



AUTISM: Helping Our Families Get the Right Support

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NEMOURS
CHILDREN'S HEALTH

Disclosure

- I have nothing to disclose

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



ADOS – 2
Normed
down to age
12 months



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CLINICAL REPORT

Guidance for the Clinician in Rendering Pediatric Care

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American Academy
of Pediatrics



1000 North Dearborn Street, Chicago, IL 60610

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, <i>MECP2</i> testing • If these studies do not reveal the etiology, proceed to step 4
4	Consider referral to genetics, workup might include WES

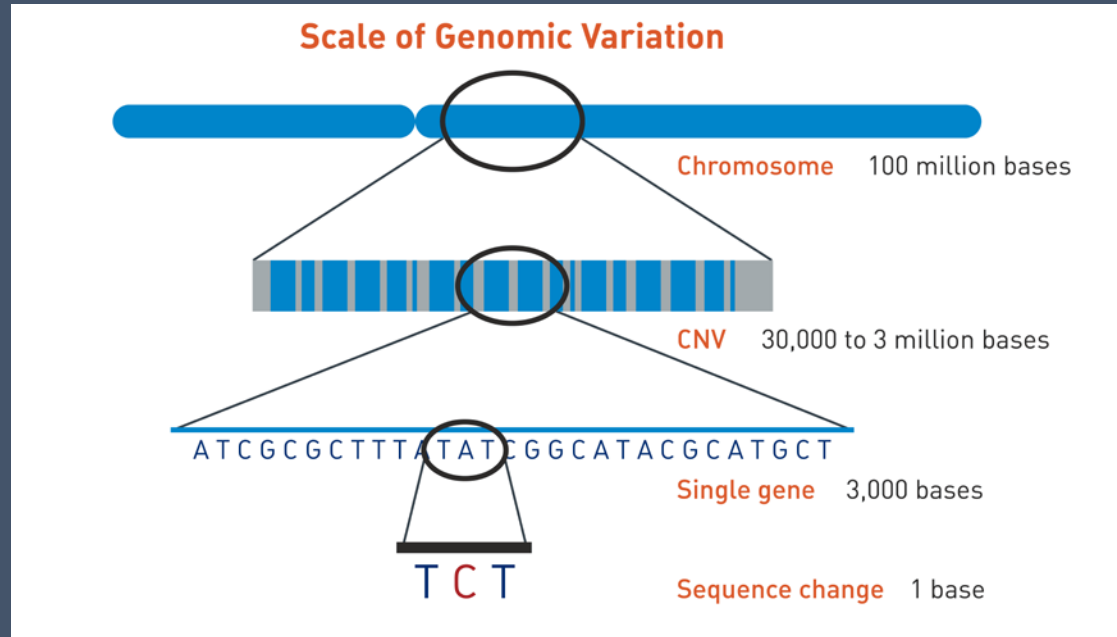
Poorly Understood

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



FLORIDA NEWBORN SCREENING

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

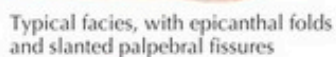
FRAGILE X SYNDROME

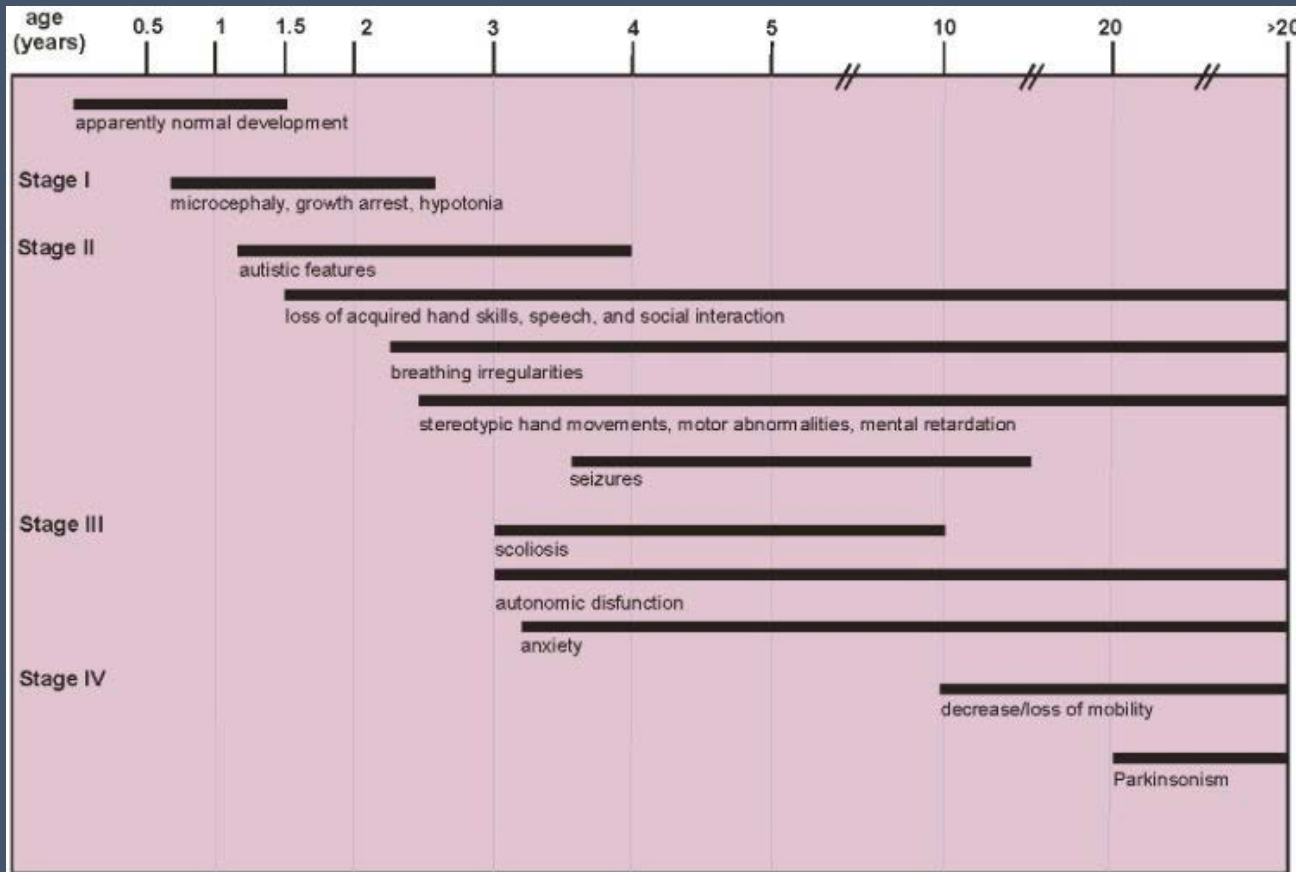
Broad forehead
Elongated face
Large prominent ears
Strabismus (crossed eyes)
Highly arched palate



Hyperextensible Joints	Hypotonia (low muscle tone)
Hand calluses	Soft, fleshy skin
Pectus Excavatum (indentation of chest)	Enlarged testicles
Mitral valve prolapse	Flat feet
	Seizures in 10%

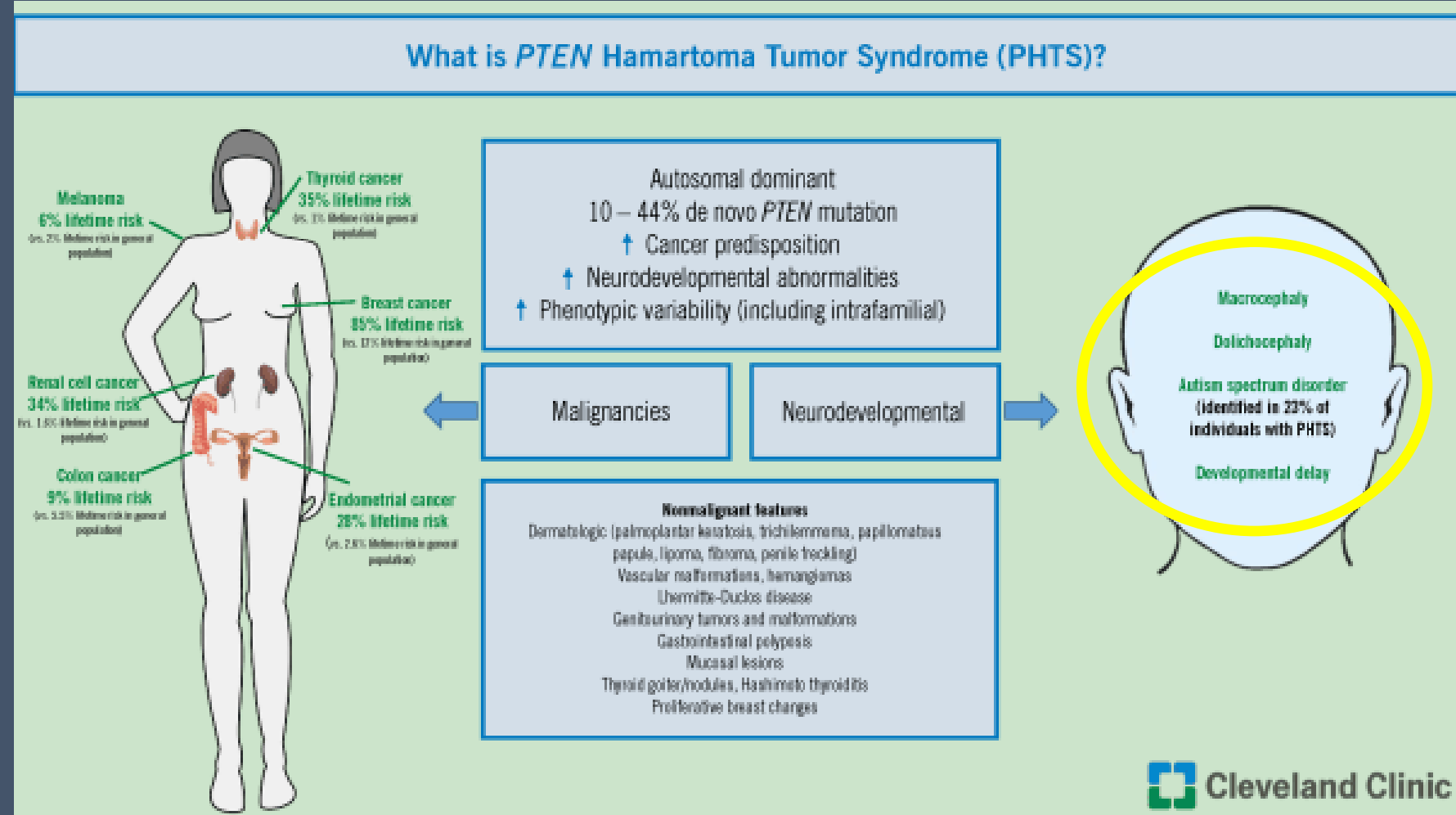
Up to 40% of children with DS have ASD





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

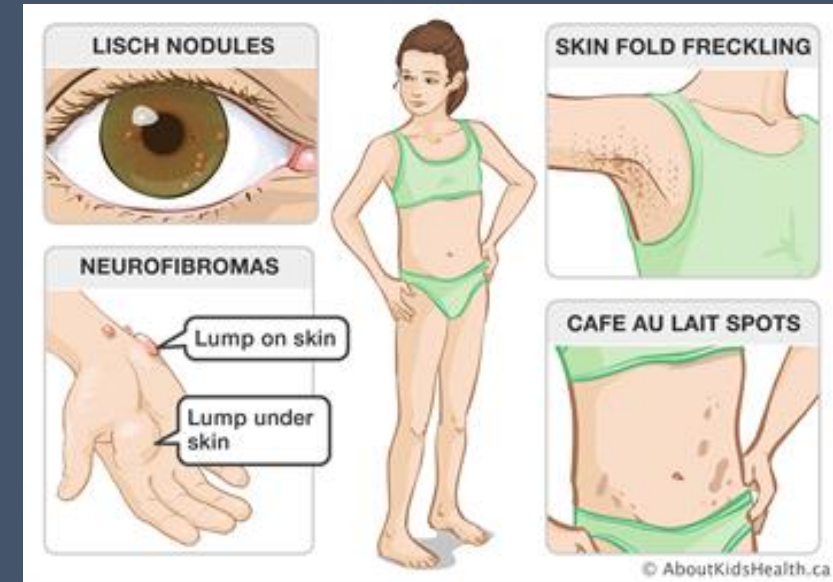
Optic Nerve Glioma

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

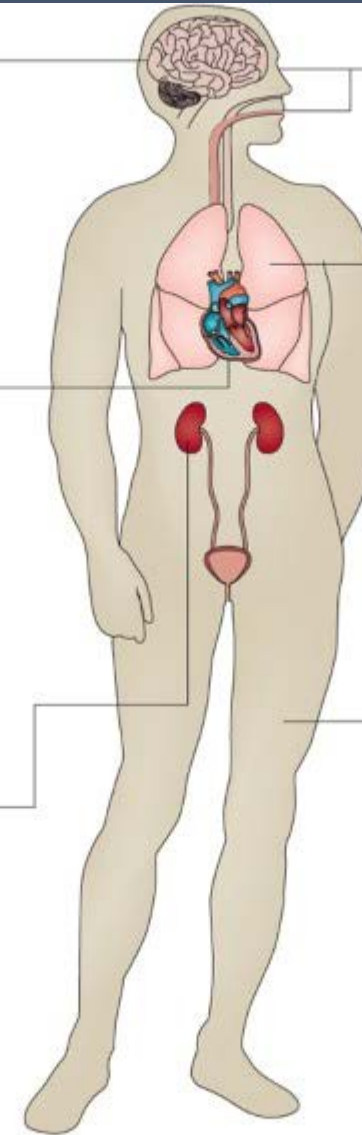
- 50% oral fibromas
- 50% retinal astrocytic hamartomas

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



Muscular Dystrophy

Not walking 16-18 mo

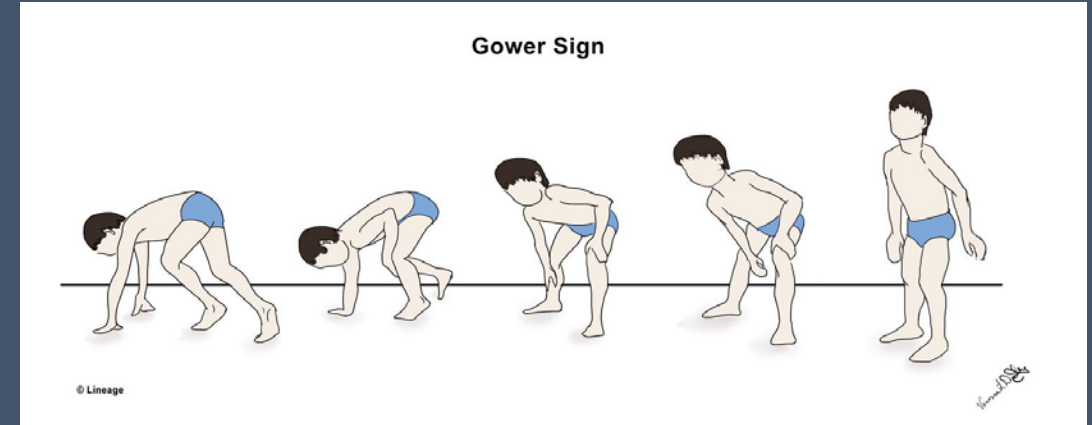
Toe Walking Calf Hypertrophy

Weakness – Prox > Distal

Hypotonia

Muscle pain or cramp

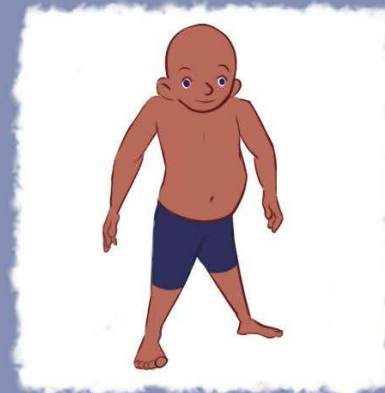
TEST – Creatine Kinase



What to Know About Types of Muscular Dystrophy (MD)

Duchenne (DMD)

- The most common type
- Begins between ages 2 and 3
 - Affects lower limbs first
 - Impacts heart and respiratory muscles later on



Becker

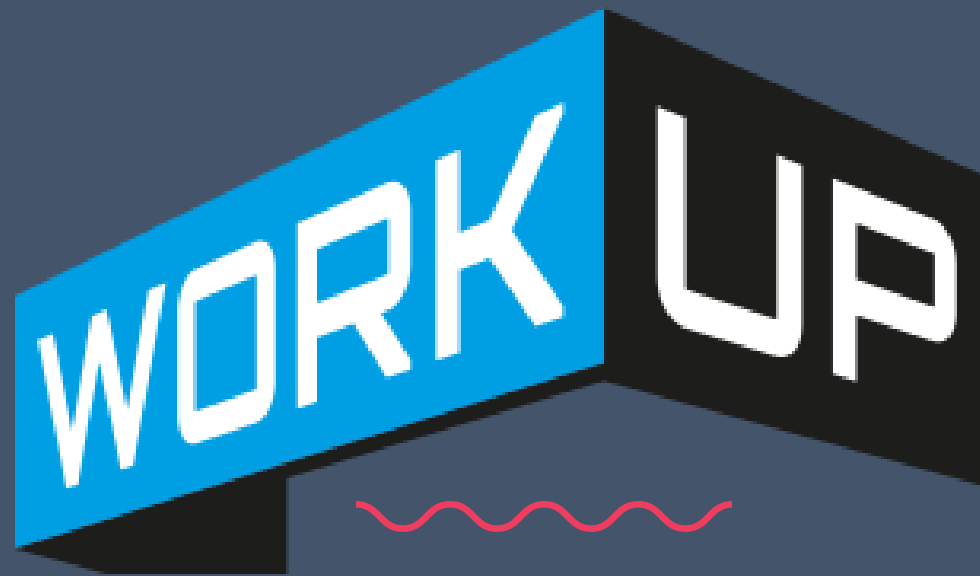
- Appears between ages 5 and 15
- Has slower progression than DMD
- Affects hips and pelvic area first
- Causes muscle weakness in the heart for most



Limb-Girdle

- Over 30 forms
- Starts in either adulthood or childhood
 - Creates muscle weakness and atrophy in hips and shoulders
 - LGMD's progression is dependent on the age of onset





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



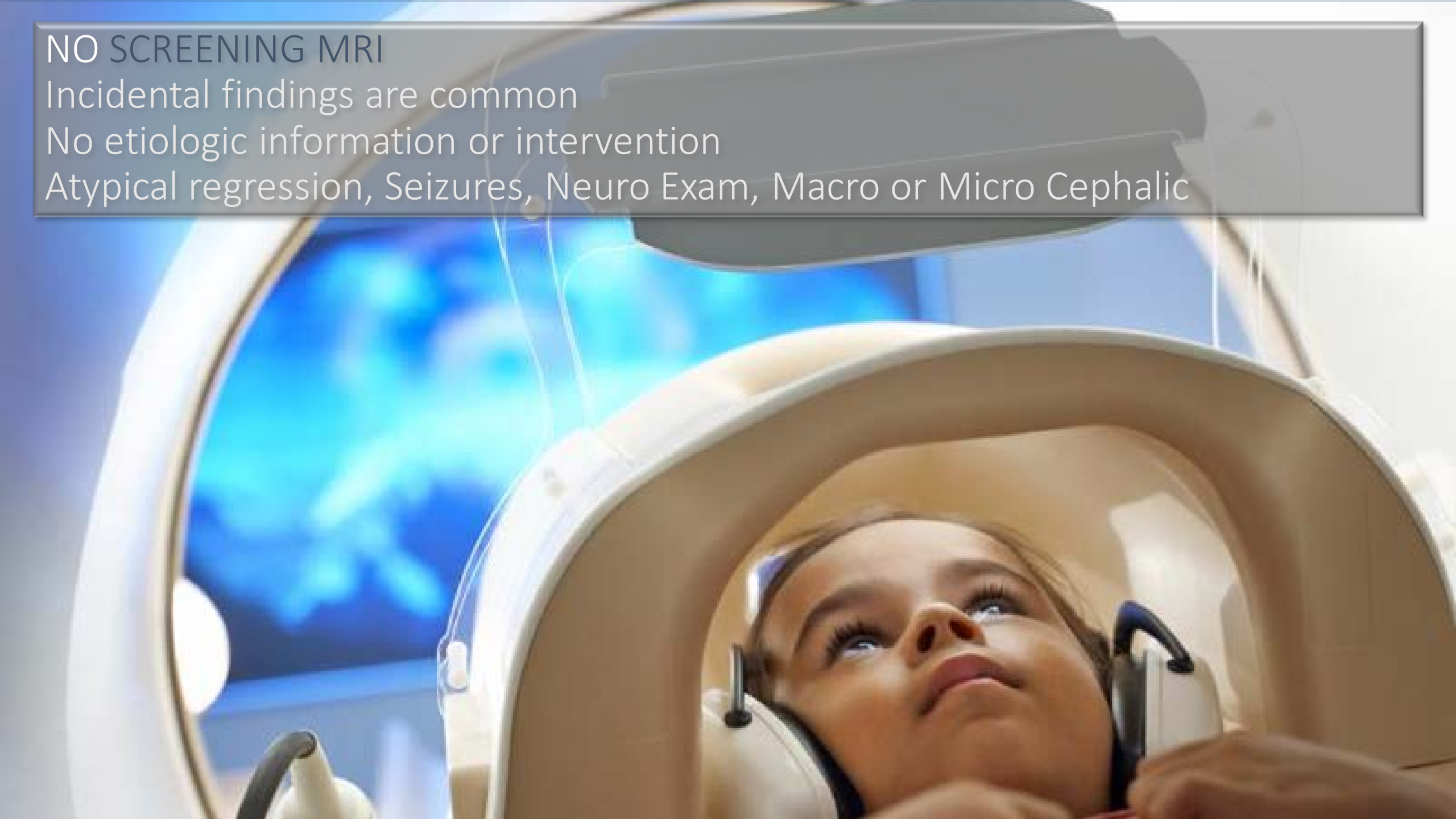
NO SCREENING EEG
Atypical Regression, History, or
Neuro Exam

NO SCREENING MRI

Incidental findings are common

No etiologic information or intervention

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



A photograph of a young child walking away from the camera on a dirt path. The child is wearing a bright blue hooded raincoat and grey shorts. A red wavy line is drawn horizontally across the child's chest area. The background is a blurred green landscape with trees and grass.

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

877.622.2884 naa@nationalautism.org f t

Donate Give Monthly

NATIONAL AUTISM ASSOCIATION

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RED DOT BOX SAFETY

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

OCCUPATIONAL THERAPY

Skills for Life

focusflorida.com

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.



TYPES OF ABA THERAPY

Natural Environment Training (NET)



NET focuses on practicing and teaching skills within the situations that they would naturally happen. In these situations, the therapist uses naturally occurring opportunities to help children learn.

The therapist might provide a coloring page but withhold the crayons until the child requests them, giving the child an empty cup and waiting for him/her to request juice, or playing a board game but withholding the dice or spinner until the child requests it are all examples of using NET.

Pivotal Response Training (PRT)



PRT uses the natural environment for teaching opportunities and consequences. PRT focuses on increasing motivation by adding items like having the child make choices/selections, taking turns, and providing reinforcement for attempts made.

More intense ABA therapy associated with optimal developmental outcome

Discrete Trial Training (DTT)

DTT teaches a skill by breaking it up into simplified, isolated tasks/steps. By breaking down tasks into short trials and using prompts, DTT increases the overall success rate of learning. DTT uses clear beginnings and ends to each trial with specific instructions and prompts.

The trials are short, permitting several teaching trials and a number of learning opportunities to occur. In addition, using one-to-one teaching allows for individualized programming.



Verbal Behavior (VB)

VB training uses a structured and one-on-one type of teaching format. This training works to teach language to children by creating and developing connections between a word and its meaning. The following are a list of VB terms that are typically implemented:



- **Echoics** occur when a speaker says something aloud and the listener repeats exactly what was said. For example, the therapist says, "Ball pit" and the child will repeat the same phrase, "Ball pit".
- **Mands** can be thought of as commands or demands, in which a person is commanding or demanding something. A mand typically results in the speaker obtaining the item that was spoken. For example, a child asking for a drink of water when he/she is thirsty and then receiving the drink.
- **Tacts** can be thought of as labeling an object. When a child sees a dog and then verbally says the word "Dog", he/she is emitting a tact.
- **Intraverbals** are similar to a conversation: a question is first asked and then an answer is provided. For example, the therapist asks, "How are you?" and the child responds, "Good!". Intraverbals can also involve filling in the blank. For example, the therapist says, "Twinkle twinkle little _____" and the child responds with "Star".



WHAT IS PARENT MANAGEMENT TRAINING (PMT)?



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



Deafness

An inability to comprehend verbal language due to a lack of hearing ability characterized by deafness



Assist students with text to speech software to manage and maximize their education

Provide students with hearing aids, cochlear implants, and sign language to help them understand

Developmental Delay

Children ages 0 through 9 with a developmental delay in physical, cognitive, social, emotional or adaptive development, etc...

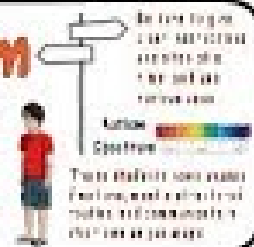


Children ages 0 through 9 with a developmental delay in physical, cognitive, social, emotional or adaptive development, etc...

Early intervention services such as speech and language therapy, occupational therapy, and physical therapy

Autism

A developmental disability significantly affecting verbal and nonverbal communication and social interaction, generally evident before age three



These students have unique challenges and a lot of strengths. They need support in social skills and communication.

Deaf-Blindness

Deaf-Blindness refers to a child with both hearing and visual disabilities evident before age three



Students need a lot of support to learn and communicate. They need support in social skills and communication.

Other students with both hearing and visual disabilities may have different needs and challenges.

Emotional Disturbed

Students that are unable to learn due to continuous behavioral problems



These students may have emotional, behavioral, and learning problems that affect their ability to learn.

Students need support in social skills and communication. They need support in social skills and communication.

Hearing Impairment

An impairment in hearing, whether permanent or fluctuating, and is not included under the definition of "deafness"



Students need support in social skills and communication. They need support in social skills and communication.

Students need support in social skills and communication. They need support in social skills and communication.

Intellectual Disability

Significantly below average general intelligence, existing with deficits in adaptive behavior



Students need support in social skills and communication. They need support in social skills and communication.

Students need support in social skills and communication. They need support in social skills and communication.

Multiple Disabilities

Students have impairments that include intellectual disability and blindness, occurring at the same time



Students need support in social skills and communication. They need support in social skills and communication.

Students need support in social skills and communication. They need support in social skills and communication.

Orthopedic Impairment

A physical impairment of the bones or muscles that adversely affects a child's educational performance



Students need support in social skills and communication. They need support in social skills and communication.

Students need support in social skills and communication. They need support in social skills and communication.

Other-Health Impairment

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

Students need support in social skills and communication. They need support in social skills and communication.



Students need support in social skills and communication. They need support in social skills and communication.

Students need support in social skills and communication. They need support in social skills and communication.

Traumatic Brain Injury

An acquired injury to the brain caused by an external physical force, resulting in total or partial functional disability



Students need support in social skills and communication. They need support in social skills and communication.

Students need support in social skills and communication. They need support in social skills and communication.

Specific Learning Disability

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



Students need support in social skills and communication. They need support in social skills and communication.

Students need support in social skills and communication. They need support in social skills and communication.

Speech Language Impairment

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



Students need support in social skills and communication. They need support in social skills and communication.

Students need support in social skills and communication. They need support in social skills and communication.

Visual Impairment

An impairment in vision, even with corrective lenses, which adversely affects a child's educational performance



Students need support in social skills and communication. They need support in social skills and communication.

Students need support in social skills and communication. They need support in social skills and communication.

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 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 DISABILITIES 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

RELEVANT 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 STUDENT'S TRANSITION 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



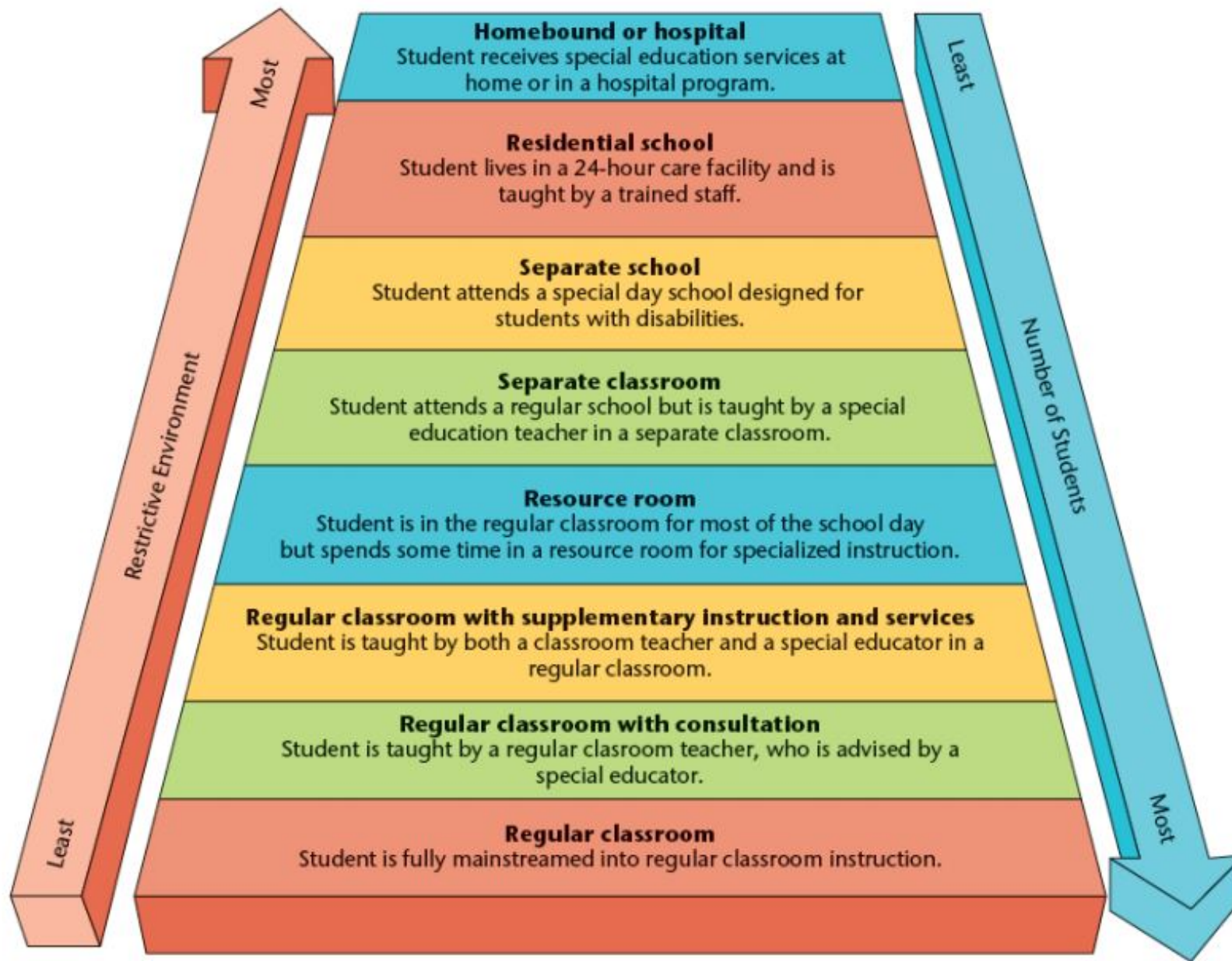
APPROPRIATE EVALUATIONS



SCHOOLS SHOULD USE A VARIETY OF ASSESSMENT METHODS IN THEIR EVALUATIONS



NEMOURS
CHILDREN'S HEALTH



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 Services IDEA Part C

What is an IFSP?



Early Intervention

Developmental delay $\geq$ 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

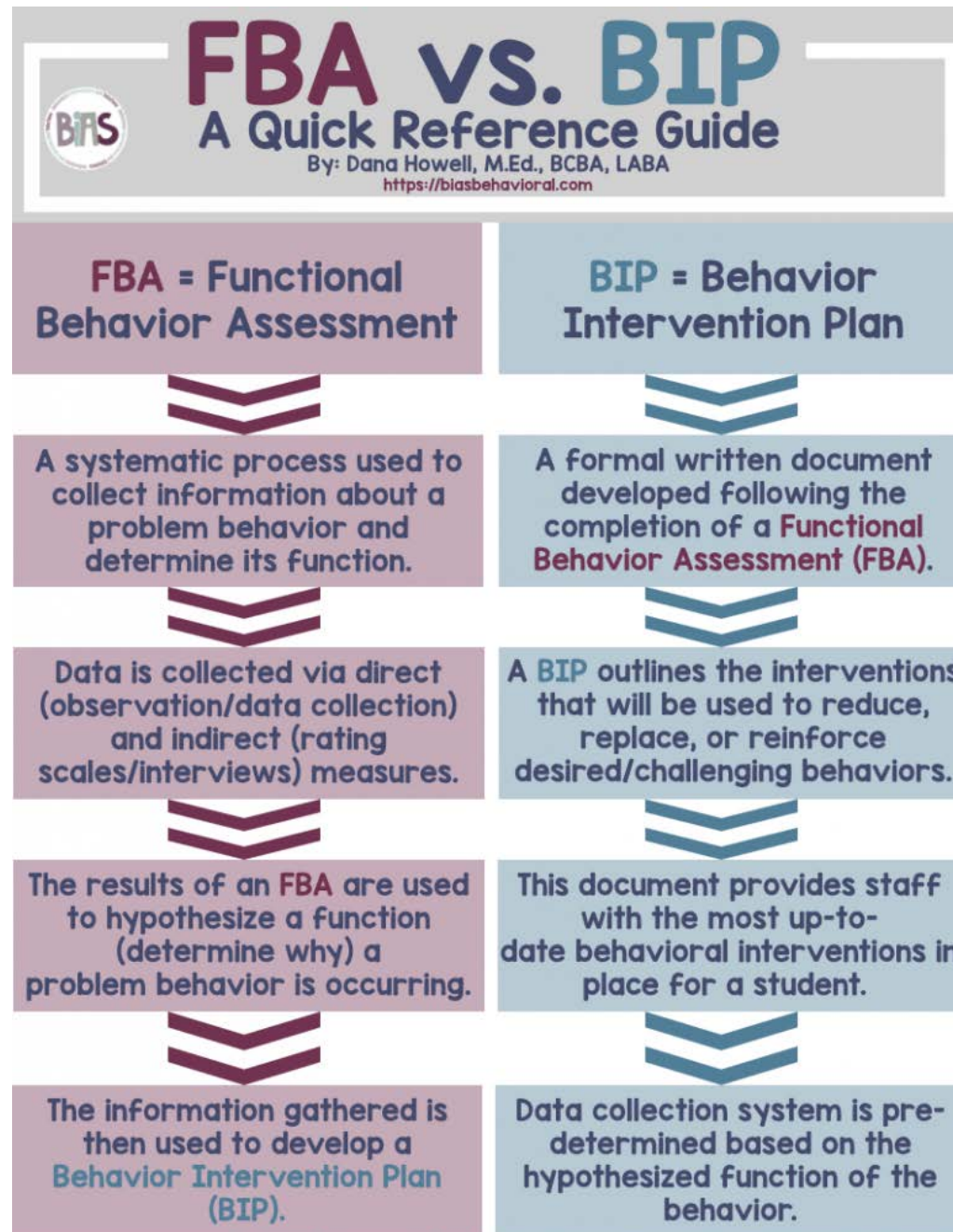
IFSP

- 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 image features decorative geometric shapes in the corners: a light blue diamond in the top-left, a yellow diamond in the top-right, and a light blue diamond in the bottom-center.

The End

The text "The End" is centered and rendered in a white, bold, sans-serif font. It is set against a vibrant background of colorful paint splashes. The splashes are primarily red, yellow, blue, and green, with some white splashes interspersed. The splashes are dynamic and energetic, with many small droplets and larger, more solid areas of color. The overall effect is one of a final, colorful burst.