AUTISM:
Helping Our Families Get the Right Support

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NEMOURS
CHILDREN'S HEALTH

Disclosure

- I have nothing to disclose

NEMOURS
CHILDREN'S HEALTH

Objectives

- Describe the recommended work up for children diagnosed with autism spectrum disorder
- Describe when a child diagnosed with autism needs an MRI of the Brain
- List the core therapies that are recommended a child receive when diagnosed with autism spectrum disorder

NEMOURS
CHILDREN'S HEALTH


Physical Health
Mental Emotional Behavioral Health
Family Dynamics
Learning
Success

ADOS – 2 Normed down to age 12 months

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CLINICAL REPORT Guidance for the Clinician in Rendering Pediatric Care

American Academy of Pediatrics
DEDICATED TO THE HEALTH OF ALL CHILDREN™

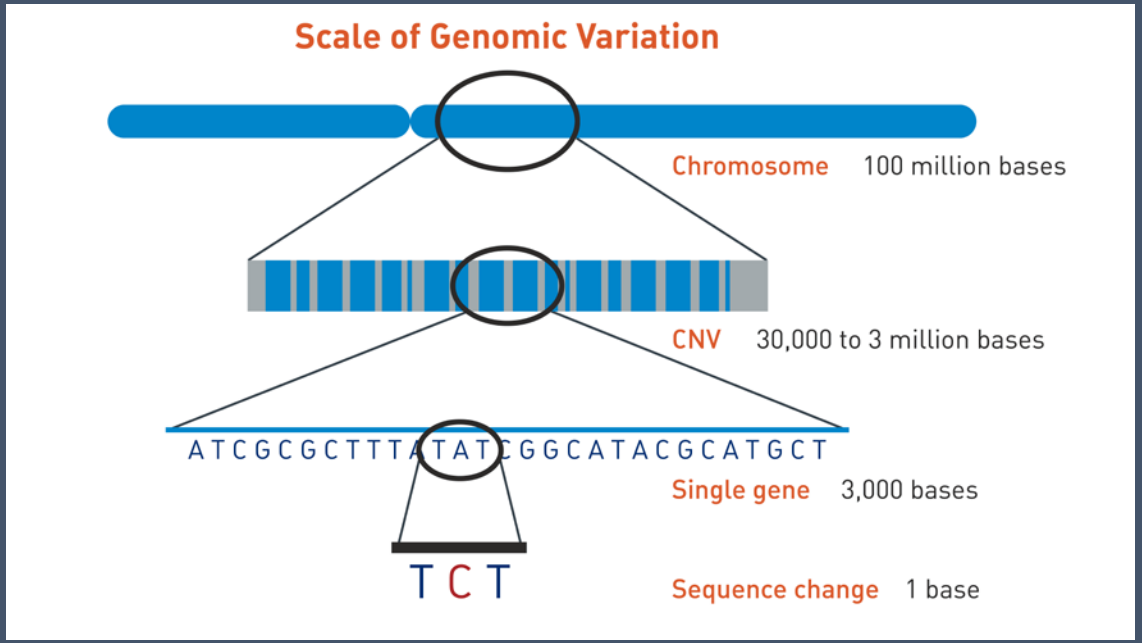
Identification, Evaluation, and Management of Children With Autism Spectrum Disorder

Susan L. Hyman, MD, FAAP,[a] Susan E. Levy, MD, MPH, FAAP,[b] Scott M. Myers, MD, FAAP,[c] COUNCIL ON CHILDREN WITH DISABILITIES, SECTION ON DEVELOPMENTAL AND BEHAVIORAL PEDIATRICS

TABLE 8 Genetic Etiologic Investigations in Patients With ASD

Step	Genetic Etiologic Investigations
1	Consider referral for pediatric genetics evaluation
2	Comprehensive history (including 3-generation family history with emphasis on individuals with ASD and other developmental, behavioral and/or psychiatric, and neurologic diagnoses) Physical examination (including dysmorphology, growth parameters [including head circumference], and skin examination) • If syndrome diagnosis or metabolic disorder is suspected, go back to step 1 (genetics and/or metabolism referral) and/or order the appropriate targeted testing • Otherwise, proceed to step 3
3	Laboratory studies • Discuss and offer CMA analysis • Discuss and offer fragile X analysis; if family history is suggestive of sex-linked intellectual disabilities, refer to genetics for additional testing • If patient is a girl, consider evaluation for Rett syndrome, *MECP2* testing • If these studies do not reveal the etiology, proceed to step 4
4	Consider referral to genetics, workup might include WES

Poorly Understood
Rare variants appear causally related
No pathologic variant accounts for >1%
No specific mutation is unique to ASD



Chromosomal Microarray
- dup/del – alter fxn of gene
- 5.4-14% pathogenic & 17-42% VUCS

Whole Exome Sequencing
- Single nucleotide variants – loss of fxn mutation and missense mutation
- Dx yield of 8-20% in those with ASD

Metabolic Testing – Low Yield

- **Atypical regression (> 2 yr)**
- **Motor regression or Multiple regressions**
- **FHx – Early Childhood Death or Metabolic d/o**
- **PE – Significant Hypotonia, Dysmorphic Features**
- **Visual or Hearing Impairment**

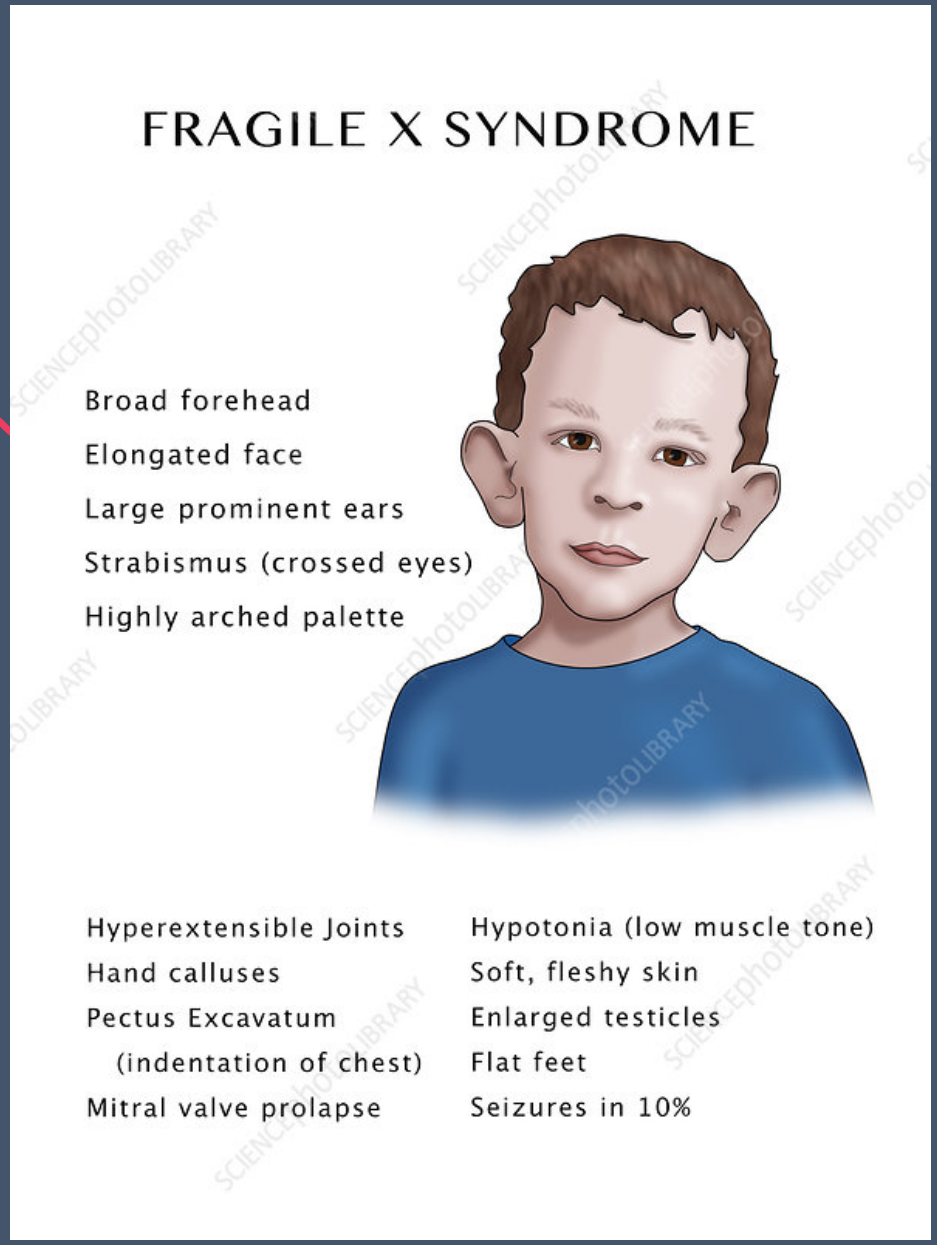
Fasting Amino Acids
Urine Organic Acids
Acylcarnitine Metabolite

FXS most common cause of inherited ID

FXS is most common known single-gene disorder in ASD cases

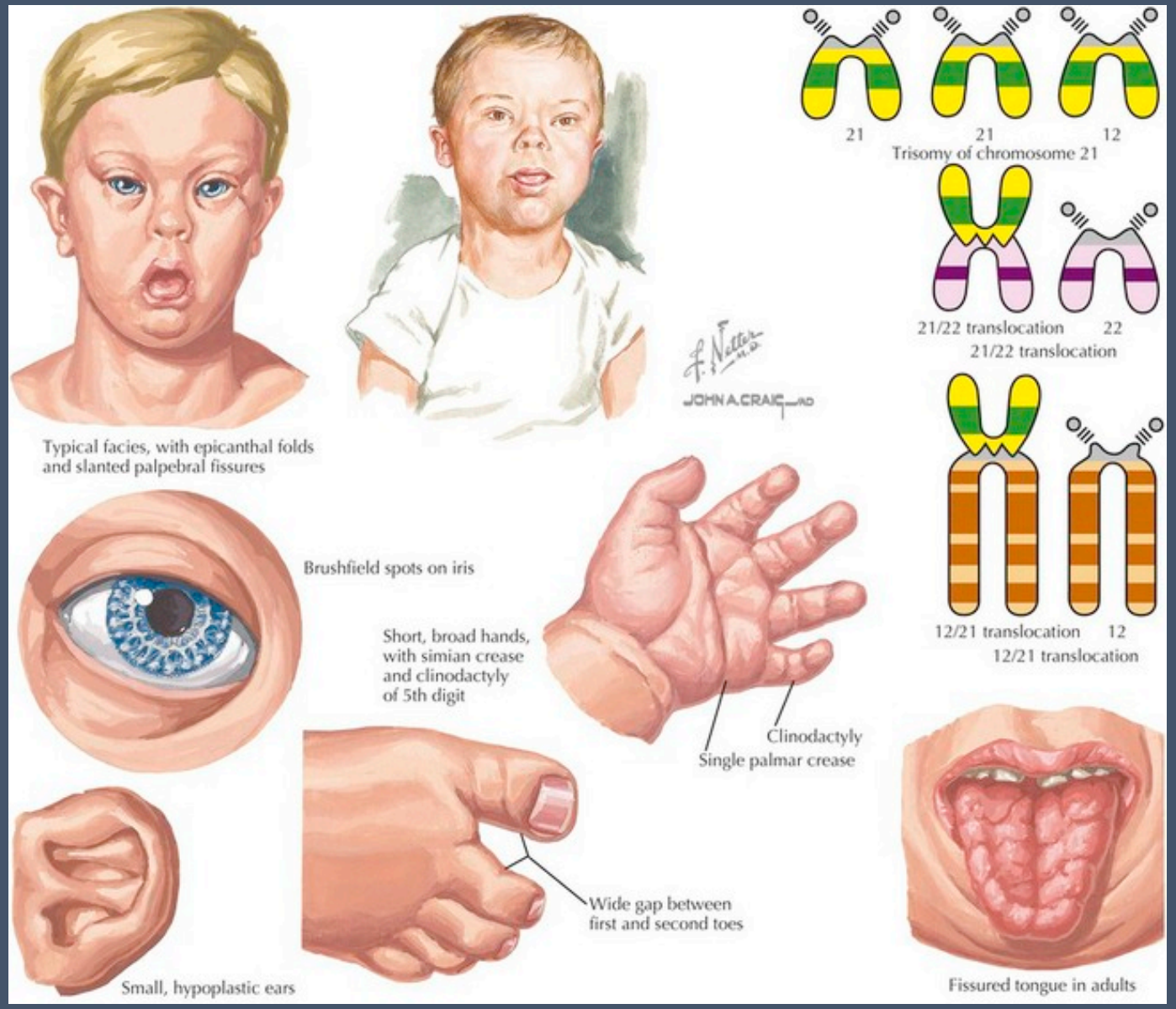
2-3% of all children with ASD have FXS

25-33% of FXS patients have ASD



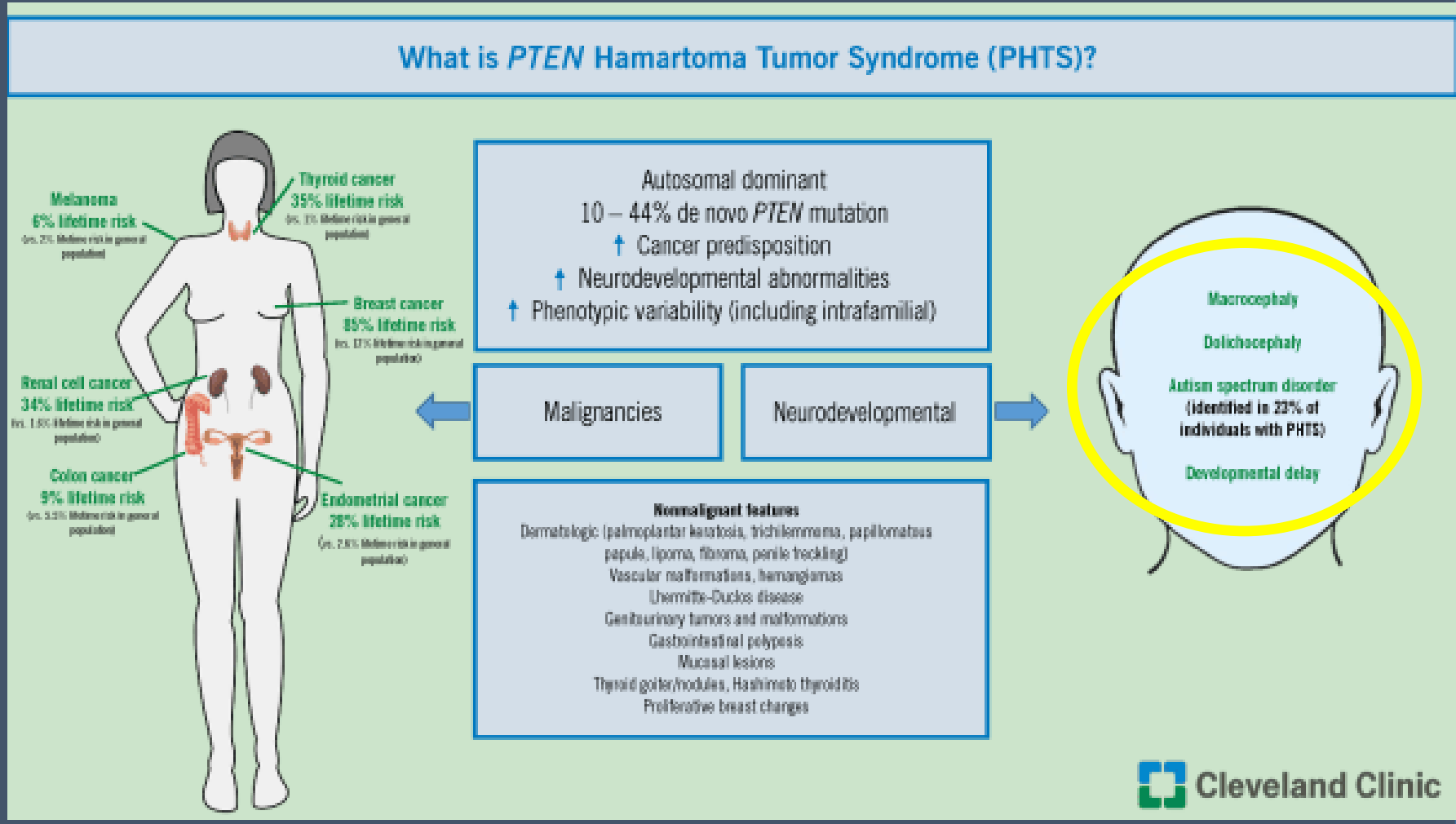
Down Syndrome

Up to 40% of children with DS have ASD



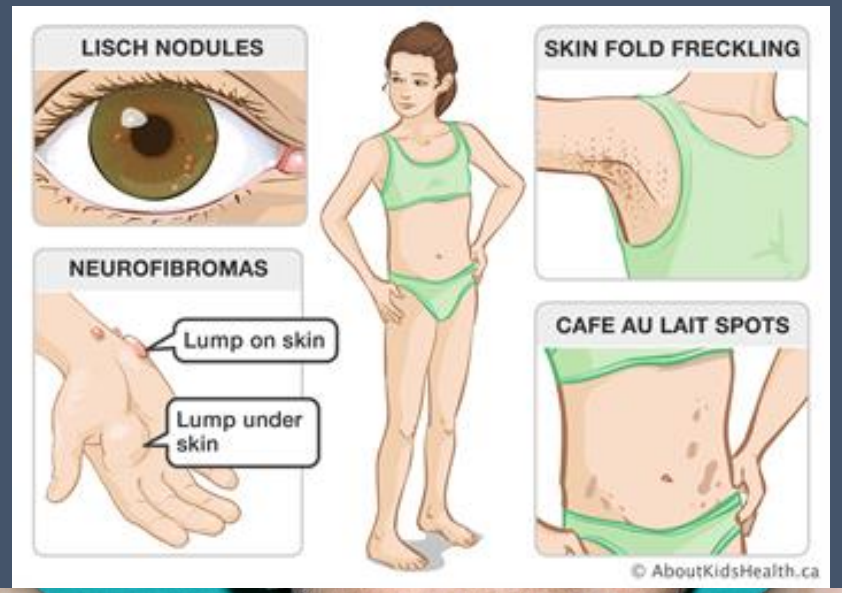
Rett Syndrome: Typical Development until 6-18 mo of age when see missed milestones or regression

PTEN is most frequently mutated tumor suppressor genes in human cancer



Neurofibromatosis

ADHD
LD Read / Math
Fine / Gross Motor
Plexiform Neurofibroma
Optic Nerve Glioma
HTN
Scoliosis
Seizures
Congenital Heart Defect



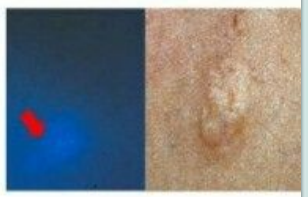
Tuberous Sclerosis

Skin signs in **Tuberous sclerosis (Epiloia)**:

- **Adenoma sebaceum:** (angiofibroma): Red papules on the face containing many blood vessels

- **Retinal hamartoma**

- **Shagreen spots:** Raised patches of skin with an orange-peel texture **often on the back**



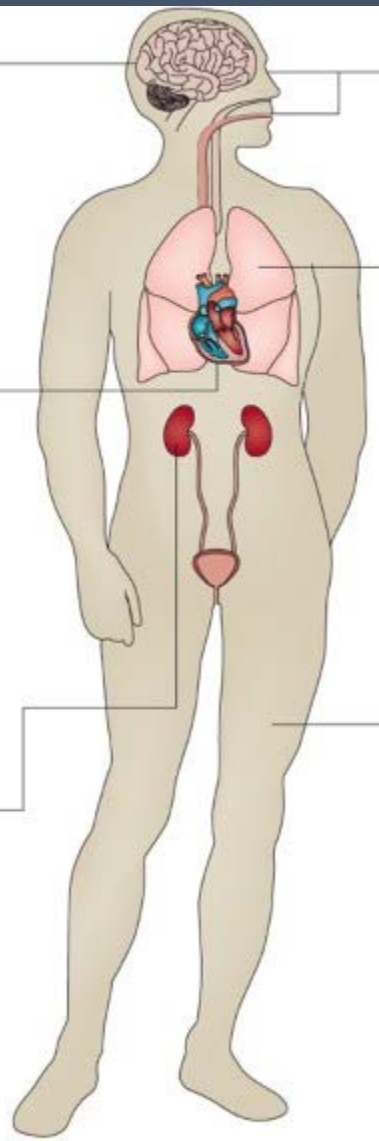
- **Ash leaf spots:** White areas of skin that look like an ash leaf

Brain
- 90% epilepsy
- 80–90% SEN
- 10–15% SEGA
- 90% TAND
- 50% intellectual disability
- 40% autism spectrum disorder

Heart
Infants
- 90% cardiac rhabdomyoma

Adults
- 20% cardiac rhabdomyoma

Kidney
- 70% angiomyolipoma
- 35% simple multiple cysts
- 5% polycystic kidney disease
- 2–3% renal cell carcinoma



Other
- 50% oral fibromas
- 50% retinal astrocytic hamartomas

Lung
Women
- 80% asymptomatic LAM
- 5–10% symptomatic LAM, can lead to respiratory failure

Men and women
- 10% MMPH

Skin
- 75% angiofibroma
- 20–80% ungual fibroma
- 25% fibrous cephalic plaques
- >50% shagreen patches
- 90% focal hypopigmentation

Nature Reviews | Disease Primers

Muscular Dystrophy

Not walking16-18 mo

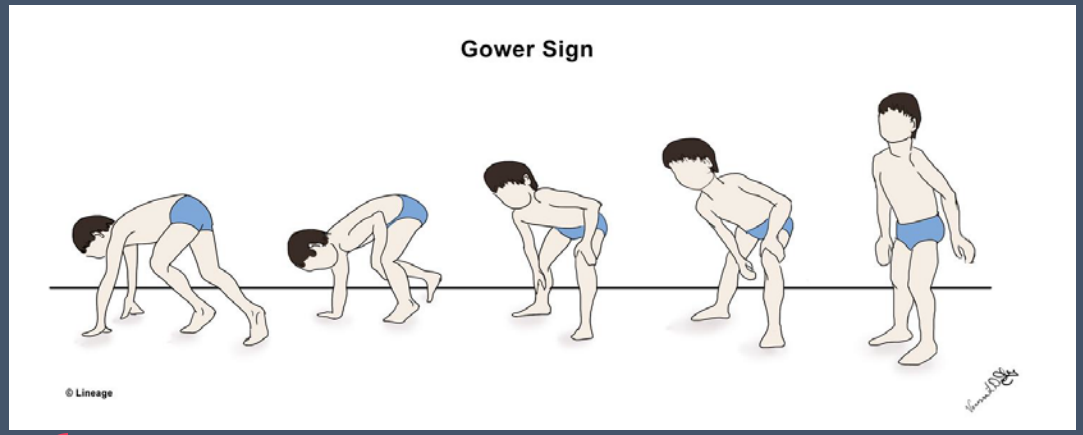
Toe Walking Calf Hypertrophy

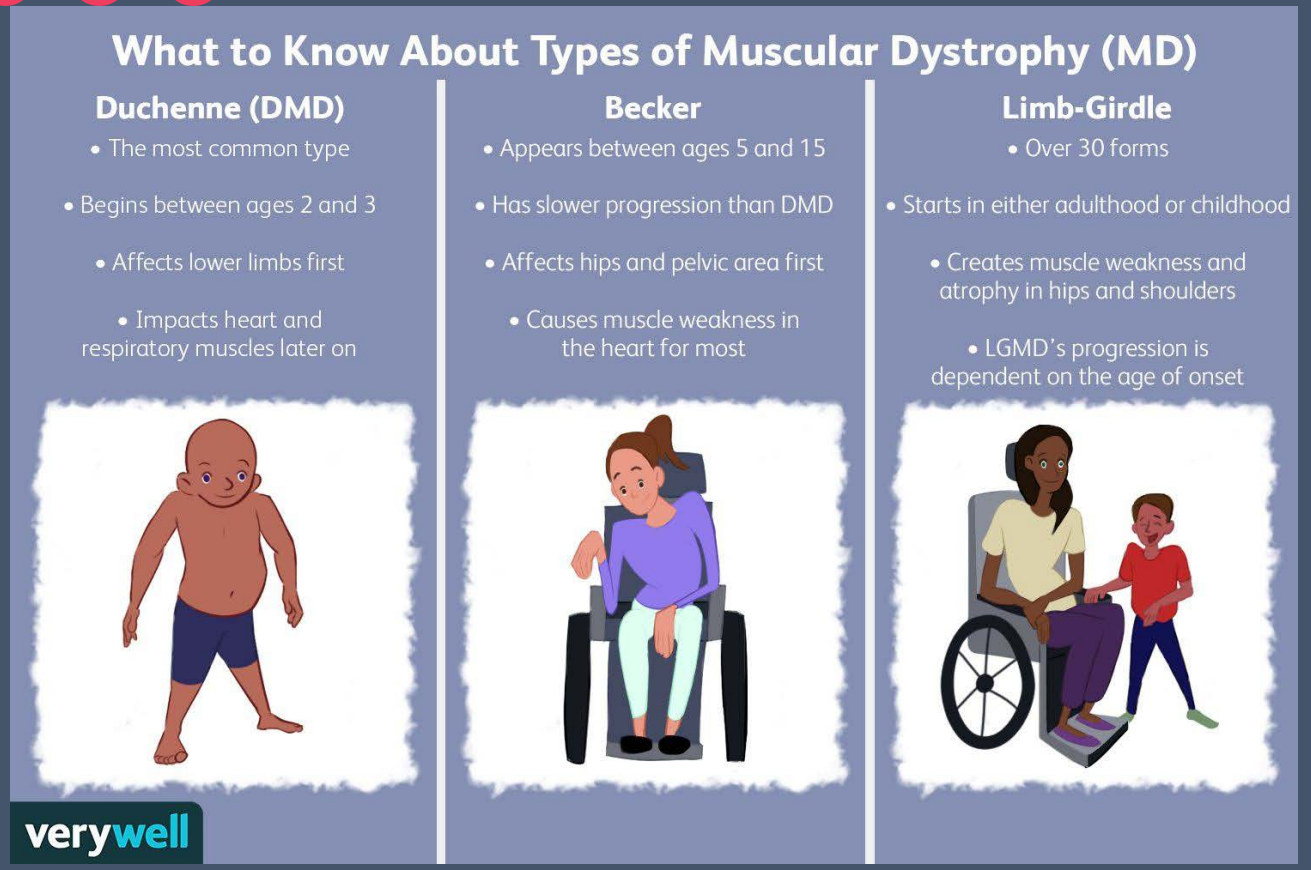
Weakness – Prox > Distal

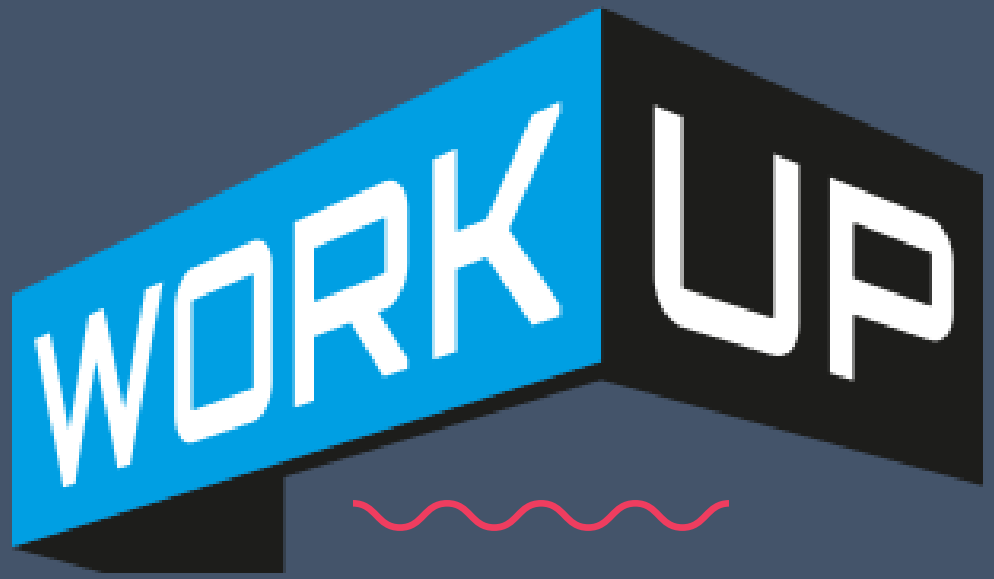
Hypotonia

Muscle pain or cramp

TEST – Creatine Kinase

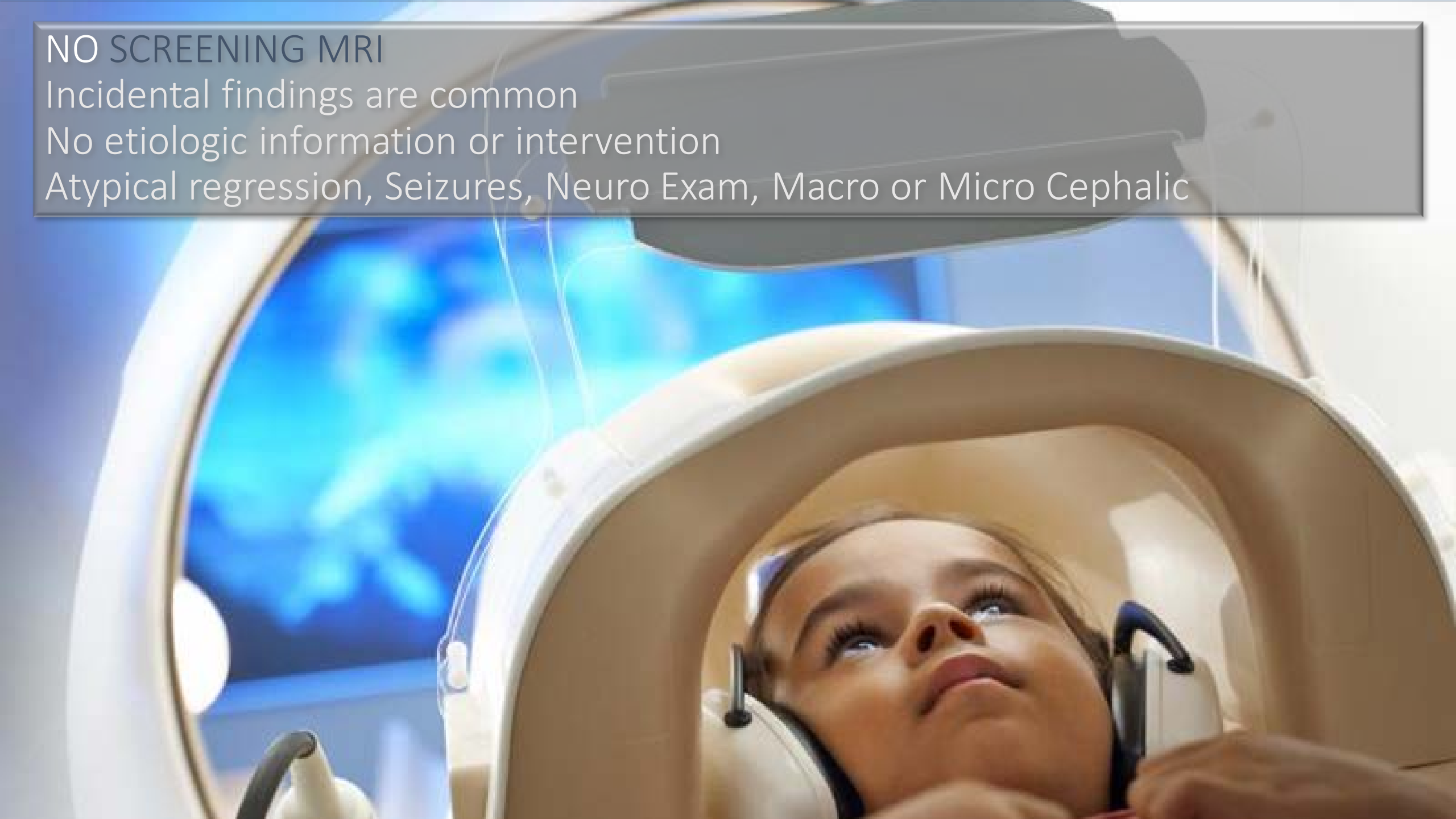






CMA – dup/del – alter fxn of gene (5.4-14% pathogenic & 17-42% VUCS)
FRX – boys and girls (FHx of Male with ID)
MECP2 – female – deceleration in head growth velocity, midline hand mvmt
PTEN – macrocephaly (pigmented macules on penis)
Creatine Kinase and TSH – motor delay or hypotonia


NO SCREENING EEG
Atypical Regression, History, or
Neuro Exam



NO SCREENING MRI

Incidental findings are common

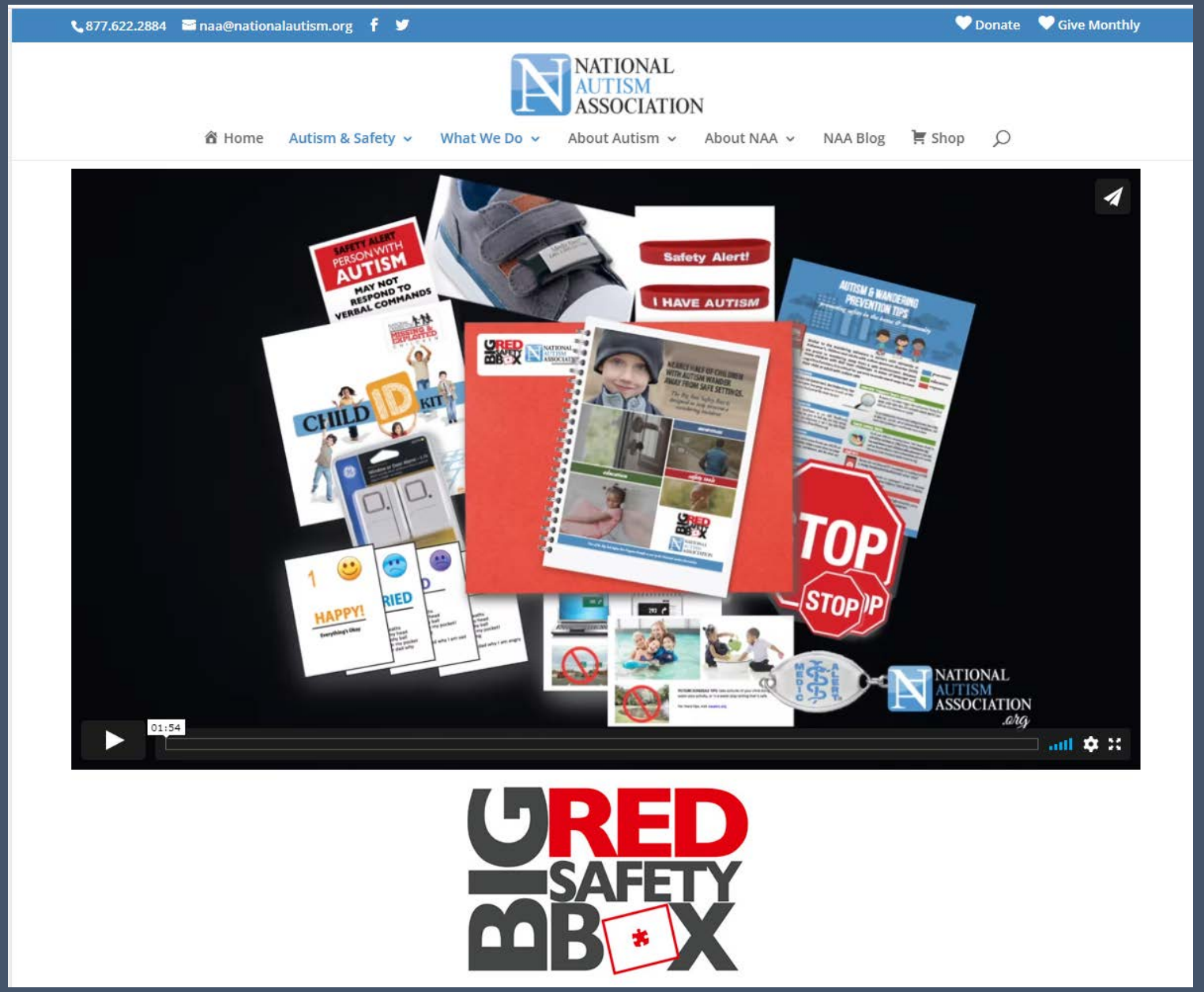
No etiologic information or intervention

Atypical regression, Seizures, Neuro Exam, Macro or Micro Cephalic

50% of 1,218 4-10 yr with ASD elope
~50% Called police
~2/3 Risk for traffic-related injury
~1/3 Near drowning


Enjoy Running
Desired Location
Pursue Intense Interest
Escape from Situation / Sensation

Prevention
- Deadbolt
- Fence
- GPS
- ID bracelet
- Alert Neighbor
- Alert LE
- Teach Safety Skill



HOW CAN SPEECH & LANGUAGE THERAPY HELP ME?

Speech Skills
- Formulate sounds into clear words
- Produce age appropriate sounds
- Help others easily understand what I am saying

Social Skills
- Play with my friends
- Attention to task
- Interactions with others
- Request help
- Express feelings
- Strengthen communication skills (eye contact & greetings)

Expressive Language Skills
- Communicate my wants and needs
- Formulate Sentences
- Put my thoughts into words
- Express Various Concepts
- Use words appropriately
- Use appropriate grammar

Cognitive Communications
- Organize thoughts
- Solve problems
- Draw conclusions
- Manage time

Receptive Language Skills
- Understand & follow directions
- Understand what I am hearing
- Respond appropriately to questions
- Recall information
- Answer yes and no questions
- Answer who, what, where, when and how questions

Voice & Fluency
- Demonstrate appropriate loudness
- Produce fluent words and phrases

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OCCUPATIONAL THERAPY

Skills for Life

WE HELP KIDS WITH:

Handwriting & cutting.

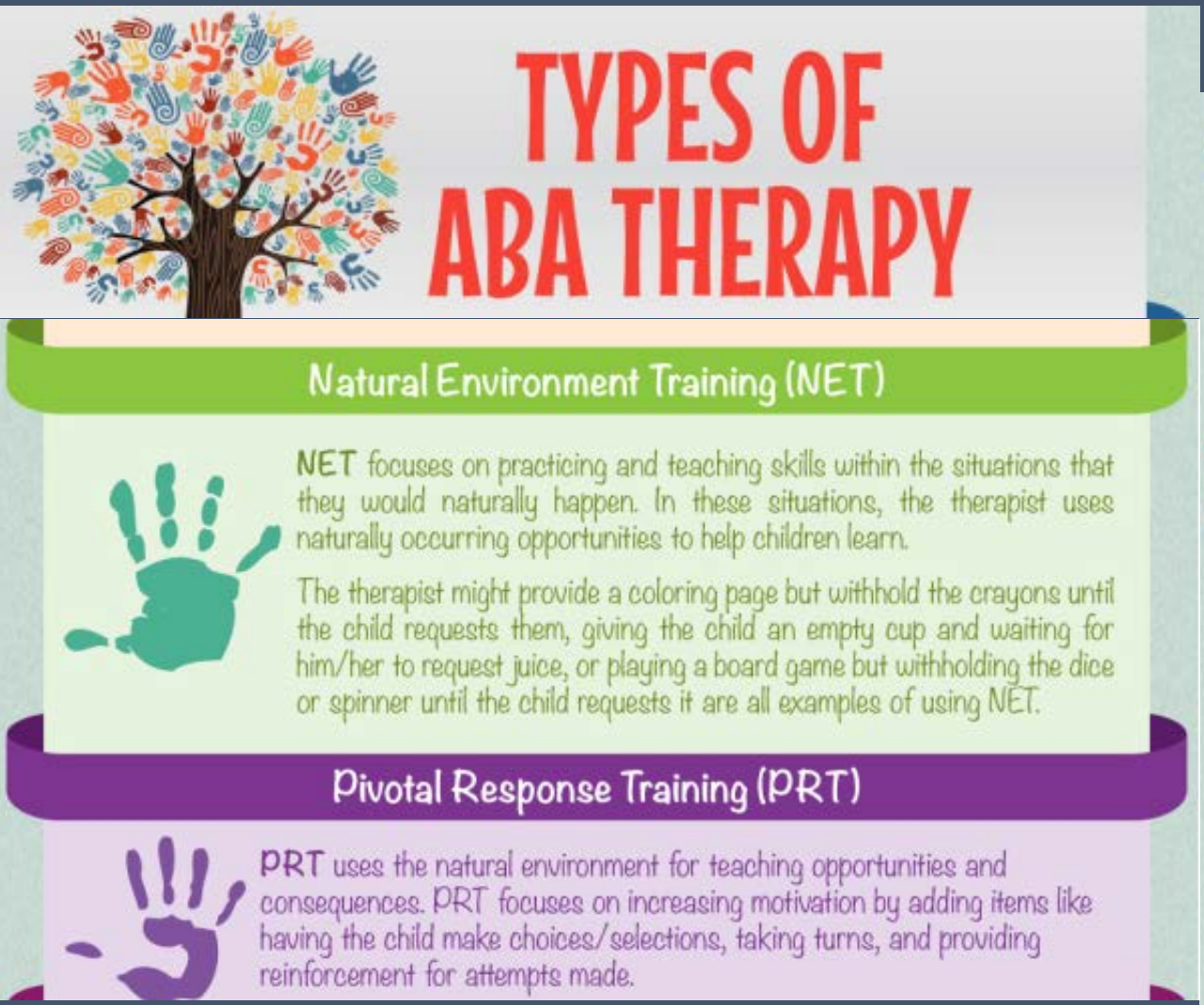
Social & emotional communication.

Core & posture strength.

Tying shoes, zippering & buttoning.

Using eating utensils & incorporating food variety.

Self regulation, calming & focus.



More intense ABA therapy associated with optimal developmental outcome


WHAT IS PARENT MANAGEMENT TRAINING (PMT)?


Parent
Support
Group

SKILLS GROUPS

Early Childhood 4-7 years	• Beginning Social Skills • School-Related Skills • Friendship Making Skills	• Dealing with Feelings • Alternatives to Aggression • Dealing with Stress
Elementary School 5-12 years	• Group Survival Skills • Friendship Making Skills • Dealing with Feelings	• Alternatives to Aggression • Dealing with Stress
Adolescents & Teens 13-18 years	• Beginning Social Skills • Advanced Social Skills • Dealing with Feelings	• Alternatives to Aggression • Dealing with Stress • Planning Skills
High Functioning Autism 10-18 years	• Relationship Skills • Social Comprehension • Self-Regulation	• Problem Solving • Understanding Emotions • School-Related Skills

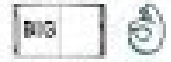
INDIVIDUAL TREATMENT

Clinic & Home Based	• Communication • Social Skills • Behavior Reduction	• Academics and Academic Preparedness • Self-help and Daily Living

The 14 Categories of Students With Disabilities

Autism

Deaf-Blindness



Deafness

Developmental Delay

Emotional Disturbed

Hearing Impairment

Intellectual Disability

Multiple Disabilities

Orthopedic Impairment

Other-Health Impairment

Includes a range of conditions such as having limited strength, vitality, or alertness, due to acute health problems

Traumatic Brain Injury

Specific Learning Disability

A disorder in basic psychological processes. The imperfect ability to listen, speak, read, write, & do math

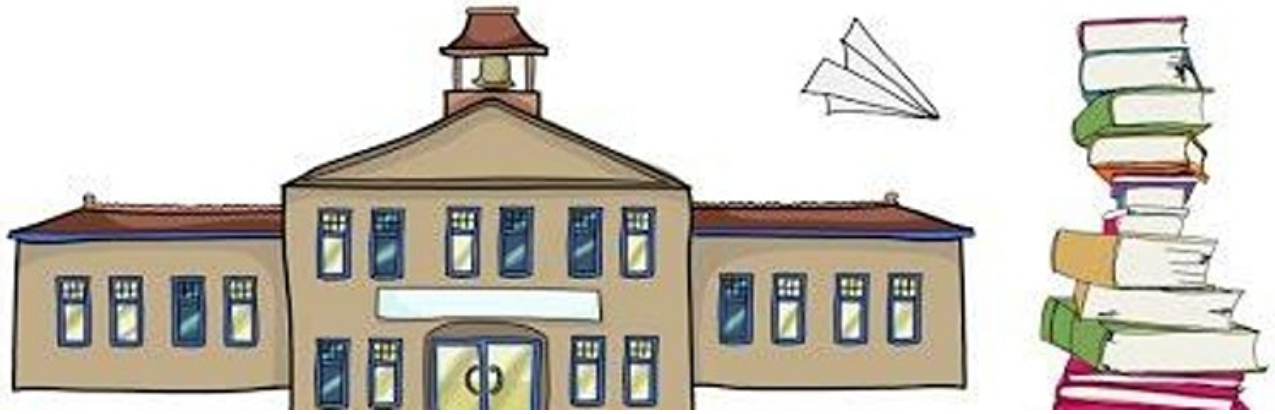
Speech Language Impairment

A communication disorder such as stuttering, impaired articulation & language, or voice impairment

Visual Impairment

IDEA

INDIVIDUALS WITH DISABILITIES EDUCATION ACT



4 PARTS OF THE I.D.E.A.

PART A

- Definitions of various terminology
- Outlines the necessity of the act
- Creates the Office of Special Education Programs

PART B

- Every child is entitled to a free and appropriate public education
- Students are entitled to evaluation of disability – meaning if a teacher believes a student to have a disability, the teacher may recommend an evaluation with a professional
- Creates the Individualized Education Program
- Child/Parent input must be taken into account in education process
- Due process to challenge inappropriate educational conditions is granted to all parents and students

PART C

- Identification and intervention services
- Individualized Family Service Plans that lay out priorities, resources, and concerns for the family
- Families have a right to participate in the creation of the IFSP
- Parents are entitled to timely resolution of all conflicts or complaints regarding these plans

PART D

- Grants to improve the education and transitional services provided to students with disabilities

The Harkin Institute | Drake University

harkininstitute.org @HarkinAtDrake The Harkin Institute

SOURCE: APA.ORG

THE INDIVIDUALS WITH DISABILITIES ACT

ORIGINALLY CALLED THE EDUCATION FOR ALL HANDICAPPED CHILDREN ACT OF 1975

FREE AND APPROPRIATE EDUCATION (FAPE)

STUDENTS GET A UNIQUE AND INDIVIDUALIZED EDUCATION AT THE EXPENSE OF THE PUBLIC

INCLUDES RELATED SERVICES

WORKS IN ACCORDANCE WITH THE I.E.P.

PARENT INVOLVEMENT

PARENTS MADE TO TAKE PART IN ELIGIBILITY DECISIONS FOR SPECIAL SERVICES.

PARENTS ARE ABLE TO CHALLENGE DECISIONS

LEAST RESTRICTIVE ENVIRONMENT (LRE)

LAW INTENDS FOR STUDENTS WITH DISABILITES TO BE EDUCATED, TO THE GREATEST EXTENT POSSIBLE, WITH STUDENTS THAT DO NOT HAVE A DISABILITY

GOAL TO MOVE THE CHILDREN INTO REGULAR CLASSROOMS

CONFIDENTIALITY PROTECTION

SPECIFIC EYES ONLY

THE SCHOOL HAS THE RESPONSIBILITY TO PROTECT THE CONFIDENTIALITY OF THE STUDENT

RELEVENT SCHOOL STAFF HAVE THE RIGHT TO REVIEW THE DOCUMENT

INDIVIDUALIZED EDUCATION PLAN (I.E.P.)

MODIFICATIONS
ACCOMMODATIONS
RELATED SERVICES
DURATION OF SERVICES
PLEP

MANIFESTATION

A CHILD SHOULD NOT BE PUNISHED FOR A BEHAVIOR IF THE BEHAVIOR IS A DIRECT RESULT OF THE STUDENT'S DISABILITY

TRANSITION SERVICES

DESIGNED TO HELP STUDENTS TRANISTION FROM FROM SCHOOL TO A WORK ENVIRONMENT.

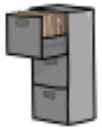
INCLUDES VOCATIONAL TRAINING

PROCEDURES FOR PROTECTION OF RIGHTS

PARENTS MUST CONSENT TO AN EVALUATION IN WRITING

IF BOTH PARENTS AND THE SCHOOLS CANNOT COME TO AN AGREEMENT, MEDIATION WILL TAKE PLACE

PARENTAL ACCESS TO STUDENT RECORDS

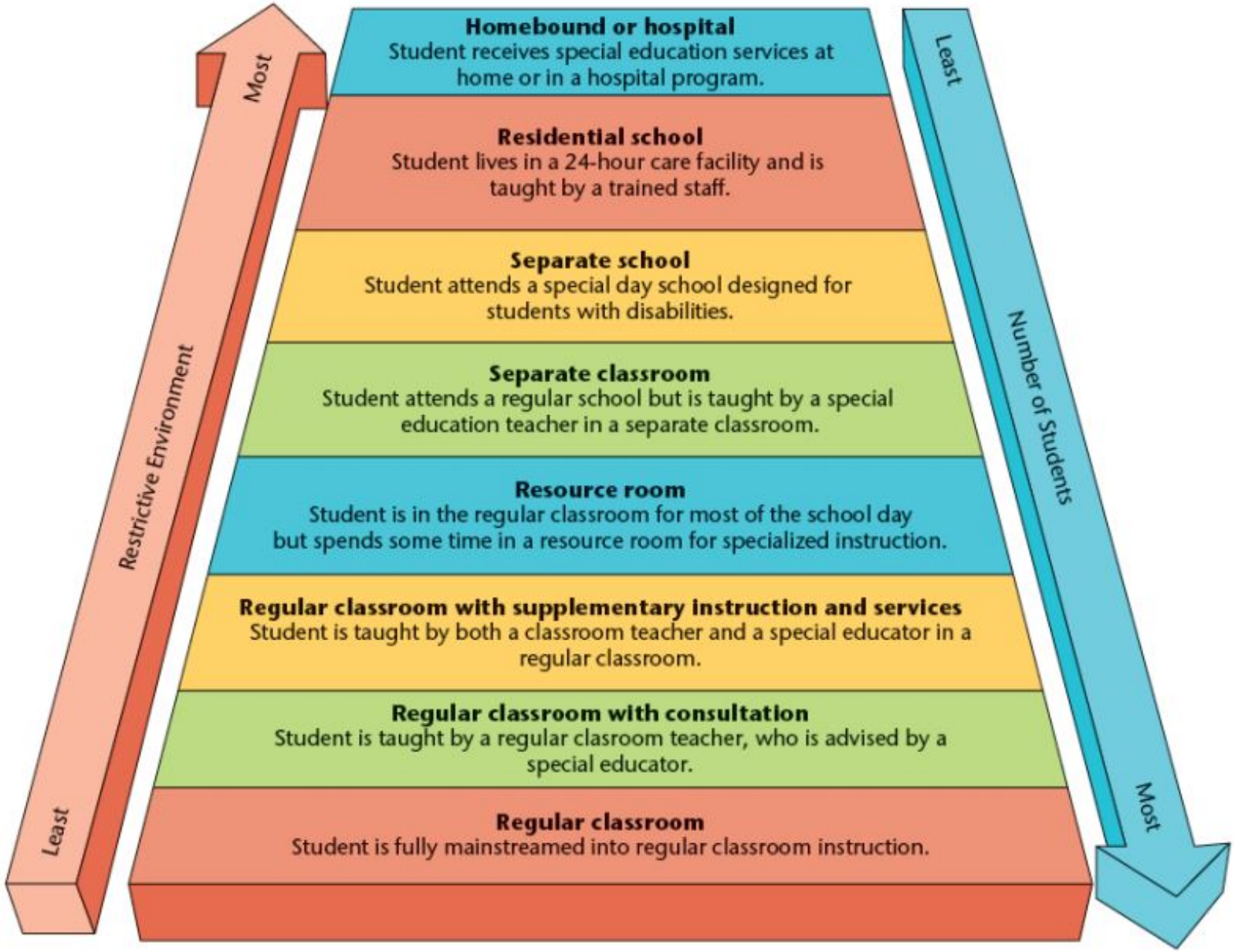


TRANSLATORS REQUIRED WHEN NEEDED

APPROPRIATE EVALUATIONS

SCHOOLS SHOULD USE A VARIETY OF ASSESSMENT METHODS IN THEIR EVALUATIONS

NEMOURS CHILDREN'S HEALTH


Most
Restrictive Environment
Least
Homebound or hospital
Student receives special education services at home or in a hospital program.
Residential school
Student lives in a 24-hour care facility and is taught by a trained staff.
Separate school
Student attends a special day school designed for students with disabilities.
Separate classroom
Student attends a regular school but is taught by a special education teacher in a separate classroom.
Resource room
Student is in the regular classroom for most of the school day but spends some time in a resource room for specialized instruction.
Regular classroom with supplementary instruction and services
Student is taught by both a classroom teacher and a special educator in a regular classroom.
Regular classroom with consultation
Student is taught by a regular clasroom teacher, who is advised by a special educator.
Regular classroom
Student is fully mainstreamed into regular classroom instruction.
Least
Number of Students
Most

Model Letter

Today's Date (include month, day, and year)

Your Name
Street Address
City, State, Zip Code
Daytime telephone number

Name of Principal or Special Education Administrator
Name of School
Street Address
City, State, Zip Code

Dear (person's name),

I am writing to request that my son/daughter, (child's name), be evaluated for special education services. I am worried that (child's name) is not doing well in school and believe he/she may need special services in order to learn. (Child's name) is in the (_) grade at (name of school). (Teacher's name) is his/her teacher.

Specifically, I am worried, because (child's name) does/does not (give a few direct examples of your child's problems at school).

We have tried the following to help (child's name): (If you or the school have done anything extra to help your child, briefly state it here).

I understand that I have to give written permission in order for (child's name) to be evaluated. Before the evaluation begins, I have some questions about the process that I need to have answered (list any questions you may have). I would be happy to talk with you about (child's name). You can send me information or call me during the day at (daytime telephone number). Thank you for your prompt attention to my request.

Sincerely,

Your name

cc: your child's principal (if letter is addressed to an administrator)
your child's teacher(s)

https://www.parentcenterhub.org/

Note: If your child has been identified as having a disability by professionals outside the school system, add the following sentence to the end of the first paragraph: "(Child's name) has been identified as having (name of disability) by (name of professional). Enclosed is a copy of the report(s) I have received that explains (child's name) condition."

Note: The "cc:" at the bottom of the letter means you are sending a copy of your letter to the people listed after the cc.

What is in the IEP

- Child's present level of academic and functional performance
- Annual Education Goals – Plan to Track Progress
- Related Services
- Timing of Services – Start, Frequency, Length
- Accommodations to learning environment
- Modifications to what child is expected to learn
- How participate in standardized tests
- How included in Gen Ed classes and school activities

Early Intervention

Developmental delay ≥ 1 domain

or

Condition with high probability to result in delay

- Enhance development of infants and toddlers with disabilities,
- minimize need for special education,
- maximize individual's long-term potential for independent living

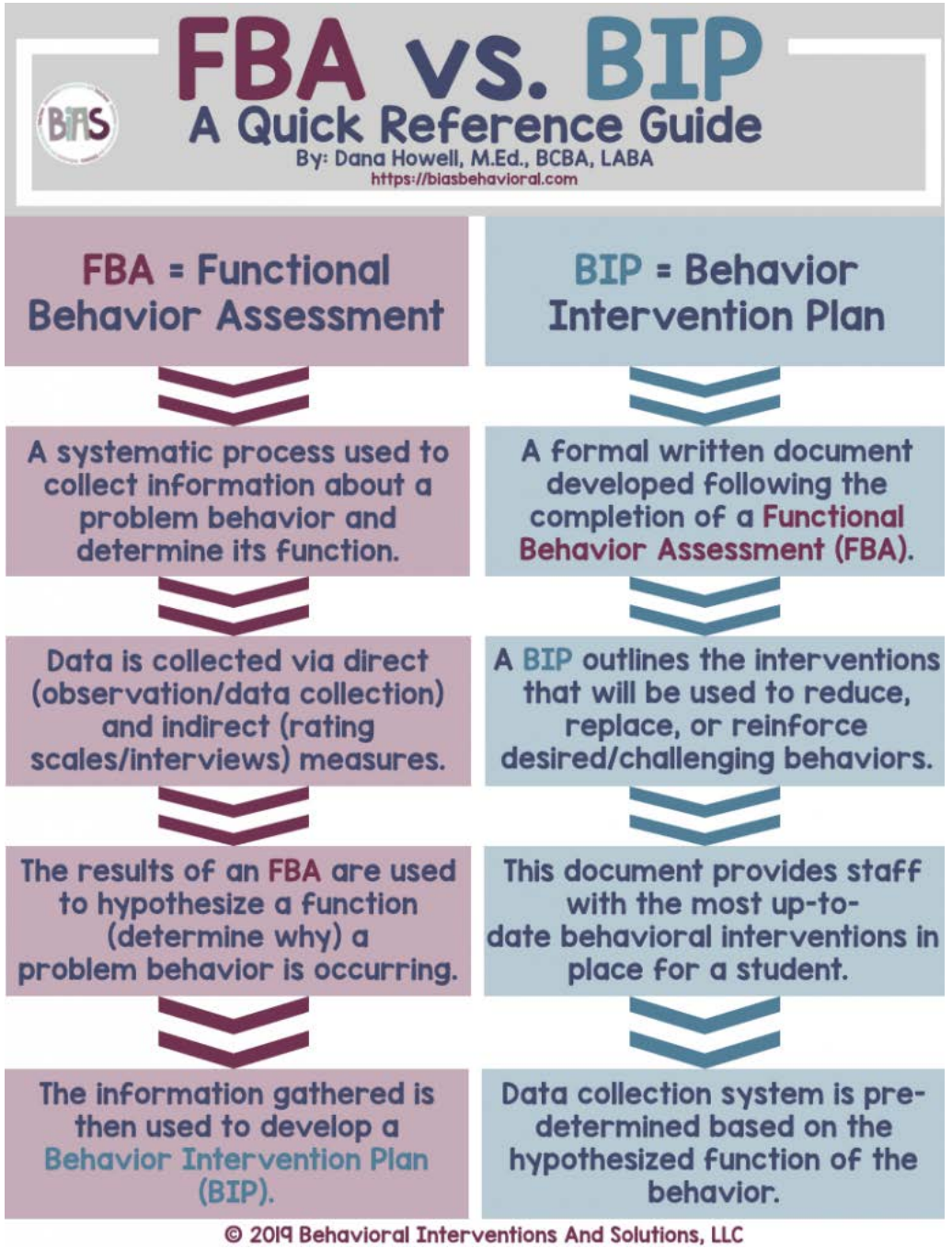
IFSP

- Part C of IDEA
- Birth – 3 yr of age
- Services in natural environment
- Service coordinator
- Transition plan (9 mo to 90 day)

IEP

- Part B of IDEA
- Age 3-21
- Services at school
- No service coordinator

Behavioral and Mental Health Issues




The End