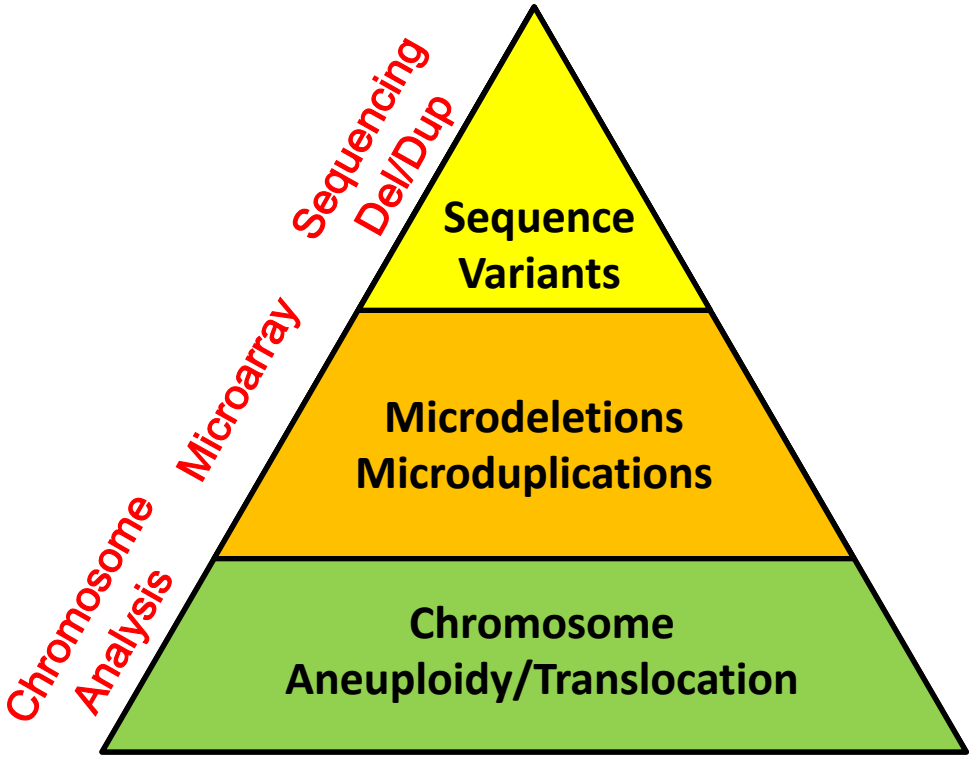
Fragile X

Normal (5-44)

Premutation (55-200)
Ovarian insufficiency
Tremors and ataxia

Full mutation (>200)
Fragile X

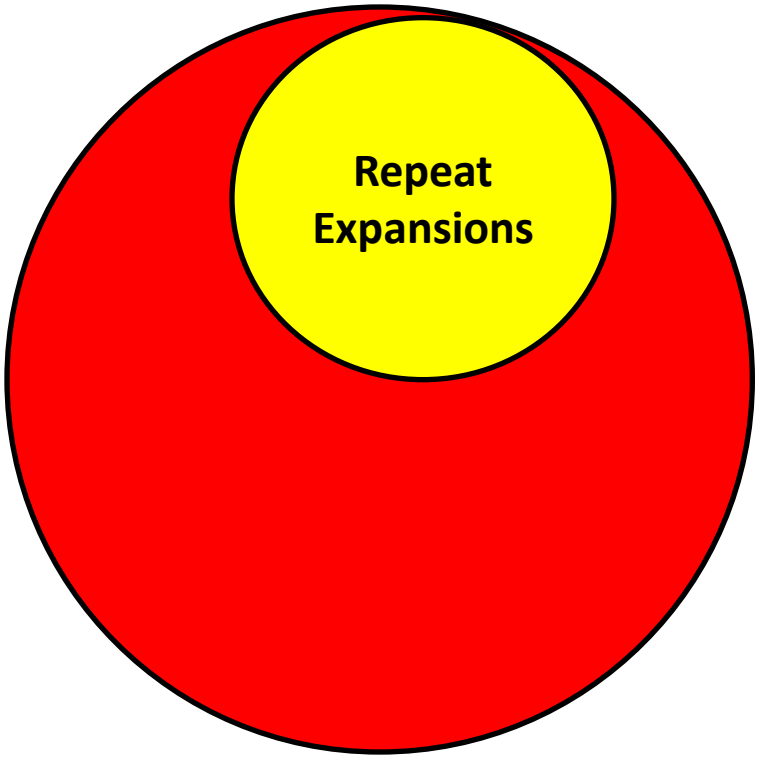
I Know the Diagnosis



Boy: Cognitive delay
Mother: Ovarian failure
Grandfather: Ataxia/Tremors

Fragile X syndrome

I Know the Diagnosis



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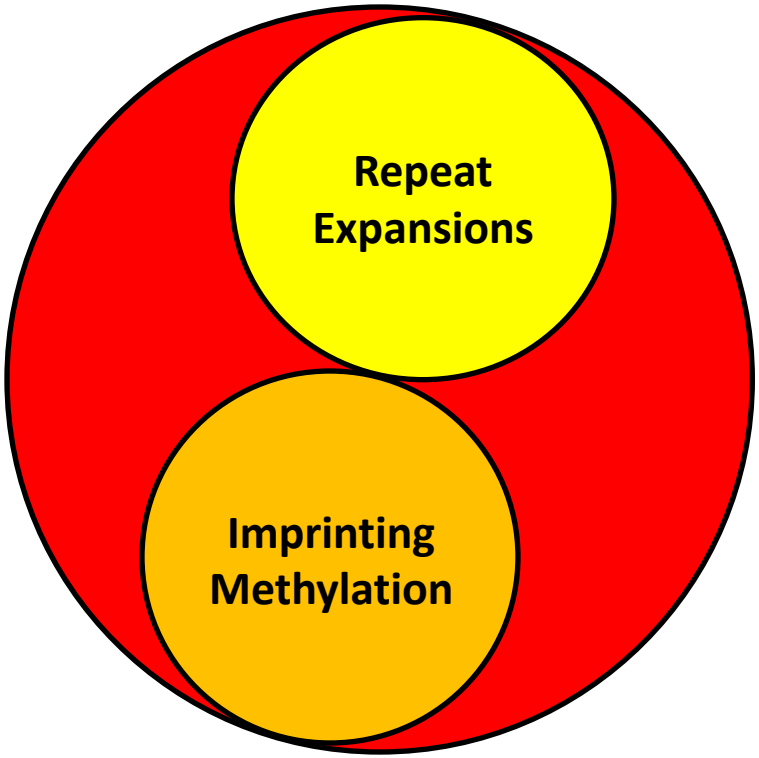
Fragile X syndrome

I Know the Diagnosis



Girl: Cognitive delay, seizures
happy face, poor speech
poor walking, loves water

Angelman syndrome

One Important Question

Evaluation

- Medical history
- Family history
- Physical exam

Do you know the diagnosis?

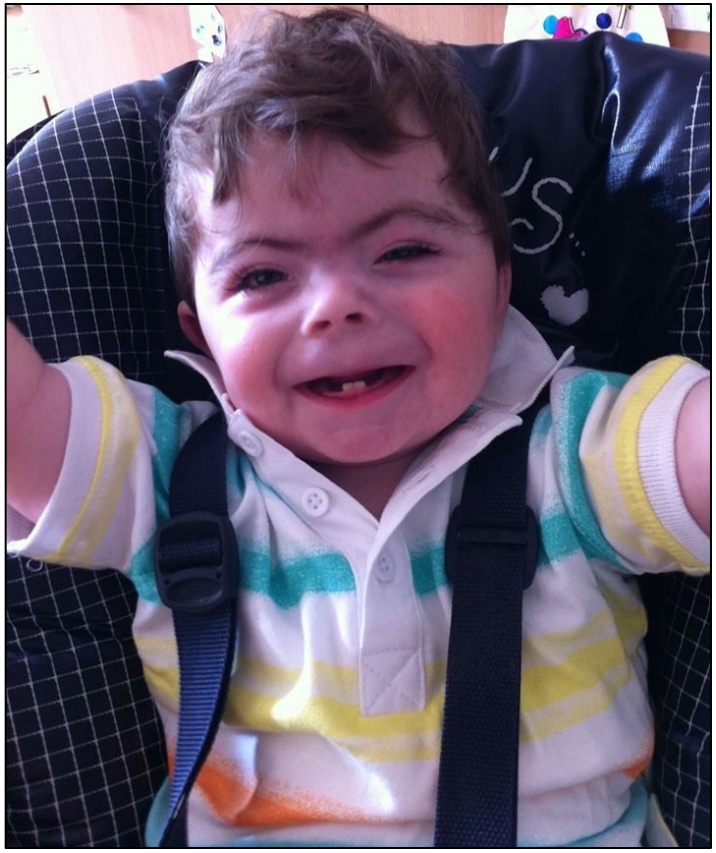
Yes—Triangle Circle

No—3Gs

G #1 = is for Google

Patient

- Short stature
- Arched eyebrows
- Synophrys
- Limb anomalies



G #1 = is for Google

Short stature arched eyebrows synophrys limb anomalies

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About 41,700 results (0.70 seconds)

Craniofacial features include **synophrys**, highly **arched eyebrows**, long eyelashes, **short** nose with anteverted nares, small widely spaced teeth, and microcephaly. IQ ranges from below 30 to 102 (mean: 53). Jan 28, 2016

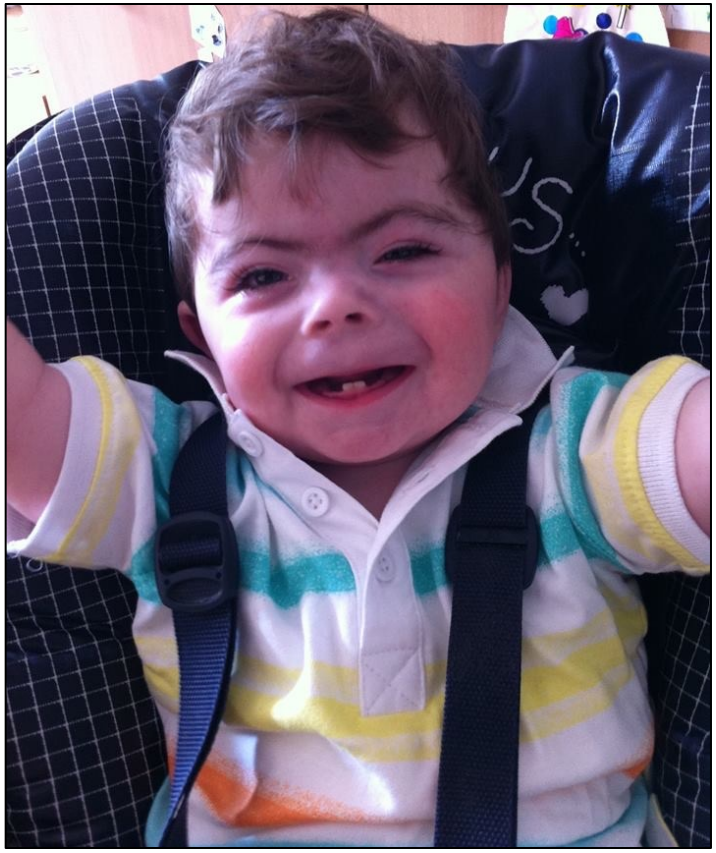
Cornelia de Lange Syndrome - GeneReviews® - NCBI Bookshelf
https://www.ncbi.nlm.nih.gov/books/NBK1104/

About this result Feedback

Cornelia de Lange Syndrome - NORD (National Organization for Rare ...
https://rarediseases.org › For Patients and Families › Rare Disease Information
... nares); neat, **arched eyebrows** that grow together (**synophrys**); long, curly eyelashes; ... In individuals with CdLS, upper **limb abnormalities** may involve one side ... may remain low in weight and exhibit abnormally **short stature** (prenatal and .... abnormally thick **eyebrows**; and/or excessive hair growth (hirsutism) on other ...

Cornelia de Lange Syndrome - GeneReviews® - NCBI Bookshelf
https://www.ncbi.nlm.nih.gov/books/NBK1104/
by MA Deardorff - 2016 - Cited by 59 - Related articles
Jan 28, 2016 - Craniofacial features include **synophrys**, highly **arched eyebrows**, long eyelashes, **short** nose with anteverted nares, small widely spaced teeth, and microcephaly. IQ ranges from below 30 to 102 (mean: 53).

De Lange syndrome (Concept Id: C0270972) - NCBI - NIH
https://www.ncbi.nlm.nih.gov/medgen/78752



Cornelia de Lange syndrome

G #2 = is for Geneticist

Patient

- Intellectual disability
- Seizures
- Mother has large front teeth

KBG syndrome

G #3 = is for Genome-Wide

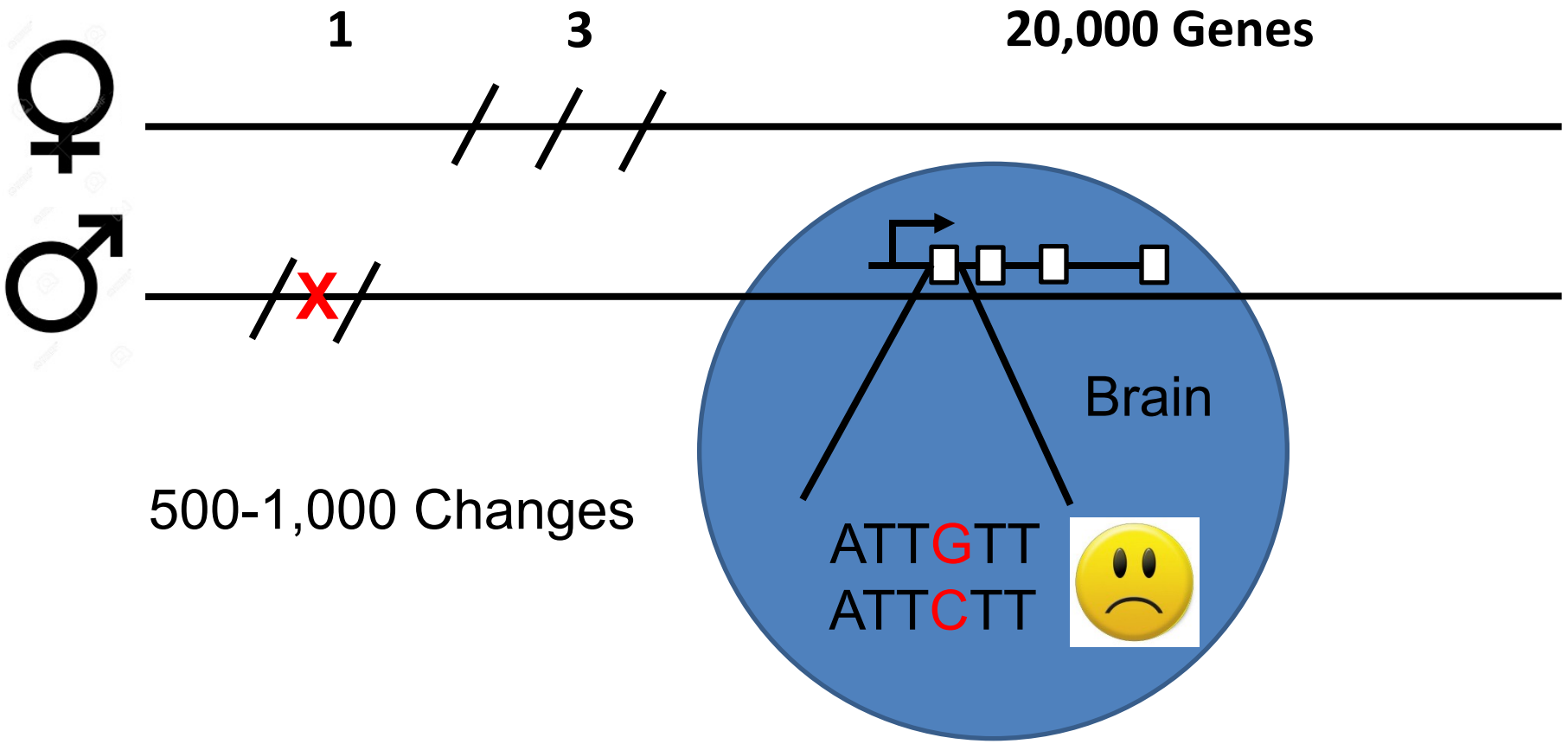
Patients

- Developmental delay
- Intellectual disability
- Autism
- 1000s of genes

Genetics Clinic

Chromosome microarray analysis

Whole Exome Sequencing



G #3 = is for Genome-Wide

Patients

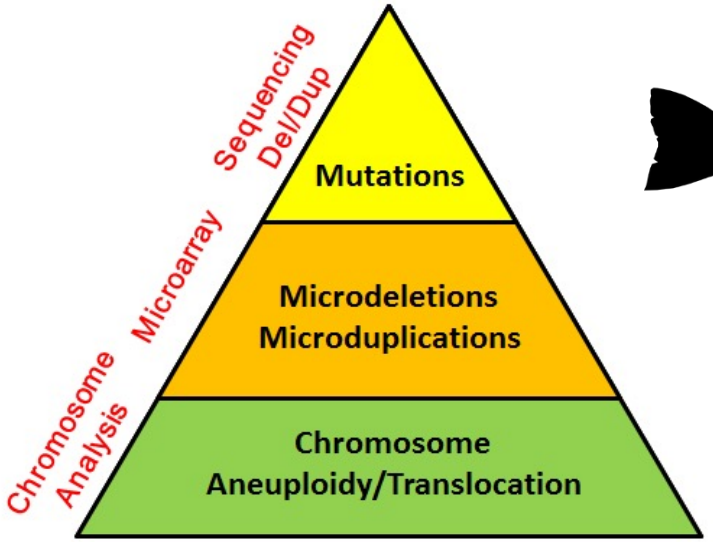
- Developmental delay
- Intellectual disability
- Autism
- 1000s of genes

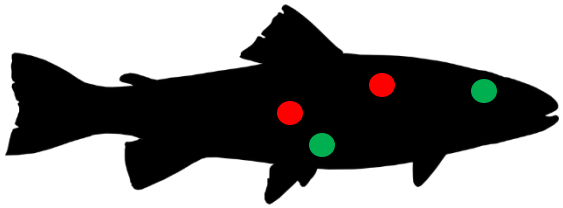
Diagnosis

- 10-15% CMA
- 25-35% WES

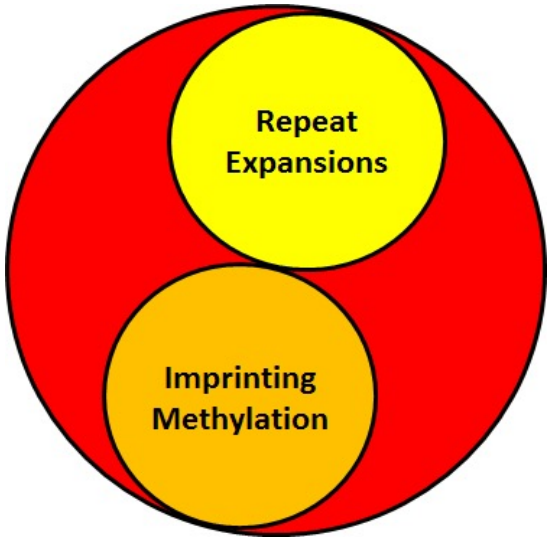
Take Home Messages

- **Do I know the diagnosis?**



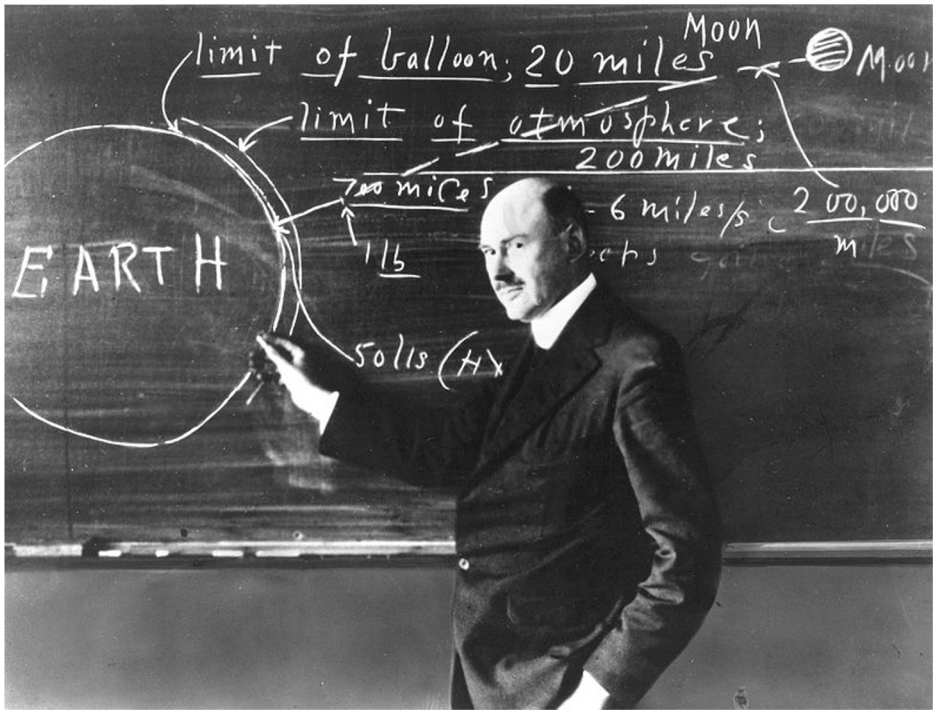


Emergency
Confirmation
Chromosome
Microdeletion
Microduplication



- **Google**
- **Geneticist**
- **Genome-Wide testing (CMA and WES)**

Ordering Genetic Testing: It's Not Rocket Science



Haley Streff, MS, CGC
Baylor College of Medicine
Texas Children's Hospital

One Important Question

- Do I know how to send the genetic test(s)?
  - Yes → Send it!
  - No →…

Follow the Steps

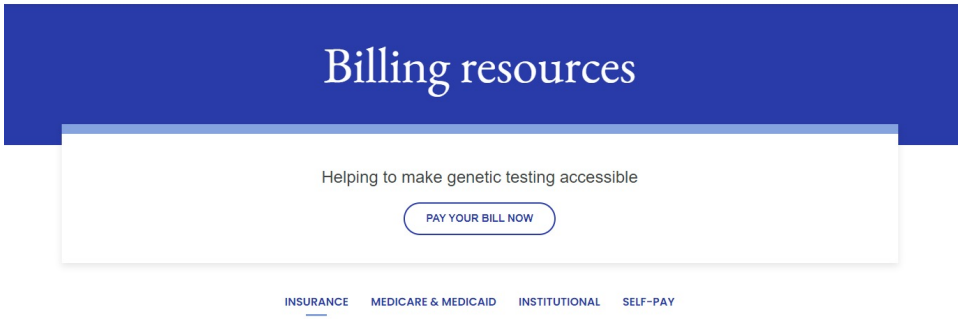
1. Lab selection
2. Billing check
3. Prior Authorization PRN
4. Order test

1. Lab Selection

- National genetic testing labs
- Check for sponsored tests: https://www.heartlandcollaborative.org/educational-resources/genetic-testing-toolkit/
- Online lookup:
  - https://www.concertgenetics.com/
  - https://www.mountainstatesgenetics.org/wp-content/uploads/sites/257/2019/02/Lab-information-Final.pdf

2. Billing Check

- Does the lab accept your state's Medicaid plan?
- Are they in-network with most private insurance companies?
- Direct v. Institutional bill

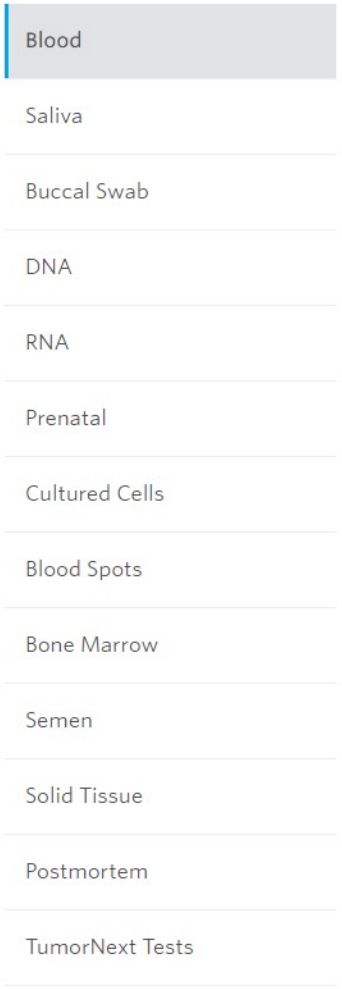


3. Prior Authorization

- By your clinic
- By the genetic testing lab
  - https://www.mountainstatesgenetics.org/wp-content/uploads/sites/257/2019/02/Lab-information-Final.pdf
- https://www.heartlandcollaborative.org/educational-resources/genetic-testing-toolkit/

4. Order the Test

- Check sample requirements
  - Buccal/saliva/blood
- Consent
  - https://www.mountainstatesgenetics.org/wp-content/uploads/sites/257/2018/10/Elements-of-Consent-for-Genetic-Testing-Final-Oct-2018.pdf
- Send the test!



Example – Texas Children's Hospital

Staff denotes Texas MCD patients in appt notes with ***

GCs have identified 2 labs that accept Texas MCD

Order same-day buccal tests for patients with *** to the 2 labs

All patients without *** are notified PA will occur on their behalf

One staff member and one GCA primarily handle all PAs and testing coordination

Mythbusters

- Insurance coverage for many genetic tests has improved.

- Most major genetic testing labs have teams to help you!
  - Test selection
  - Follow-up genetic counseling
  - Billing and PA
  - Regional reps

CHAT CHECK

Question

1) Type answer into the chat
2) Do not push enter
3) When given the signal push enter
4) We will reveal the correct answer

CHAT CHECK

What test should you order?

Time Limit: 15 sec

A) Single gene
B) Gene panel
C) Chromosomes
D) FISH
E) CMA
F) CMA then WES

Don't Push Enter Until I Ask You To!

CHAT CHECK

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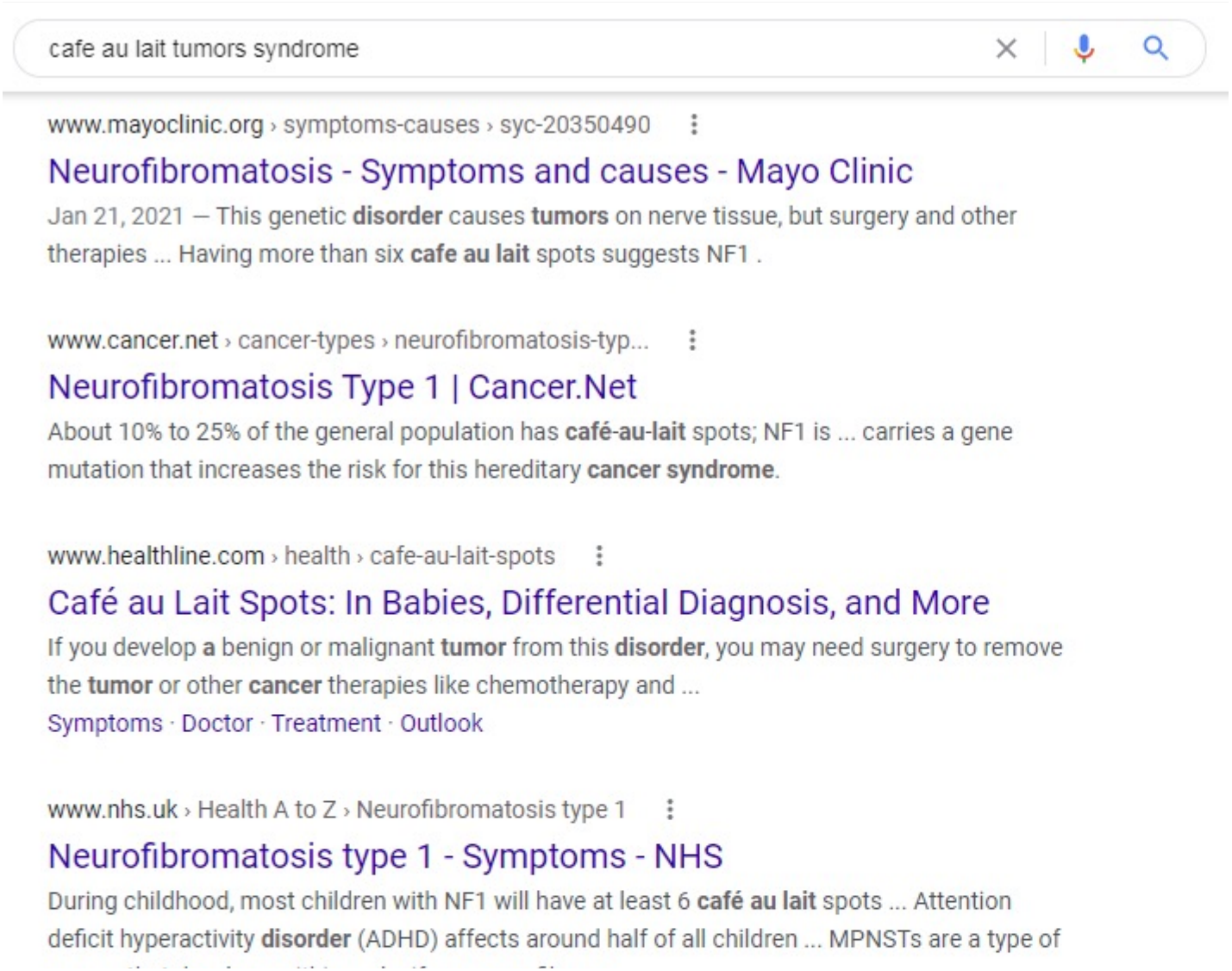
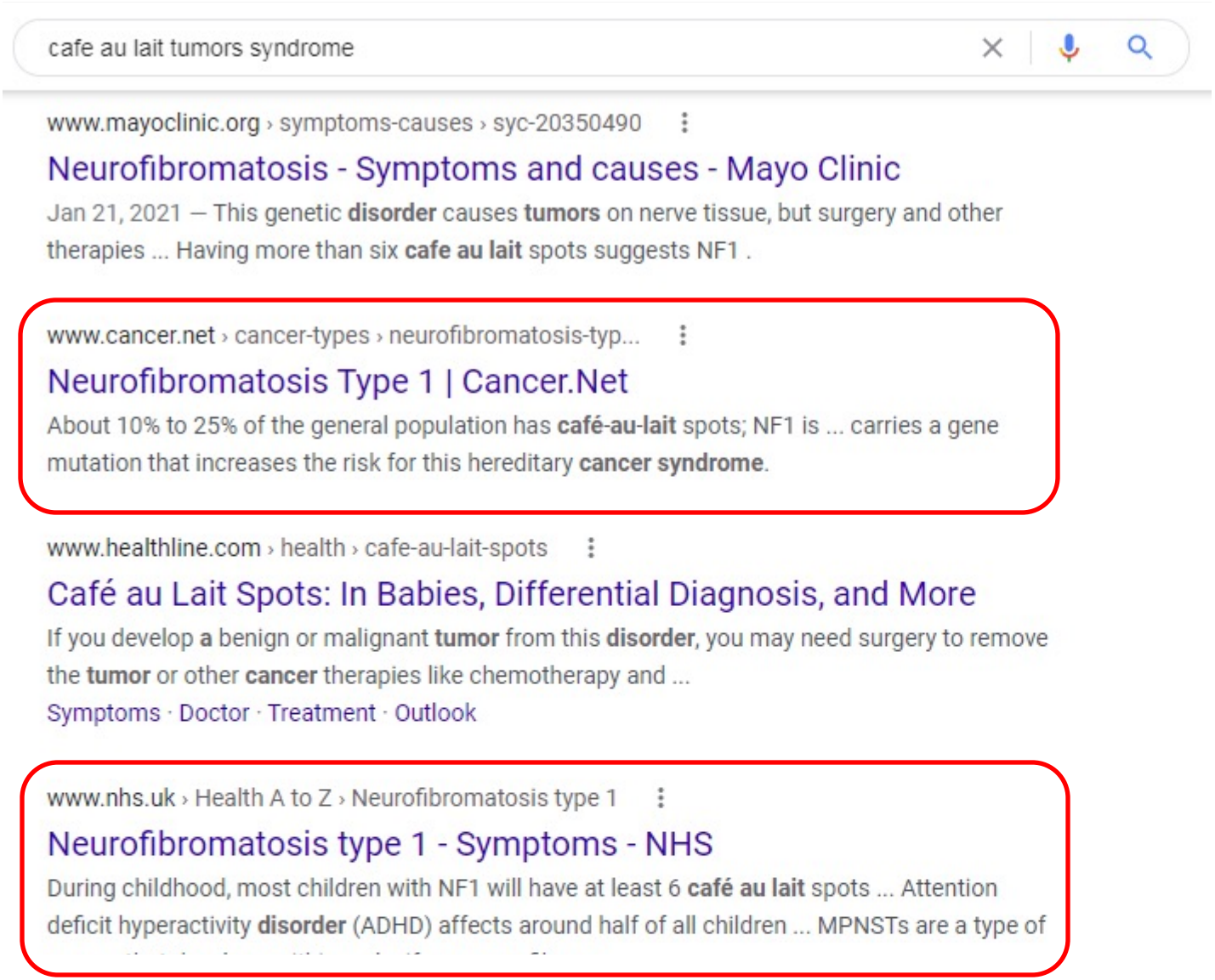
Actas Dermosifiliogr. 2016;107:454–64

CHAT CHECK


cafe au lait tumors syndrome
www.mayoclinic.org › symptoms-causes › syc-20350490
Neurofibromatosis - Symptoms and causes - Mayo Clinic
Jan 21, 2021 — This genetic disorder causes tumors on nerve tissue, but surgery and other therapies ... Having more than six cafe au lait spots suggests NF1 .
www.cancer.net › cancer-types › neurofibromatosis-typ...
Neurofibromatosis Type 1 | Cancer.Net
About 10% to 25% of the general population has café-au-lait spots; NF1 is ... carries a gene mutation that increases the risk for this hereditary cancer syndrome.
www.healthline.com › health › cafe-au-lait-spots
Café au Lait Spots: In Babies, Differential Diagnosis, and More
If you develop a benign or malignant tumor from this disorder, you may need surgery to remove the tumor or other cancer therapies like chemotherapy and ...
Symptoms · Doctor · Treatment · Outlook
www.nhs.uk › Health A to Z › Neurofibromatosis type 1
Neurofibromatosis type 1 - Symptoms - NHS
During childhood, most children with NF1 will have at least 6 café au lait spots ... Attention deficit hyperactivity disorder (ADHD) affects around half of all children ... MPNSTs are a type of

CHAT CHECK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Neurofibromatosis - Symptoms and causes - Mayo Clinic
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Café au Lait Spots: In Babies, Differential Diagnosis, and More
If you develop a benign or malignant tumor from this disorder, you may need surgery to remove the tumor or other cancer therapies like chemotherapy and ...
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Neurofibromatosis Type 1

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Neurofibromatosis Type 1 = *NF1*

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Neurofibromatosis Type 1 = *NF1*

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CHAT CHECK

www.massgeneral.org › tsc › patient-education › how-t...

How TSC Affects the Skin - Massachusetts General Hospital

Hypomelanotic (**ash leaf**) macule on the arm ... These harmless "white **spots**," as they are sometimes called, are seen in more than 90 ... and unlike many other skin lesions associated with the **disorder**, they are common in infants with TSC. ... **Angiofibromas** typically appear in people with TSC between age two and age five, ...

www.ncbi.nlm.nih.gov › pmc › articles › PMC3750905

Do you know this syndrome? - NCBI - NIH

by GMA Gontijo · 2013 · Cited by 1 · Related articles

The most characteristic hypochromic spots are those which have the form of leaves, called "**ash-leaf spots**". Facial **angiofibromas**: apparent between the second ...

www.aafp.org › afp

Recognizing an Index Case of Tuberous Sclerosis - American ...

by JS Hurst · 2000 · Cited by 47 · Related articles

Feb 1, 2000 — Characteristic signs of tuberous sclerosis vary widely in severity and can include hypopigmented "**ash-leaf spots**," fibrous plaques on the forehead, **angiofibromas** on the face (adenoma sebaceum), a shagreen patch on the lower back and fibromas of the nails.

CHAT CHECK

www.massgeneral.org › tsc › patient-education › how-t...

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www.aafp.org › afp

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Tuberous Sclerosis

CHAT CHECK

What test should you order?

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A) Single gene
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Tuberous Sclerosis = *TS1* & *TS2*

CHAT CHECK

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Tuberous Sclerosis = *TS1* & *TS2*

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CHAT CHECK

What test should you order?

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D) FISH
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F) CMA then WES

Trisomy 13

CHAT CHECK

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Trisomy 13

CHAT CHECK

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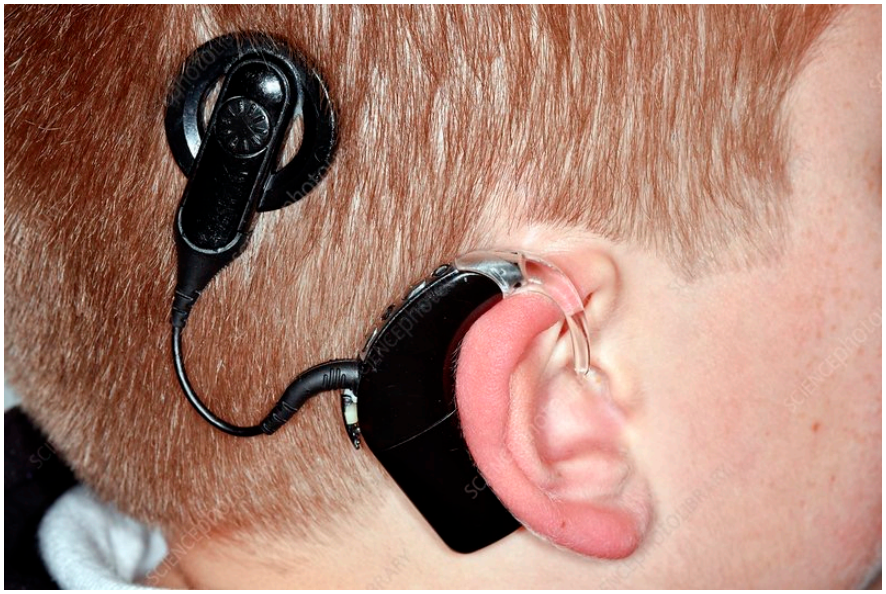

Hearing loss = >100 genes

CHAT CHECK

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Hearing loss = >100 genes

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Hearing loss = >100 genes

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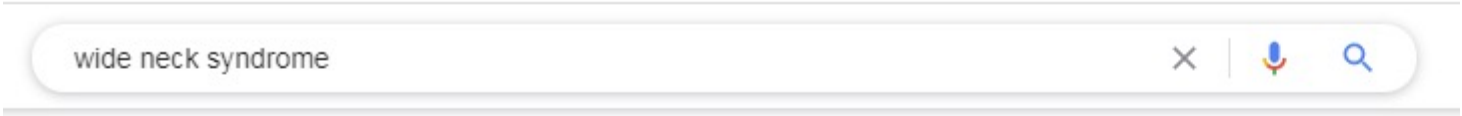


rarediseases.org › Rare Diseases

Noonan Syndrome - NORD (National Organization for Rare ...

The **disorder** is characterized by a **wide** spectrum of symptoms and physical ... of the head and facial (craniofacial) area, a **broad** or webbed **neck**, short stature, ...

General Discussion · Signs & Symptoms · Causes · Related Disorders

www.nhs.uk › Health A to Z › Turner syndrome

Turner syndrome - Symptoms - NHS

a particularly short, **wide neck** (webbed neck) · a broad chest and widely spaced nipples · arms that turn out slightly at the elbows · a low hairline · mouth ...

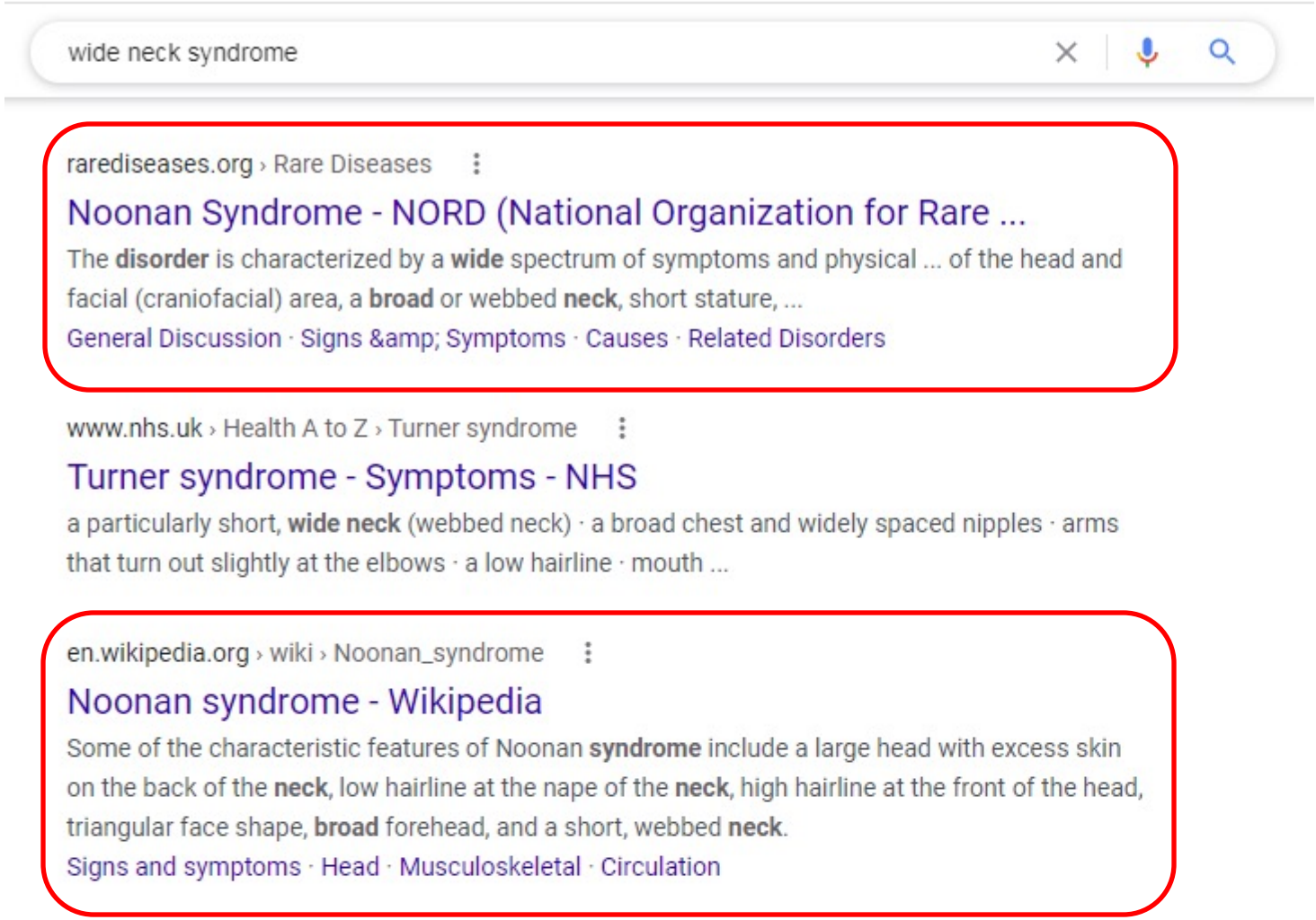
en.wikipedia.org › wiki › Noonan_syndrome

Noonan syndrome - Wikipedia

Some of the characteristic features of Noonan **syndrome** include a large head with excess skin on the back of the **neck**, low hairline at the nape of the **neck**, high hairline at the front of the head, triangular face shape, **broad** forehead, and a short, webbed **neck**.

Signs and symptoms · Head · Musculoskeletal · Circulation

CHAT CHECK


wide neck syndrome
rarediseases.org › Rare Diseases
Noonan Syndrome - NORD (National Organization for Rare ...
The disorder is characterized by a wide spectrum of symptoms and physical ... of the head and facial (craniofacial) area, a broad or webbed neck, short stature, ...
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en.wikipedia.org › wiki › Noonan_syndrome
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Noonan syndrome

CHAT CHECK

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Noonan syndrome = 14 genes

CHAT CHECK

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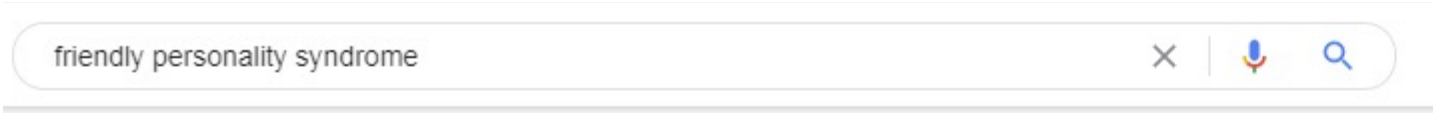


en.wikipedia.org › wiki › Williams_syndrome

Williams syndrome - Wikipedia

Williams **syndrome** (WS) is a genetic **disorder** that affects many parts of the body. Facial ... Some studies suggest that the amygdala of a **person** with Williams **syndrome** has greater volume than the average ... This is possibly due to the fact that although they are very **friendly** to strangers and love meeting new people, they ...

Kabuki syndrome · Gabrielle Marion-Rivard · Supravalvular aortic stenosis

abcnews.go.com › Health › story

Friendly to the Extreme: Meet Kids and Adults With Williams ...

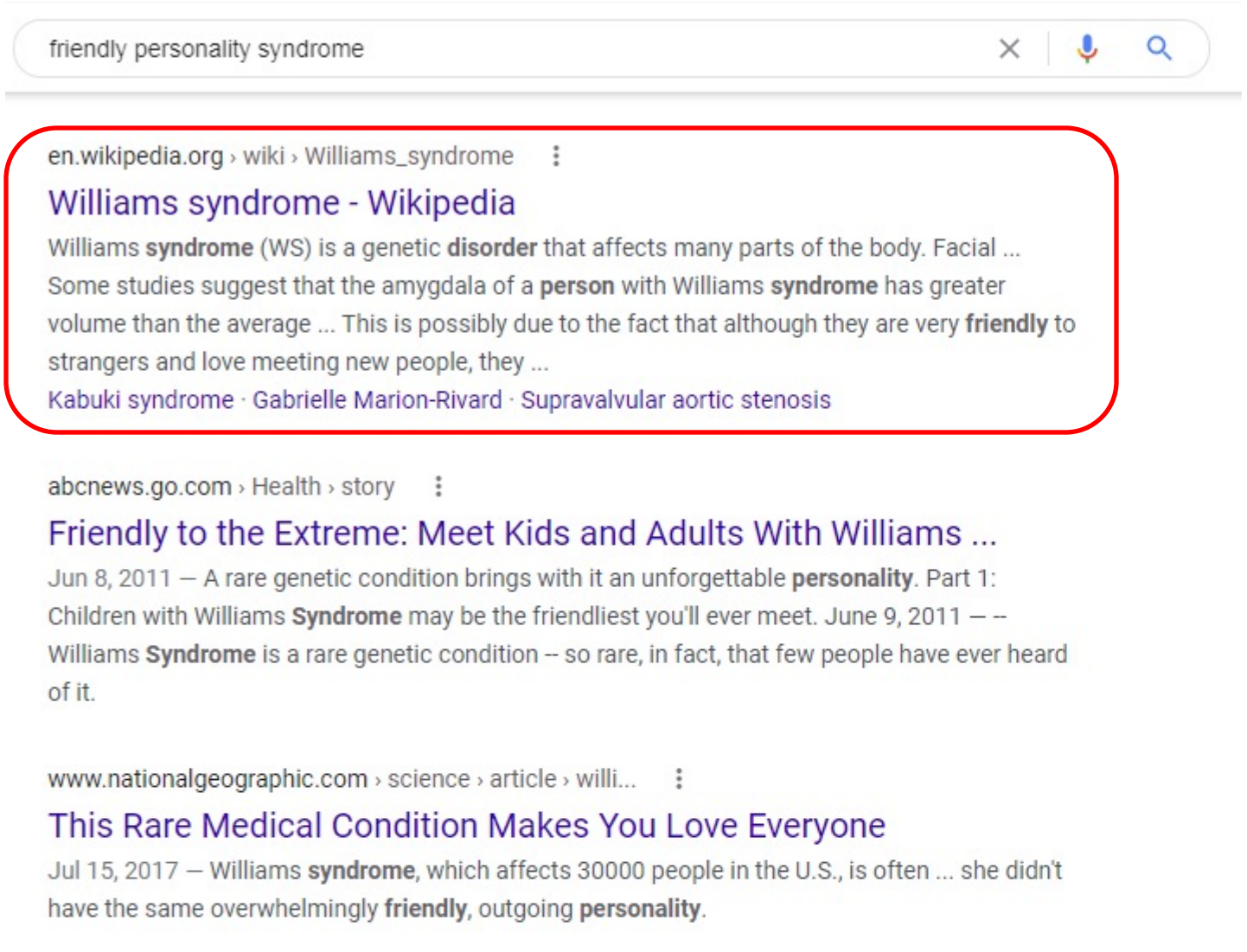
Jun 8, 2011 — A rare genetic condition brings with it an unforgettable **personality**. Part 1: Children with Williams **Syndrome** may be the friendliest you'll ever meet. June 9, 2011 — -- Williams **Syndrome** is a rare genetic condition -- so rare, in fact, that few people have ever heard of it.

www.nationalgeographic.com › science › article › willi...

This Rare Medical Condition Makes You Love Everyone

Jul 15, 2017 — Williams **syndrome**, which affects 30000 people in the U.S., is often ... she didn't have the same overwhelmingly **friendly**, outgoing **personality**.

CHAT CHECK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www.nationalgeographic.com › science › article › willi...
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William syndrome

CHAT CHECK

What test should you order?

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C) Chromosomes
D) FISH
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William syndrome = del 7q11.23

CHAT CHECK

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William syndrome = del 7q11.23

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Seizures

CHAT CHECK

What test should you order?

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Turner syndrome; 45,X

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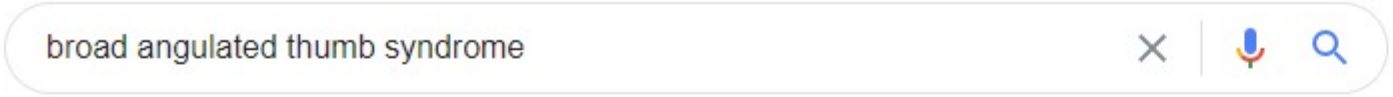


rarediseases.org › Rare Diseases

Rubinstein Taybi Syndrome - NORD (National Organization ...

RSTS is characterized by growth delays, distinctive facial features, intellectual disability (with an average IQ of 36-51), abnormally **broad** and often **angulated thumbs** and great toes (halluces), and feeding difficulties (dysphagia).

General Discussion · Signs & Symptoms · Causes · Related Disorders

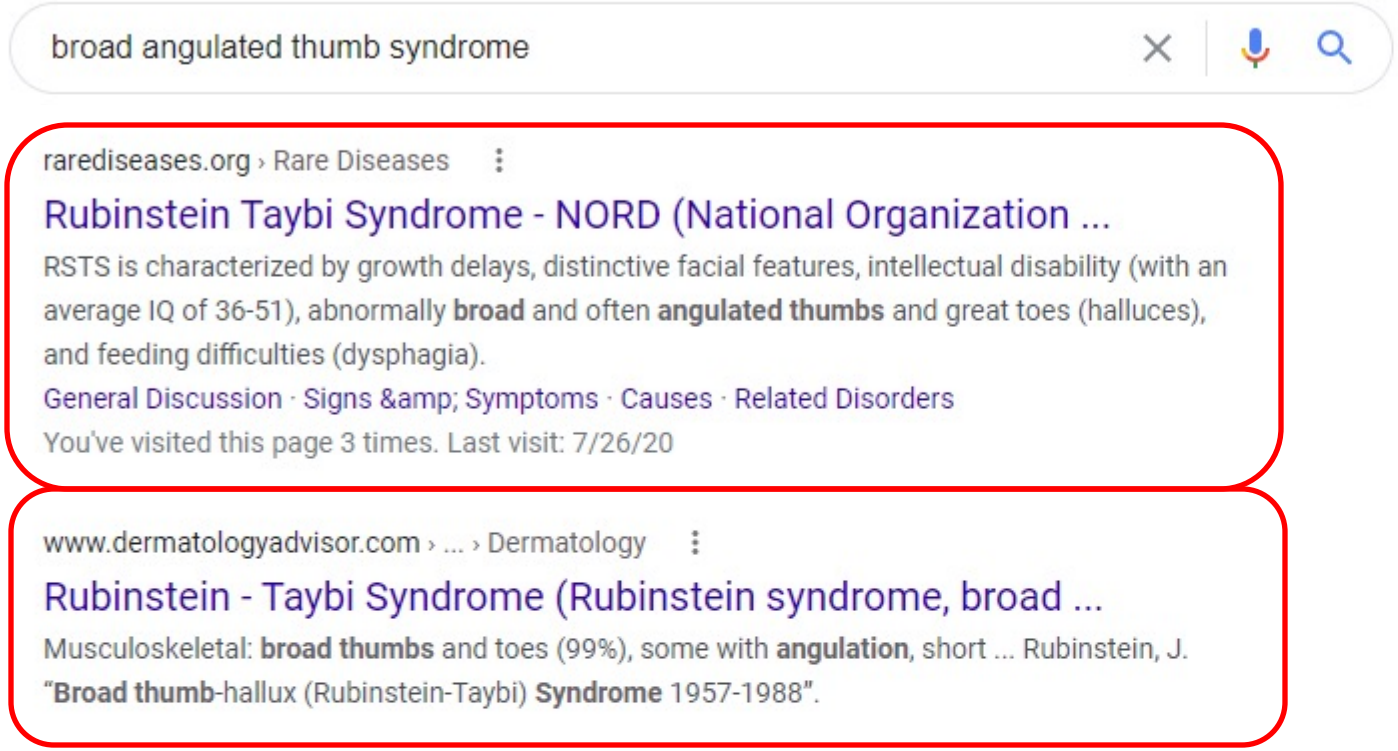
You've visited this page 3 times. Last visit: 7/26/20

www.dermatologyadvisor.com › ... › Dermatology

Rubinstein - Taybi Syndrome (Rubinstein syndrome, broad ...

Musculoskeletal: **broad thumbs** and toes (99%), some with **angulation**, short ... Rubinstein, J. "**Broad thumb**-hallux (Rubinstein-Taybi) **Syndrome** 1957-1988".

CHAT CHECK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rarediseases.org › Rare Diseases
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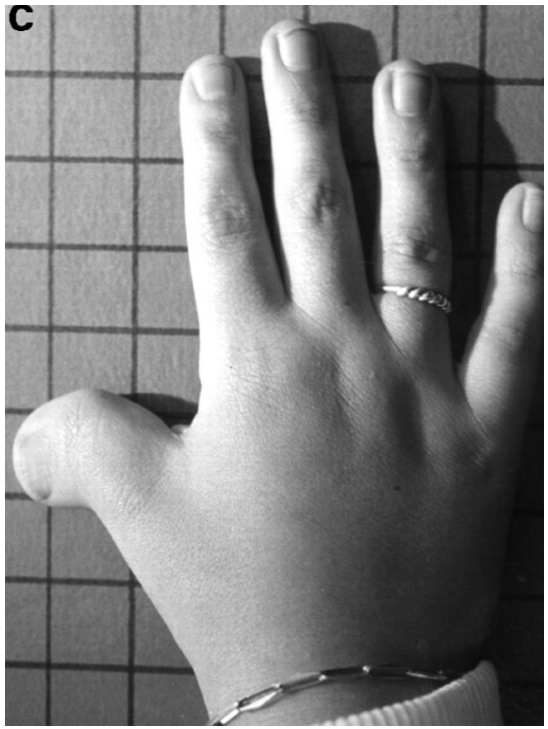

Rubinstein-Taybi

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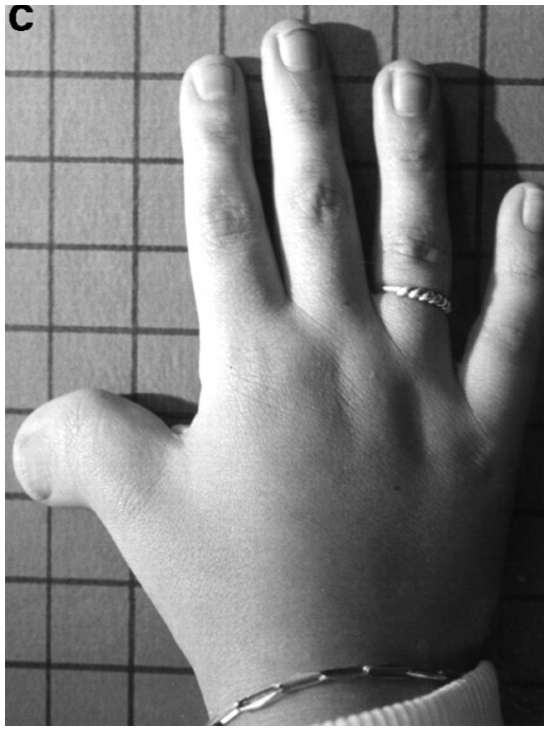
E) CMA
F) CMA then WES



Rubinstein-Taybi = 2 genes

CHAT CHECK

What test should you order?

Time Limit: 15 sec

A) Single gene
B) Gene panel
C) Chromosomes
D) FISH
E) CMA
F) CMA then WES

Rubinstein-Taybi = 2 genes

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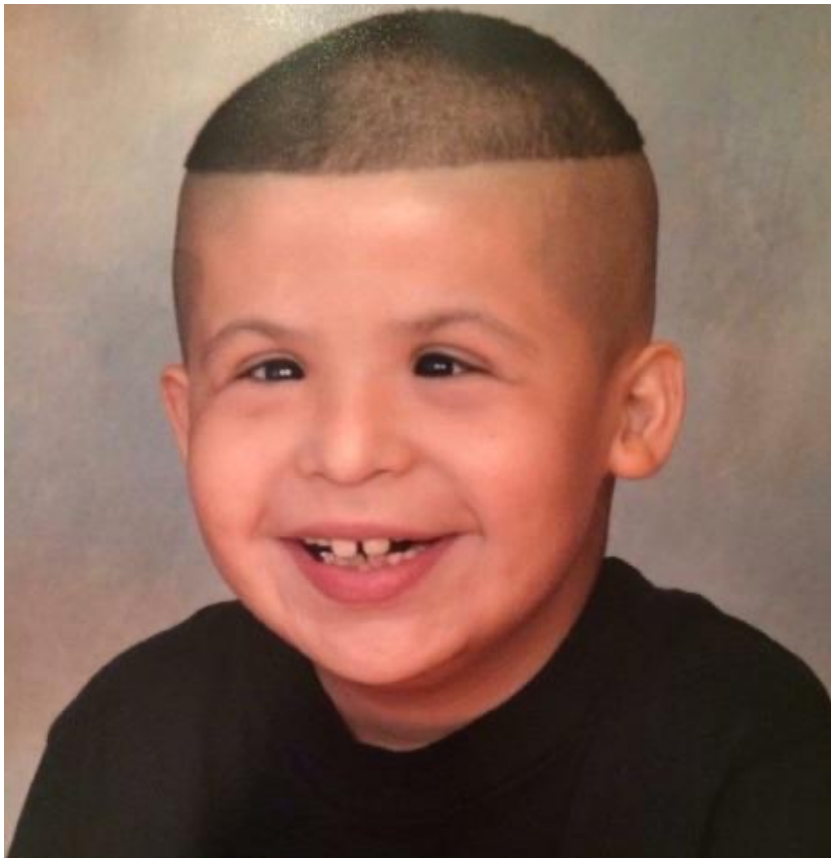

VSD, LVNC, DD, ID, G-Tube

CHAT CHECK

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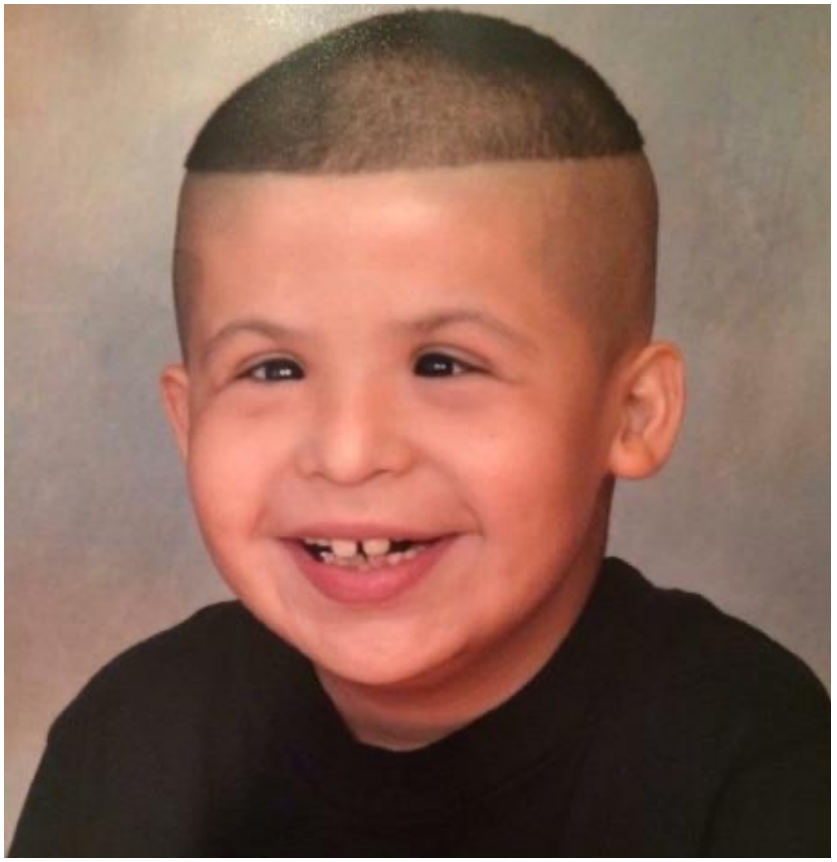

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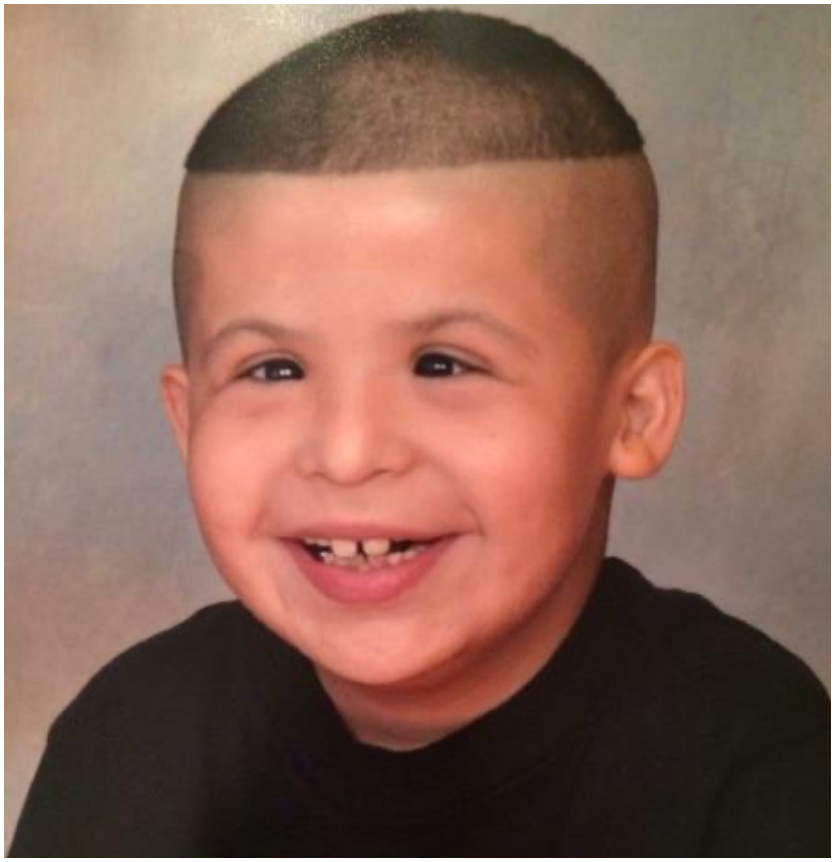


NONO-Related Disorder

CHAT CHECK

What test should you order?

Time Limit: 15 sec

A) Single gene
B) Gene panel
C) Chromosomes
D) FISH
E) CMA
F) CMA then WES

Intellectual disability

CHAT CHECK

What test should you order?

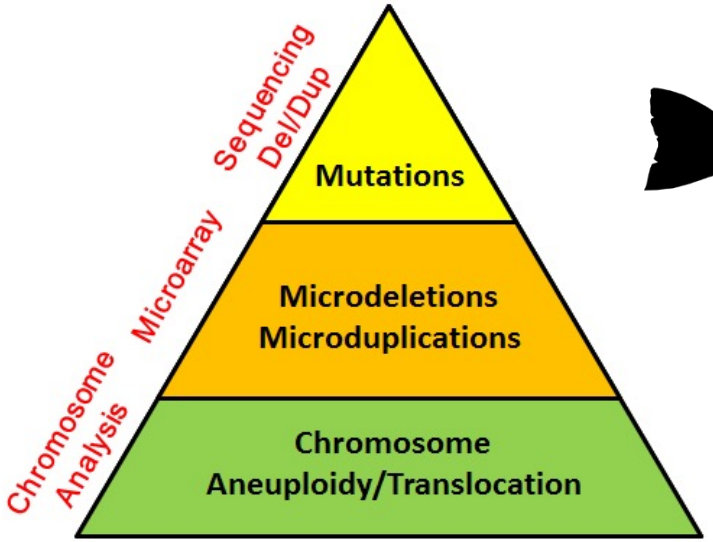
Time Limit: 15 sec

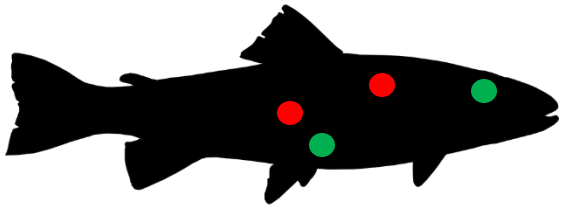
A) Single gene
B) Gene panel
C) Chromosomes
D) FISH
E) CMA
F) CMA then WES

Intellectual disability

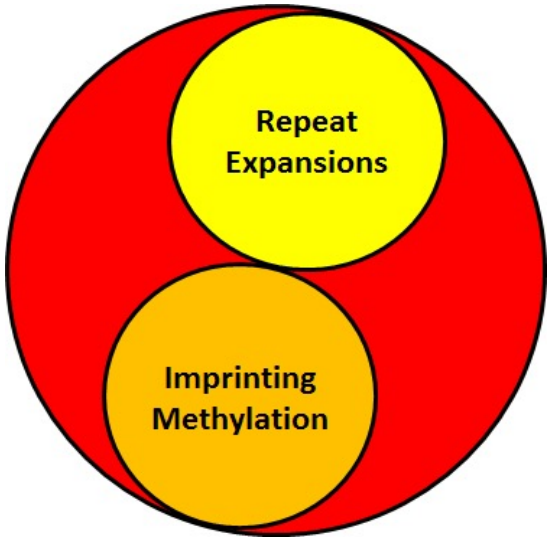
Take Home Messages

- **Do I know the diagnosis?**





Emergency
Confirmation
Chromosome
Microdeletion
Microduplication



- **Google**
- **Geneticist**
- **Genome-Wide testing (CMA and WES)**