April 23, 2026

Genetics and Speech-Language Pathology What does the future hold?

Knowledge is Power: Translating Genetic Insights into Proactive and Personalized Interventions

/beʔát^{h}ə/

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Disclosures

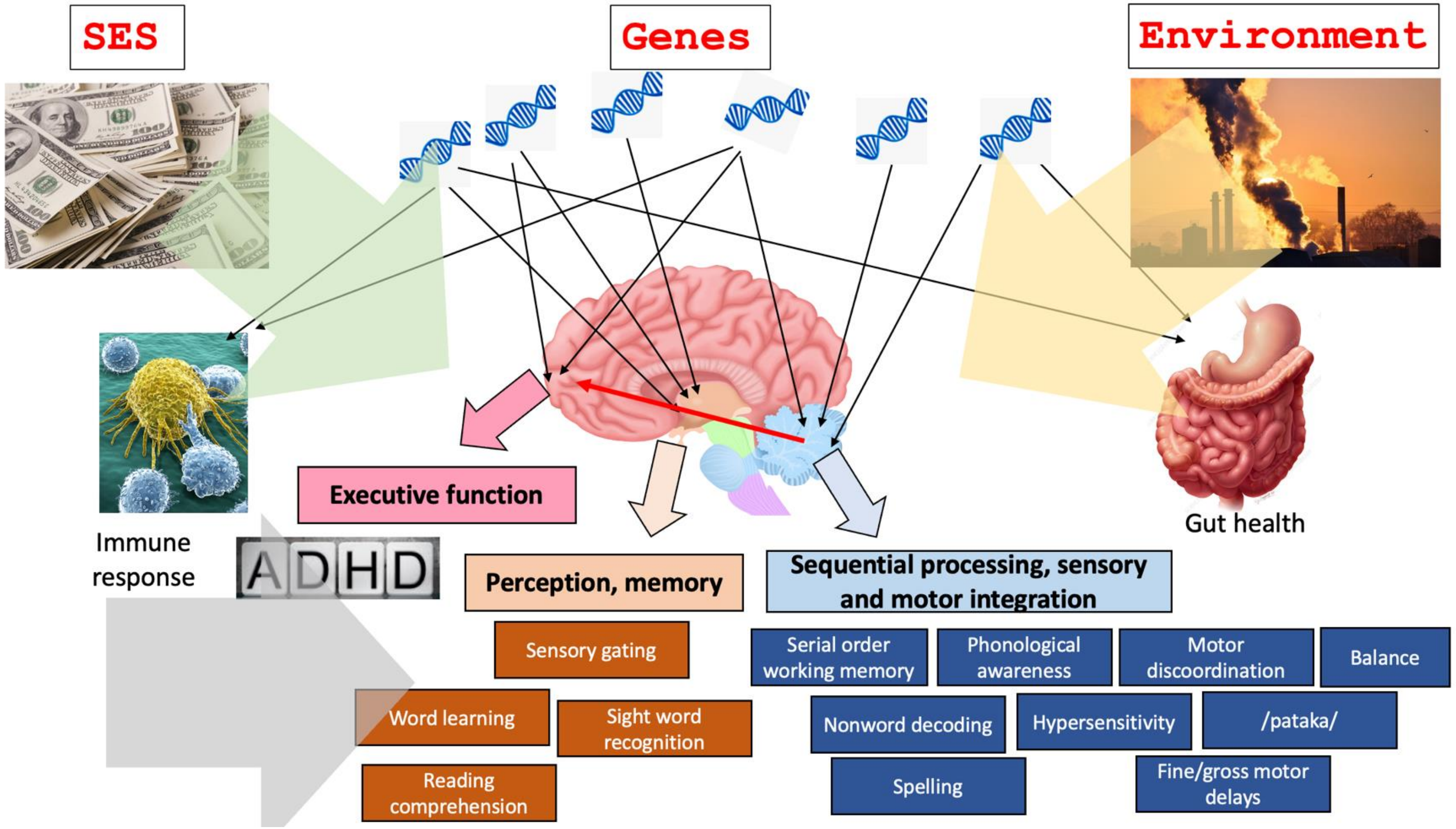
- No conflicts of interest
- Parts of this presentation were shown at the 2024 ASHA Convention Research Symposium “Genetics” (BP was the facilitator)

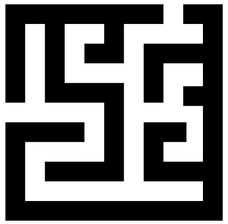
If a child has a speech disorder due to a genetic cause, we don’t treat that child because we can’t change the DNA.

School district representative
Personal communication, Dr. Barabara Lewis

Is DNA really this deterministic? Our fate?

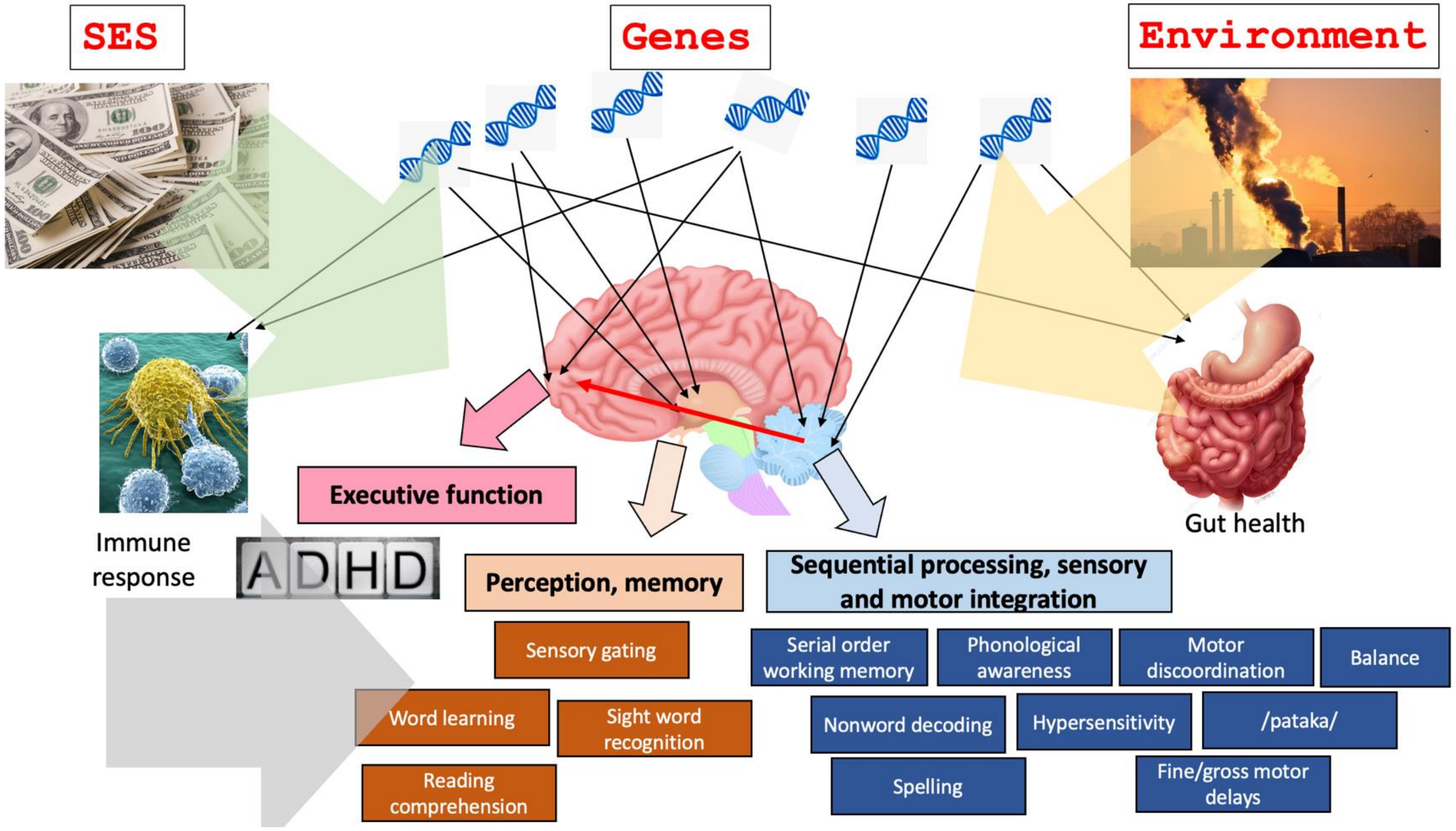
Biology of speech and language: A complicated picture

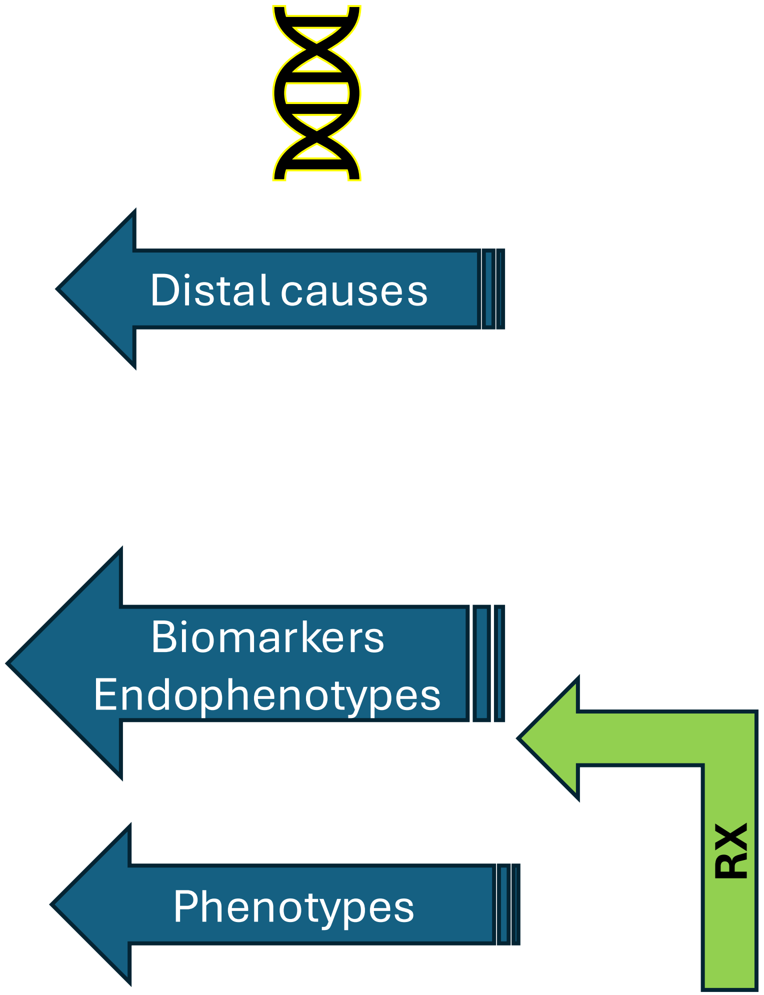




It's all wildly complicated! How will we ever use any of this in the clinic?

Goals:
1. Unraveling cause-effect relationships
2. Clinical translations





Personalized
Proactive

1. Unraveling cause-effect relationships

HUGE STRIDES!
Kudos to Professor Morgan!!!

Early neuroimaging markers of *FOXP2* intragenic deletion

Frédérique J. Liégeois[1,2], Michael S. Hildebrand[3,5], Alexandra Bonthrone[1,2], Samantha J. Turner[3,6], Ingrid E. Scheffer[3,4,5,8], Melanie Bahlo[3,7], Alan Connelly[3,4] & Angela T. Morgan[3,6,8]

nature portfolio

Mol Psychiatry. 2024 Feb 16;29(5):1281–1292. doi: 10.1038/s41380-024-02409-8

Genetic architecture of childhood speech disorder: a review

Angela T Morgan [1,2,3,✉], David J Amor [1,4], Miya D St John [1,2], Ingrid E Scheffer [1,5], Michael S Hildebrand [1,5]

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PMCID: PMC11189821 PMID: 38366112

PNAS RESEARCH ARTICLE | PSYCHOLOGICAL AND COGNITIVE SCIENCES GENETICS
OPEN ACCESS
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Genome-wide analyses of individual differences in quantitatively assessed reading- and language-related skills in up to 34,000 people

Else Eising[a], Nazanin Mirza-Schreiber[b], Eveline L. de Zeeuw[c], Carol A. Wang[d,e], Dongnhu T. Truong[f], Andrea G. Allegrini[g], Chin Yang Shapland[h,i], Gu Zhu[j], Karen G. Wigg[k], Margot L. Gerritse[a], Barbara Molz[a], Gökberk Alagöz[a], Alessandro Gialluisi[l,m,n], Filippo Abbondanza[o], Kaili Rimfeld[g,p], Marjolein van Donkelaar[a], Zhijie Liao (廖志洁)[q], Philip R. Jansen[r,s,t], Till F. M. Andlauer[l,u], Timothy C. Bates[v], Manon Bernard[w], Kirsten Blokland[x], Milene Bonte[y], Anders D. Børglum[z,aa,bb], Thomas Bourgeron[cc], Daniel Brandeis[dd,ee,ff,gg], Fabiola Ceroni[hh,ii], Valéria Csépe[jj,kk], Philip S. Dale[ll], Peter F. de Jong[mm], John C. DeFries[nn,oo], Jean-François Démonet[pp], Ditte Demontis[z,aa], Yu Feng[k], Scott D. Gordon[j], Sharon L. Guger[qq], Marianna E. Hayiou-Thomas[rr], Juan A. Hernández-Cabrera[ss], Jouke-Jan Hottenga[c], Charles Hulme[tt], Juha Kere[uu,vv], Elizabeth N. Kerr[qq,ww,xx], Tanner Koomar[yy], Karin Landerl[zz,aaa], Gabriel T. Leonard[bbb], Maureen W. Lovett[x,xx], Heikki Lyytinen[ccc], Nicholas G. Martin[j], Angela Martinelli[o], Urs Maurer[ddd], Jacob J. Michaelson[yy], Kristina Moll[eee], Anthony P. Monaco[fff], Angela T. Morgan[ggg,hhh,iii], Markus M. Nöthen[jjj], Zdenka Pausova[w,kkk], Craig E. Pennell[d,e,lll], Bruce F. Pennington[mmm], Kaitlyn M. Price[k,x,nnn], Veera M. Rajagopal[z,aa], Franck Ramus[ooo], Louis Richer[ppp], Nuala H. Simpson[qqq], Shelley D. Smith[rrr], Margaret J. Snowling[qqq,sss], John Stein[ttt], Lisa J. Strug[uuu,vvv], Joel B. Talcott[www], Henning Tiemeier[r,xxx], Marc P. van der Schroeff[yyy,zzz], Ellen Verhoef[a], Kate E. Watkins[qqq], Margaret Wilkinson[x], Margaret J. Wright[aaaa], Cathy L. Barr[k,x,nnn], Dorret I. Boomsma[c,bbbb,cccc], Manuel Carreiras[dddd,eeee,ffff], Marie-Christine J. Franken[yyy], Jeffrey R. Gruen[f], Michelle Luciano[v], Bertram Müller-Myhsok[l,gggg], Dianne F. Newbury[ii], Richard K. Olson[nn], Silvia Paracchini[o], Tomáš Paus[hhhh], Robert Plomin[g], Sheena Reilly[ggg,iiii], Gerd Schulte-Körne[eee], J. Bruce Tomblin[iiii], Elsje van Bergen[c,bbbb,kkkk], Andrew J. O. Whitehouse[llll], Erik G. Willcutt[oo], Beate St Pourcain[a,h,mmmm,1], Clyde Francks[a,mmmm,nnnn,1], and Simon E. Fisher[a,mmmm,2]

Molecular Psychiatry
https://doi.org/10.1038/s41380-018-0020-x

ARTICLE

Check for updates

A set of regulatory genes co-expressed in embryonic human brain is implicated in disrupted speech development

Else Eising[1] · Amaia Carrion-Castillo[1] · Arianna Vino[1] · Edythe A. Strand[2] · Kathy J. Jakielski[3] · Thomas S. Scerri[4,5] · Michael S. Hildebrand[6] · Richard Webster[7] · Alan Ma[8] · Bernard Mazoyer[9] · Clyde Francks[1,10] · Melanie Bahlo[4,5] · Ingrid E. Scheffer[6,11] · Angela T. Morgan[6,12] · Lawrence D. Shriberg[13] · Simon E. Fisher[1,10]

Neurosci Biobehav Rev. 2023 Sep:152:105293. doi: 10.1016/j.neubiorev.2023.105293. Epub 2023 Jun 22.

To speak may draw on epigenetic writing and reading: Unravelling the complexity of speech and language outcomes across chromatin-related neurodevelopmental disorders

Miya St John [1], Tanya Tripathi [2], Angela T Morgan [3], David J Amor [4]

2. Clinical translations

Precision medicine (generally considered analogous to personalized medicine or individualized medicine) is an innovative approach that uses information about an individual's genomic, environmental and lifestyle information to guide decisions related to their medical management. The goal of precision medicine is to provide more a precise approach for the prevention, diagnosis and treatment of disease.

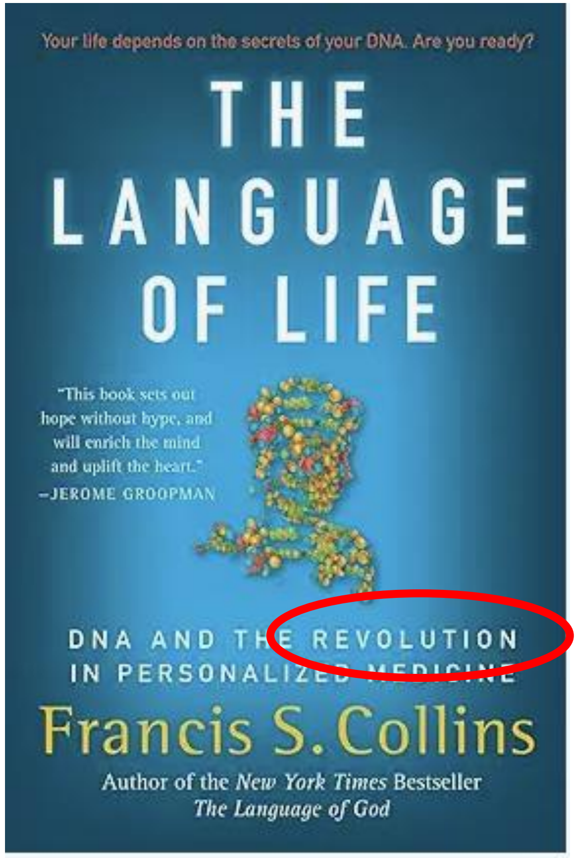
There is no such thing as the "average patient"

https://www.genome.gov/genetics-glossary/Precision-Medicine#:~:text=Definition&text=Precision%20medicine%20(generally%20considered%20analogous,diagnosis%20and%20treatment%20of%20disease.

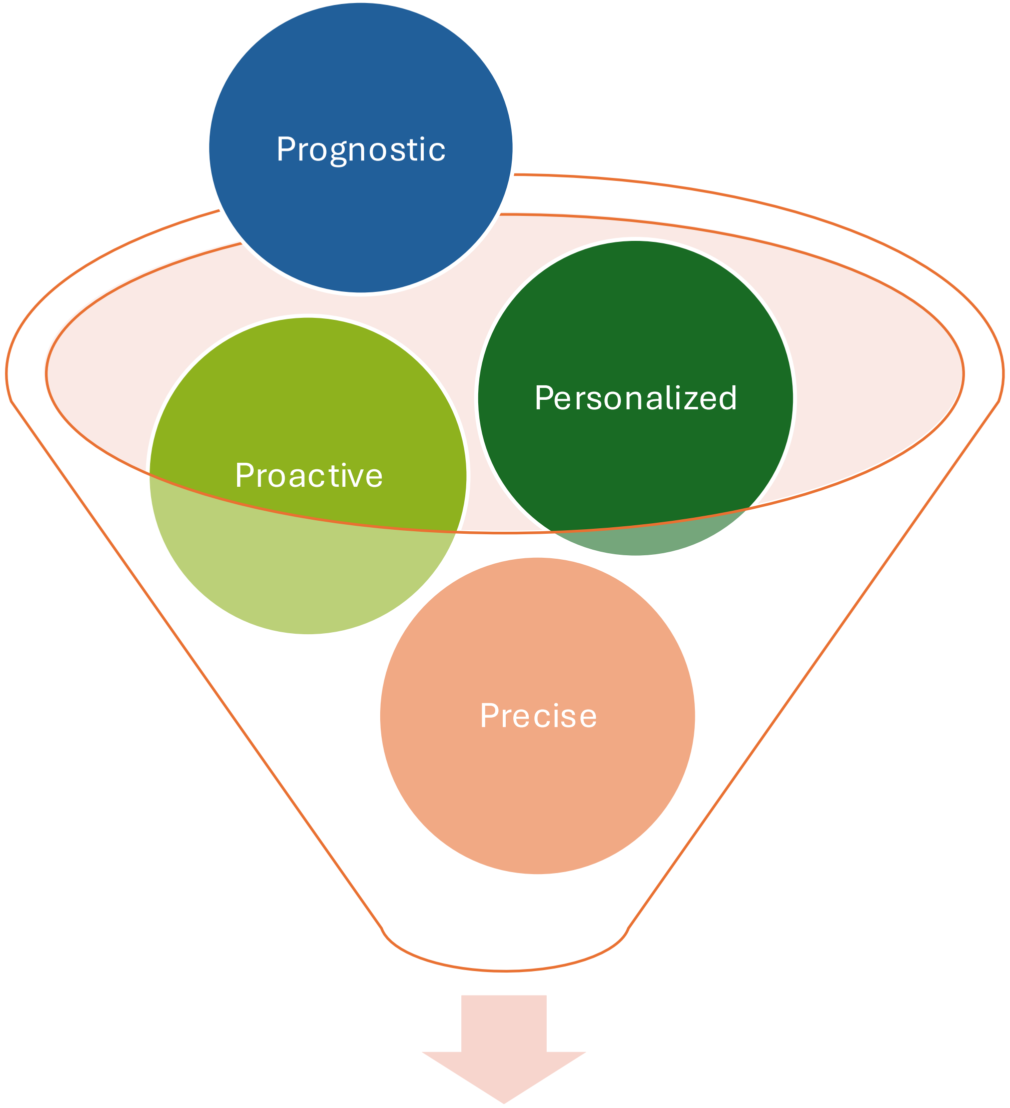
Francis Collins, 2010, About Consumer DNA Services

Francis S. Collins, M.D., Ph.D.

Director, National Institutes of Health, August 17, 2009 - December 19, 2021



- …recent discoveries place us in a position to make several strong statements: (1) for each disease, specific genetic and environmental risk factors exist, and are rapidly being identified; (2) these discoveries are providing powerful new insights into both treatment and prevention; (3) the more you know about all this, the more you can adjust your own lifestyle and medical surveillance to prevent illnesses or catch them in early and treatable stages (p. 61)
- The right drug at the right dose for the right person (p. 231 ff)



Improved Outcomes

Each person is unique!

- Genetic profile
- Environment
- Epigenetic profile
- Lifestyle
- Nutrition
- ...

Some examples:

Pharmacogenomics:
Selecting the right type and dosage of blood thinners and statins based on the patient's genotype

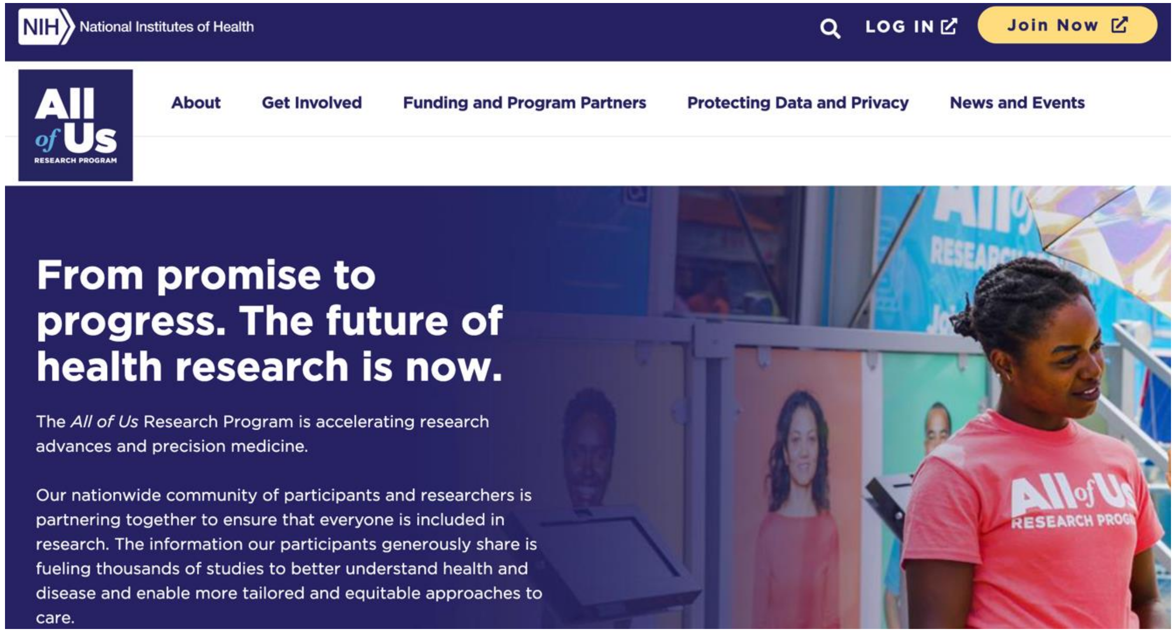
Precision nutrition:
Managing diet-related chronic illnesses with diet changes and supplements

Precision Oncology:
Genotyping the cancer cells, then selecting the most effective therapy for that cell type

Exposome:
Zip code, how the environment interacts with DNA and health

Adapted from https://www.nih.gov/about-nih/what-we-do/nih-turning-discovery-into-health/promise-precision-medicine

https://obamawhitehouse.archives.gov/precision-medicine



Goals:

1. *Enroll 1 million people*
2. *Collect surveys, health data streams, a whole genome sequence, environmental data, and physical measures.*
3. *Launch partnered research studies*
4. *Global community of >100,000 researchers*
5. *Participant return of value*
6. *Management and organization*

The NEW ENGLAND JOURNAL of MEDICINE

MEDICINE, writ large

SPECIAL REPORT

The "All of Us" Research Program

Author: The All of Us Research Program Investigators Author Info & Affiliations

Published August 14, 2019 | N Engl J Med 2019;381:668-676 | DOI: 10.1056/NEJMsr1809937
VOL. 381 NO. 7 | Copyright © 2019

Well, what about speech and language disorders?

1-10%

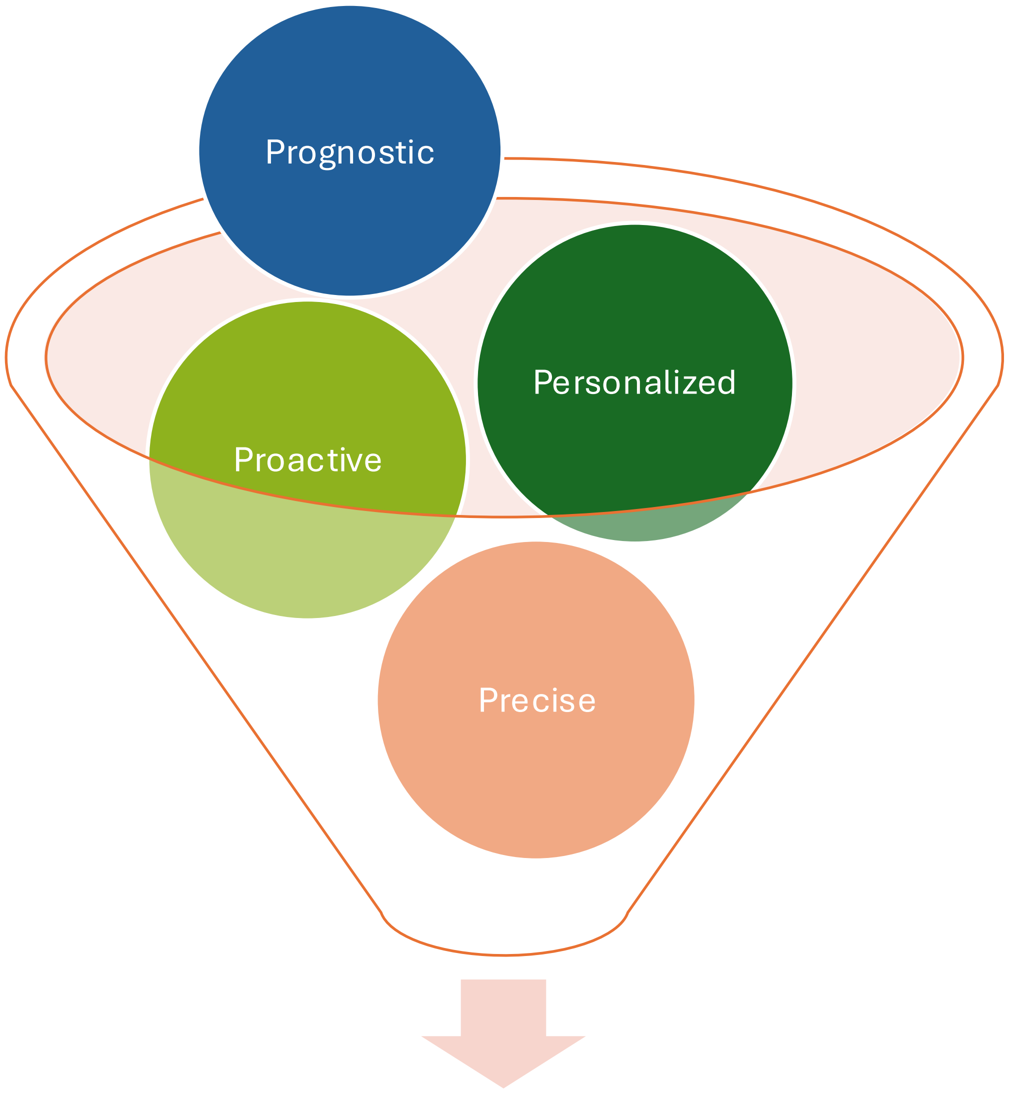
Table S1: Estimated baseline prevalences and incidences of disease conditions in one million people.

These data are taken from the Precision Medicine Initiative (PMI) Working Group Report to the Advisory Committee to the Director and were derived from individuals volunteering to large-scale EHR biobanks in the NHGRI Electronic Medical Records and Genomics (eMERGE) Network.[1] As these case estimates derived from longitudinally followed patients at large academic healthcare systems, they may represent sicker populations than enrolling in *All of Us*.

Disease	Estimated prevalent cases	Estimated incident cases 5 years	10 years	Total in 10 years
Essential hypertension	360,794 36%	99,963	231,165	591,959
Type 2 Diabetes	135,658 14%	40,411	94,493	230,151
Depression	110,091 11%	40,133	91,893	201,984
Atrial fibrillation	78,272 8%	35,047	56,292	134,564
Chronic renal failure	74,700	28,944	49,926	124,626
Congestive heart failure	73,723	21,315	40,322	114,045
Asthma	62,149	17,292	44,036	106,185
COPD	48,728	15,396	33,584	82,312
Rheumatoid arthritis	45,835	11,466	23,875	69,710
Myocardial infarction	39,273	14,981	27,112	66,385
Epilepsy	33,426	4,161	11,248	44,674
Thrombosis	26,746	11,559	21,169	47,915
Breast cancer (female)	20,470	12,068	21,382	41,852
Stroke	16,016	8,969	15,598	31,614
Lupus	14,659	3,283	6,738	21,397
Prostate cancer	13,861	6,241	13,848	27,709
Dementia	13,373	7,028	9,656	23,029
Lung cancer	11,432 1%	2,866	4,828	16,260
Colorectal cancer	9,407	3,745	6,844	16,251
Abdominal aortic aneurysms	8,729	2,451	5,518	14,247
Melanoma	6,109 0.6%	2,727	3,873	9,982
Parkinson's disease	4,311 0.4%	2,127	4,032	8,343

From medical to clinical applications of precision medicine principles

Disorder	%	Age	Reference
Reading disorder (dyslexia)	7-10	General population	Thomas, T, Khalaf, S., Grigorenko, E. A systematic review and meta-analysis of imaging genetics studies of specific reading disorder. *Cognitive Neuropsychology*, 2021-04, Vol.38 (3), p.179-204. Peterson RL, Pennington BF. Developmental dyslexia. Lancet 2012; 379:1997–2007.
Speech Sound Disorder	5	Early school age	Shriberg, LD; Tomblin, JB; McSweeny, JL (1999): Prevalence of speech delay in 6-year-old children and comorbidity with language impairment. J Speech Lang Hear Res. Dec;42(6):1461-81. doi: 10.1044/jslhr.4206.1461.
Developmental Language Disorder	3.3	Children, adolescents	Black, LI; Vahratian, A; Hoffman, HJ (2015): Communication disorders and use of intervention services among children aged 3–17 years: United States, 2012. NCHS data brief. Hyattsville, MD: National Center for Health Statistics. Jun:(205):1-8
Stuttering	1	Lifespan	Yairi, E; Ambrose, N (2013): Epidemiology of stuttering: 21st century advances. J Fluency Disord. Jun;38(2):66-87. doi: 10.1016/j.jfludis.2012.11.002.



Improved Outcomes

But hey, we are already doing that:
- Select interventions for different types of speech sound disorder
- Recommend the most effective intervention dosage
- …
- Uh … what about ”prognostic” and “proactive”????

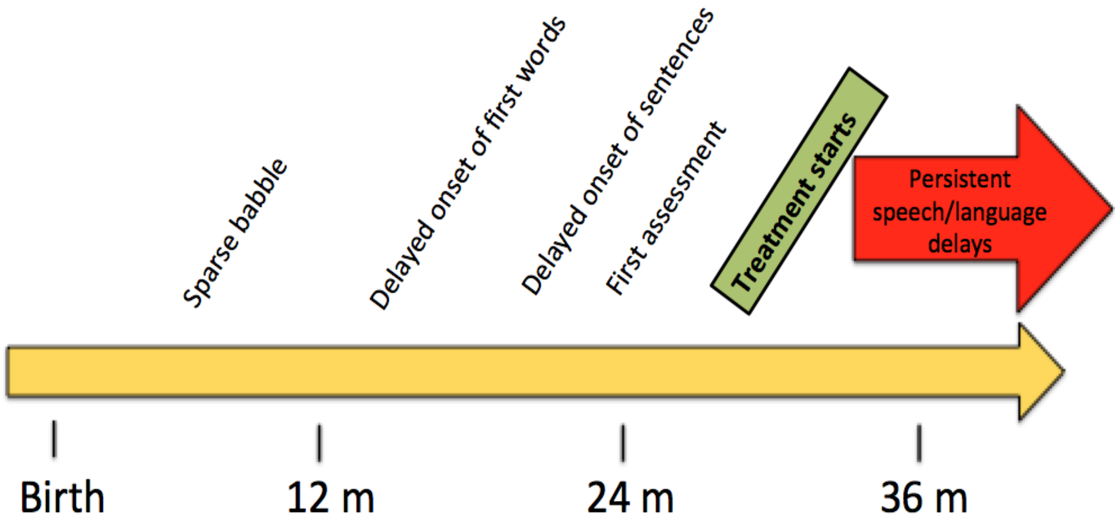
Imagine a world where …

We could know children‘s genetic risk for speech/language/fluency etc. at birth

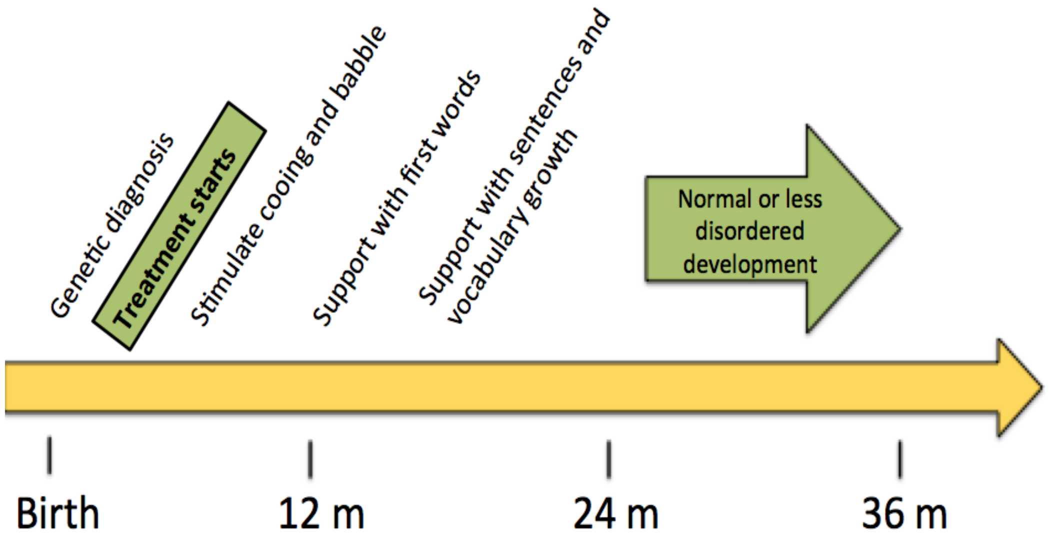
We could develop earliest preventive approaches for them

We could taylor clinical management based on the patient‘s genetic profile

… kind of like ***precision medicine***



Current status:
"Wait until you see signs of speech or language problems."



Imagine that disorder risks could be predicted at birth and addressed with prevention …

Proof-of-concept study:

Classic Galactosemia

- Metabolic disease
- Diagnosed via newborn screening
- High risk for speech and language disorders
- Rare in the US: 1:~40,000 births

Why classic galactosemia?

Trait	General Population	Classic Galactosemia
Speech sound disorder	4-6%	40-85%
Childhood Apraxia of Speech	<1%	24-63%
Language delay	7-10%	50-78 (more expressive than receptive)

Recommended Uniform Screening Panel
Core Conditions
(As of January 2023)

X: Condition is in this category --: Condition is not in this category

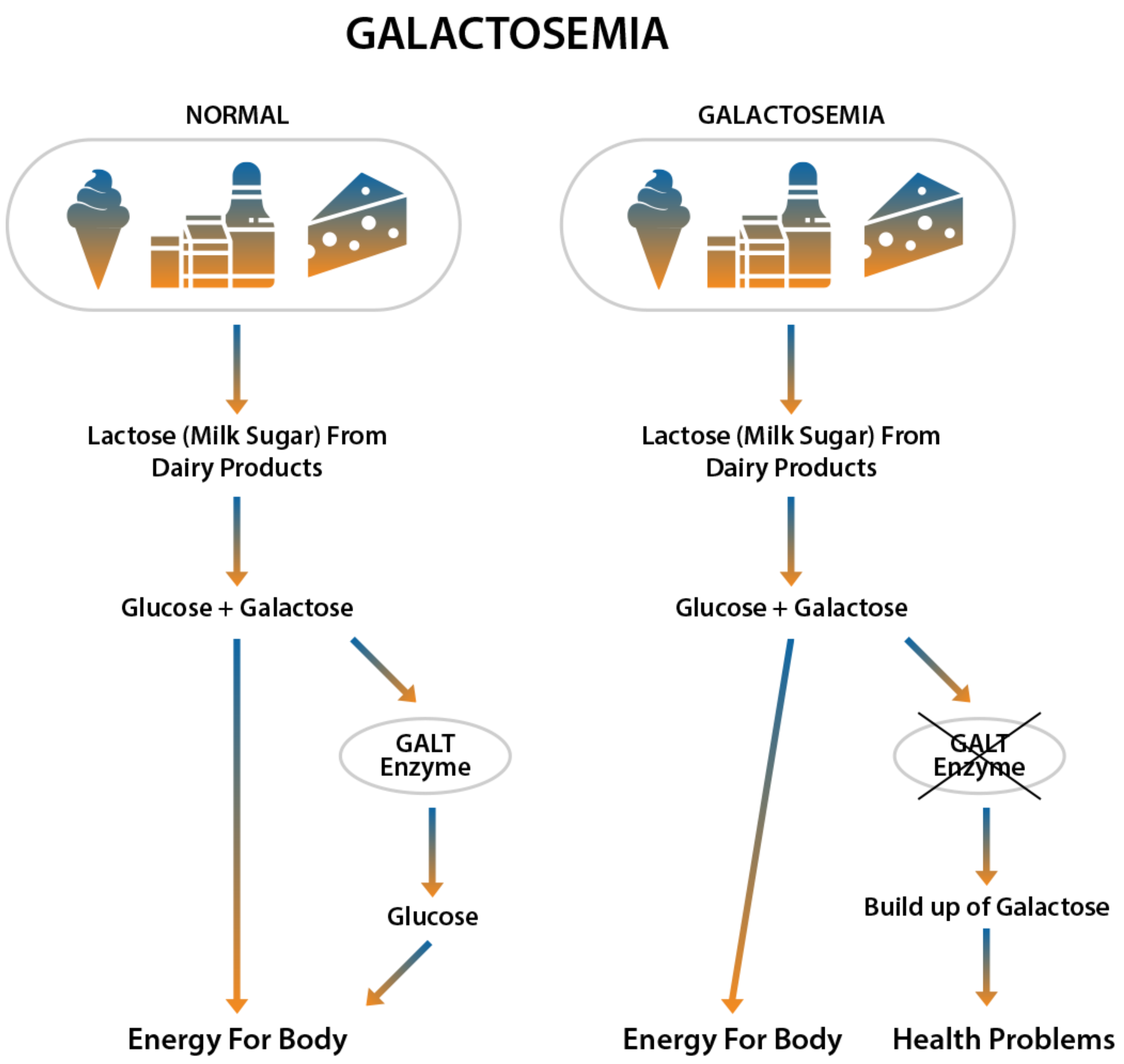
Core Condition	Metabolic Disorder - Organic acid condition	Metabolic Disorder - Fatty acid oxidation disorder	Metabolic Disorder - Amino acid disorder	Endocrine Disorder	Hemoglobin Disorder	Other Disorder
3-Hydroxy-3-Methyglutaric Aciduria	X	--	--	--	--	--
3-Methylcrotonyl-CoA Carboxylase Deficiency	X	--	--	--	--	--
ß-Ketothiolase Deficiency	X	--	--	--	--	--
Glutaric Acidemia Type I	X	--	--	--	--	--
Holocarboxylase Synthase Deficiency	X	--	--	--	--	--
Isovaleric Acidemia	X	--	--	--	--	--
Methylmalonic Acidemia (Cobalamin disorders)	X	--	--	--	--	--
Methylmalonic Acidemia (methylmalonyl-CoA mutase)	X	--	--	--	--	--
Propionic Acidemia	X	--	--	--	--	--
Carnitine Uptake Defect/Carnitine Transport Defect	--	X	--	--	--	--
Long-chain L-3 Hydroxyacyl-CoA Dehydrogenase Deficiency	--	X	--	--	--	--
Medium-chain Acyl-CoA Dehydrogenase Deficiency	--	X	--	--	--	--
Trifunctional Protein Deficiency	--	X	--	--	--	--
Very Long-chain Acyl-CoA Dehydrogenase Deficiency	--	X	--	--	--	--
Argininosuccinic Aciduria	--	--	X	--	--	--
Citrullinemia, Type I	--	--	X	--	--	--
Classic Phenylketonuria	--	--	X	--	--	--
Homocystinuria	--	--	X	--	--	--
Maple Syrup Urine Disease	--	--	X	--	--	--
Tyrosinemia, Type I	--	--	X	--	--	--
Congenital adrenal hyperplasia	--	--	--	X	--	--
Primary Congenital Hypothyroidism	--	--	--	X	--	--
S, βeta-Thalassemia	--	--	--	--	X	--
S,C Disease	--	--	--	--	X	--
S,S Disease (Sickle Cell Anemia)	--	--	--	--	X	--
Biotinidase Deficiency	--	--	--	--	--	X
Classic Galactosemia	--	--	--	--	--	X
Critical Congenital Heart Disease	--	--	--	--	--	X
Cystic Fibrosis	--	--	--	--	--	X
Glycogen Storage Disease Type II (Pompe)	--	--	--	--	--	X
Guanidinoacetate Methyltransferase Deficiency	--	--	--	--	--	X
Hearing Loss	--	--	--	--	--	X
Mucopolysaccharidosis Type I	--	--	--	--	--	X
Mucopolysaccharidosis Type II	--	--	--	--	--	X
Severe Combined Immunodeficiencies	--	--	--	--	--	X
Spinal Muscular Atrophy due to homozygous deletion of exon 7 in SMN1	--	--	--	--	--	X
X-linked Adrenoleukodystrophy	--	--	--	--	--	X

Recommended Uniform Screening Panel[1]
SECONDARY[2] CONDITIONS[3]
(As of January 2023)

Secondary Condition	Metabolic Disorder - Organic acid condition	Metabolic Disorder - Fatty acid oxidation disorder	Metabolic Disorder - Amino acid disorder	Endocrine Disorder	Hemoglobin Disorder	Other Disorder
2-Methyl-3-hydroxybutyric aciduria	X	--	--	--	--	--
2-Methylbutyrylglycinuria	X	--	--	--	--	--
3-Methylglutaconic aciduria	X	--	--	--	--	--
Isobutyrylglycinuria	X	--	--	--	--	--
Malonic acidemia	X	--	--	--	--	--
Methylmalonic acidemia with homocystinuria	X	--	--	--	--	--
2,4 Dienoyl-CoA reductase deficiency	--	X	--	--	--	--
Carnitine acylcarnitine translocase deficiency	--	X	--	--	--	--
Carnitine palmitoyltransferase type I deficiency	--	X	--	--	--	--
Carnitine palmitoyltransferase type II deficiency	--	X	--	--	--	--
Glutaric acidemia type II	--	X	--	--	--	--
Medium/short-chain L-3-hydroxyacyl-CoA dehydrogenase deficiency	--	X	--	--	--	--
Medium-chain ketoacyl-CoA thiolase deficiency	--	X	--	--	--	--
Short-chain acyl-CoA dehydrogenase deficiency	--	X	--	--	--	--
Argininemia	--	--	X	--	--	--
Benign hyperphenylalaninemia	--	--	X	--	--	--
Biopterin defect in cofactor biosynthesis	--	--	X	--	--	--
Biopterin defect in cofactor regeneration	--	--	X	--	--	--
Citrullinemia, type II	--	--	X	--	--	--
Hypermethioninemia	--	--	X	--	--	--
Tyrosinemia, type II	--	--	X	--	--	--
Tyrosinemia, type III	--	--	X	--	--	--
Various other hemoglobinopathies	--	--	--	--	X	--
Galactoepimerase deficiency	--	--	--	--	--	X
Galactokinase deficiency	--	--	--	--	--	X
T-cell related lymphocyte deficiencies	--	--	--	--	--	X

1. Selection of conditions based upon "Newborn Screening: Towards a Uniform Screening Panel and System." *Genetic Med.* 2006; 8(5) Suppl: S12-S252" as authored by the American College of Medical Genetics (ACMG) and commissioned by the Health Resources and Services Administration (HRSA).
2. Disorders that can be detected in the differential diagnosis of a core disorder.
3. Nomenclature for Conditions based upon "Naming and Counting Disorders (Conditions) Included in Newborn Screening Panels." *Pediatrics.* 2006; 117 (5) Suppl: S308-S314.

https://www.hrsa.gov/sites/default/files/hrsa/advisory-committees/heritable-disorders/rusp/rusp-august-2023.pdf



Classic galactosemia is

- Diagnosed via newborn screening
- Treated with lactose-restricted diet
- But despite this restricted diet:
  - ***Severe speech and language disorders***
  - ***Fine and gross motor disorders***

https://www.andersondiagnostics.com/newbornscreening/galactosemia/

You’d think these babies get earliest attention but …

Our FB survey of 22 families with kids with CG found:

- **Mean age of first screening or assessment was 22 months** (SD = 15.5; range [3, 72])
- **Mean age at start of therapy was 28 months** (SD = 21.5; range [4, 96])
- **Only 5 had typical babble behaviors**

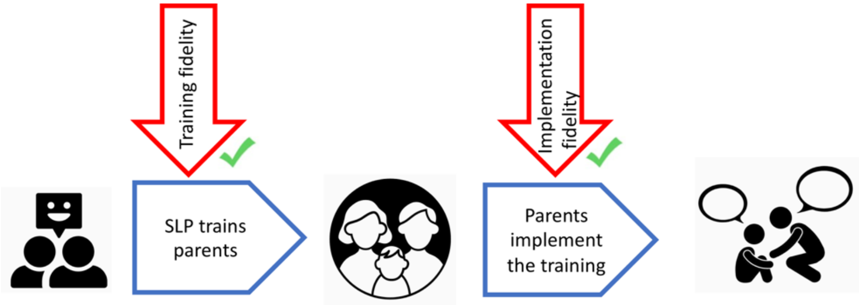
We asked ourselves:

- Babies with CG tend to ***not vocalize or babble*** much … Will teaching them to babble help their speech development?
- ***First words and sentences appear late*** and are underdeveloped … Will attention to words and sentences help communication outcomes in the long run?
- Besides the risk for speech and language disorders, there is also a risk ***for cognitive delays*** … Will a focus on communication skills also help cognitive development?

The big idea:

Babble Boot Camp©

Parent training via telehealth



A speech-language pathologist trains parents to

- Stimulate coo and babble
- Increase consonant and vowel inventory
- Elicit first words on time
- Shape early sentences
- Use language to convey meaning
- ...

https://news.asu.edu/20190514-study-aims-prevent-children-speech-and-language-disorders

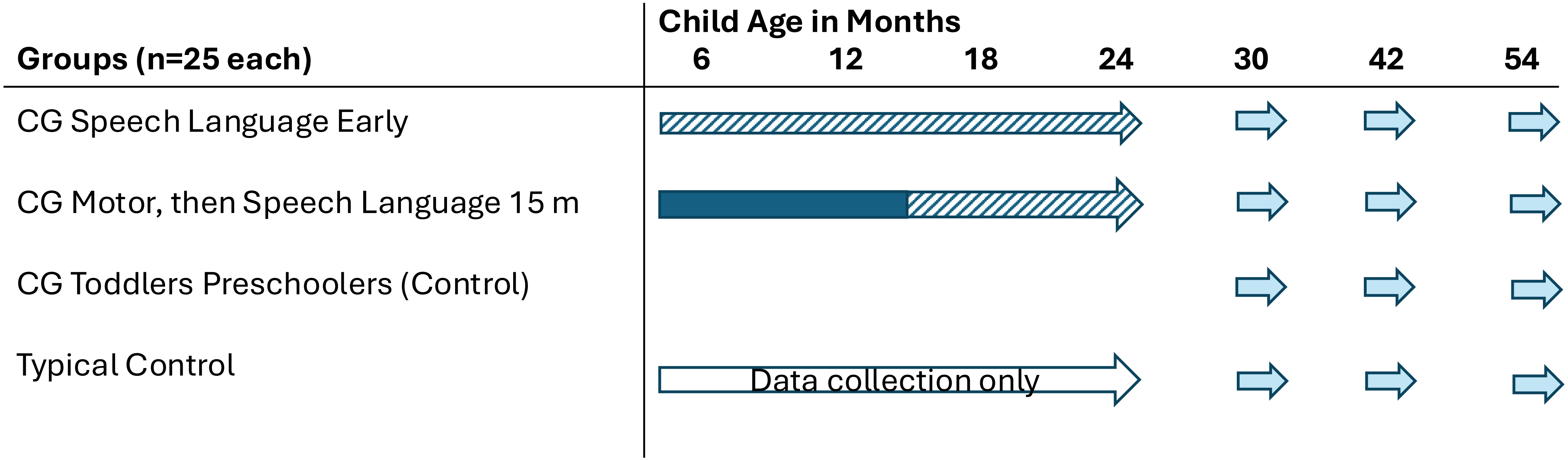
Active phase: infant age 2 – 24 months
Follow-up testing at child age 3 and 4 years

Note:
We start with prelinguistic skills!

Huge dosage b/c implemented daily at home

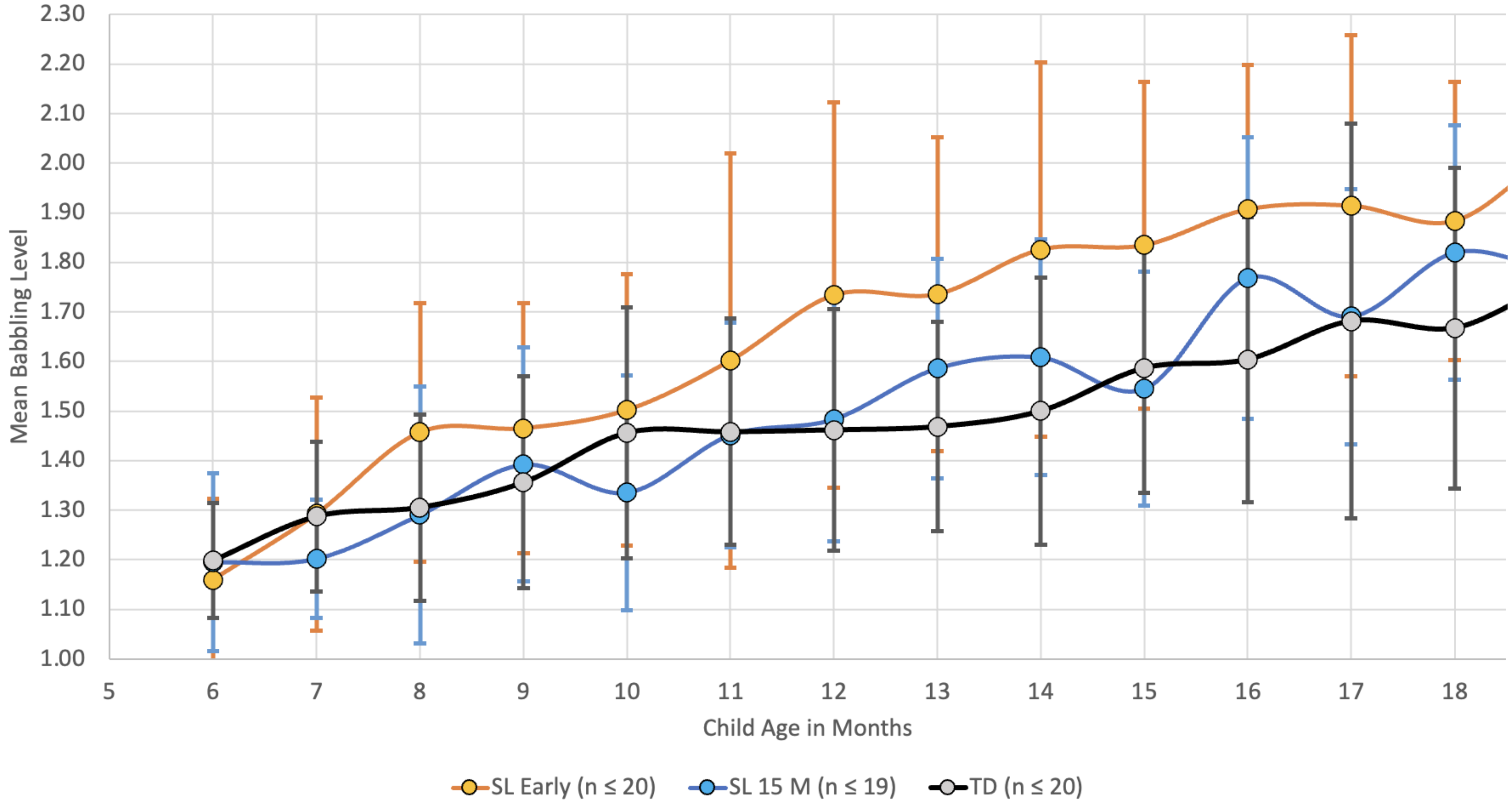
Birth …

nt Behavior	Target Child Behavior	Target Chil
"Word bath" using infant-directed prosody	Attend to the input	Perceptual coupling of visual and auditory input Building phoneme boundaries
Facial expression of emotions	Attend to the input	Communicative awareness
Respond to infant vocalizations	Increase vocalization rate	Sensorimotor association of movement and auditory percept Turn-taking
Model and respond to babble	Increase babble quantity and complexity	Motor speech control during prespeech sound production
Say words for persons and objects and narrate events	Imitate words Request more verbal labels by pointing or vocalizing	Receptive and expressive vocabulary Speech sound production Phonetic and phonemic representation
Recast child utterances with slight expansions	Produce more complex utterances	Syntactic complexity



Peter, B., Davis, J., Cotter, S., Belter, A., Williams, E., Stumpf, M., Bruce, L., Eng, L., Kim, Y., Finestack, L., Stoel-Gammon, C., Williams, D., Scherer, N., VanDam, M., & Potter, N. (2021) Towards preventing speech and language disorders of known genetic origin: First post-intervention results of Babble Boot Camp© in children with classic galactosemia. *American Journal of Speech-Language Pathology.* https://doi.org/10.1044/2021_AJSLP-21-00098

Mean Babbling Level: Phonotactics

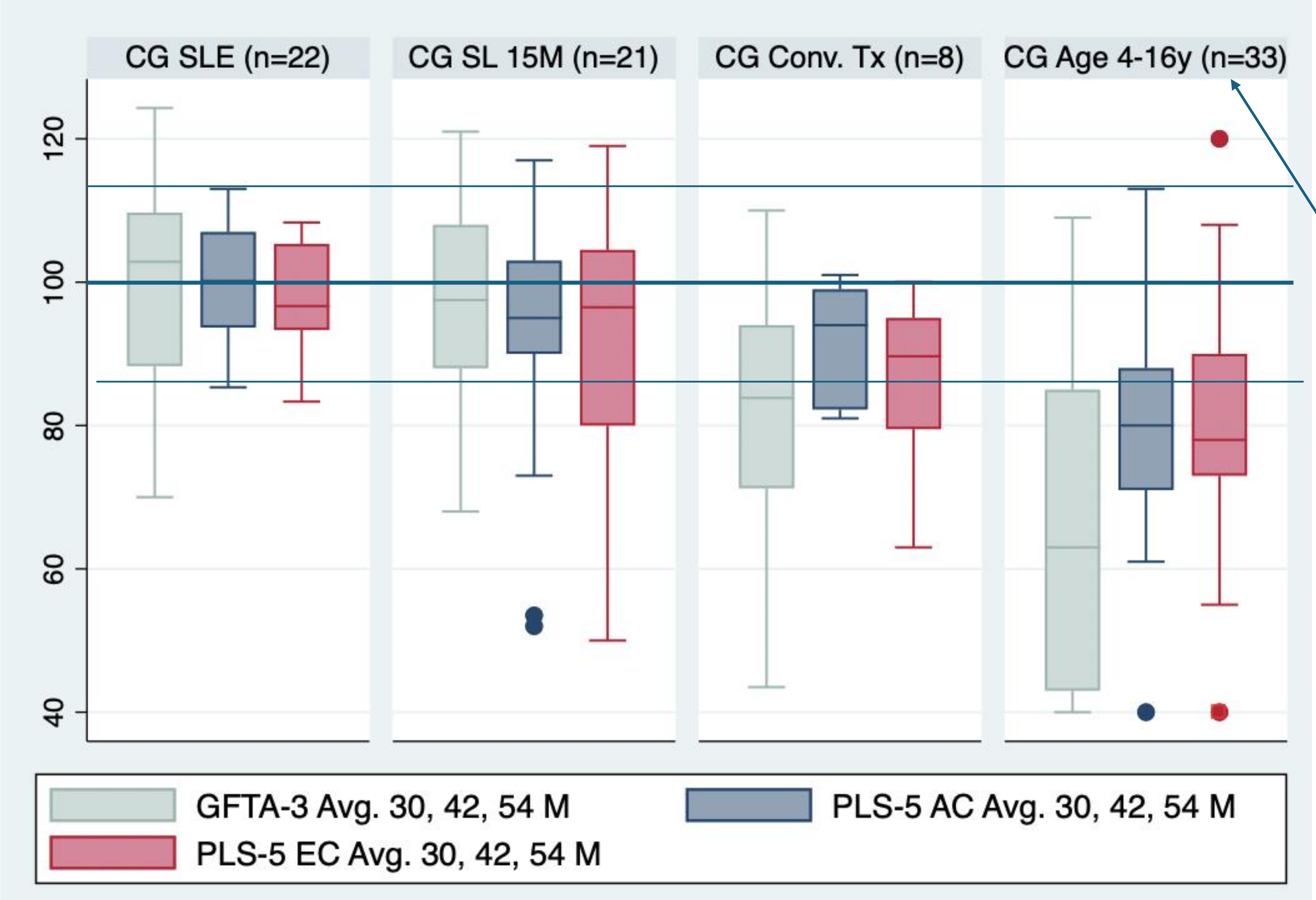


1 = vowel glide, glottal stop, [h] [a], [ha], [joja]

2 = vowel plus 1 true consonant [ba], [bopa]

3 = vowel plus 2+ true consonants [baga], [dapa]

Peter, B., Davis, J., Bruce, L., Finestack, L., Kornelis, M., Eng, L., Kim, Y., Scherer, N., Potter, N., VanDam, M., Thompson, L., Loveall, S., Stoel-Gammon, C., Raaz, C., Armstrong-Heimsoth, A. & Buckley, S. (2025). Translating the power of precision medicine into the world of communication disorders. *Journal of Speech, Language & Hearing Research, 11*(5174-5189)



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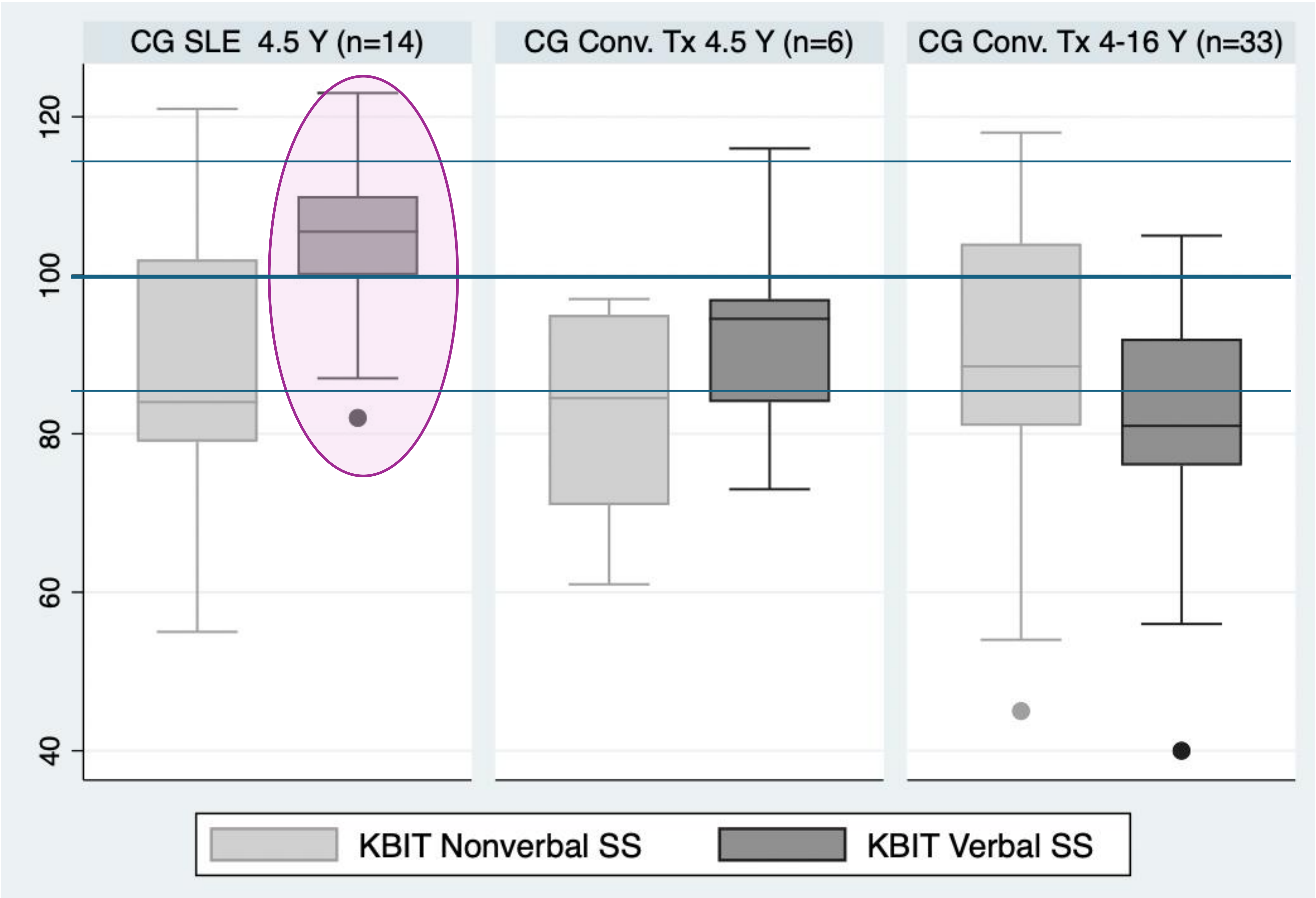


CG SLE 4.5 Y (n=14)
CG Conv. Tx 4.5 Y (n=6)
CG Conv. Tx 4-16 Y (n=33)
120
100
80
60
40
KBIT Nonverbal SS
KBIT Verbal SS

Babble Boot Camp training is now available online:

NSS
Northern Speech Services

Babble Boot Camp© Basic Training #e321

Presenters: Nancy Potter, PhD, CCC-SLP & Victoria Heinlen, MS, CCC-SLP

https://www.northernspeech.com/early-intervention/babble-boot-camp-basic-training/

Branching out:
New populations of infants

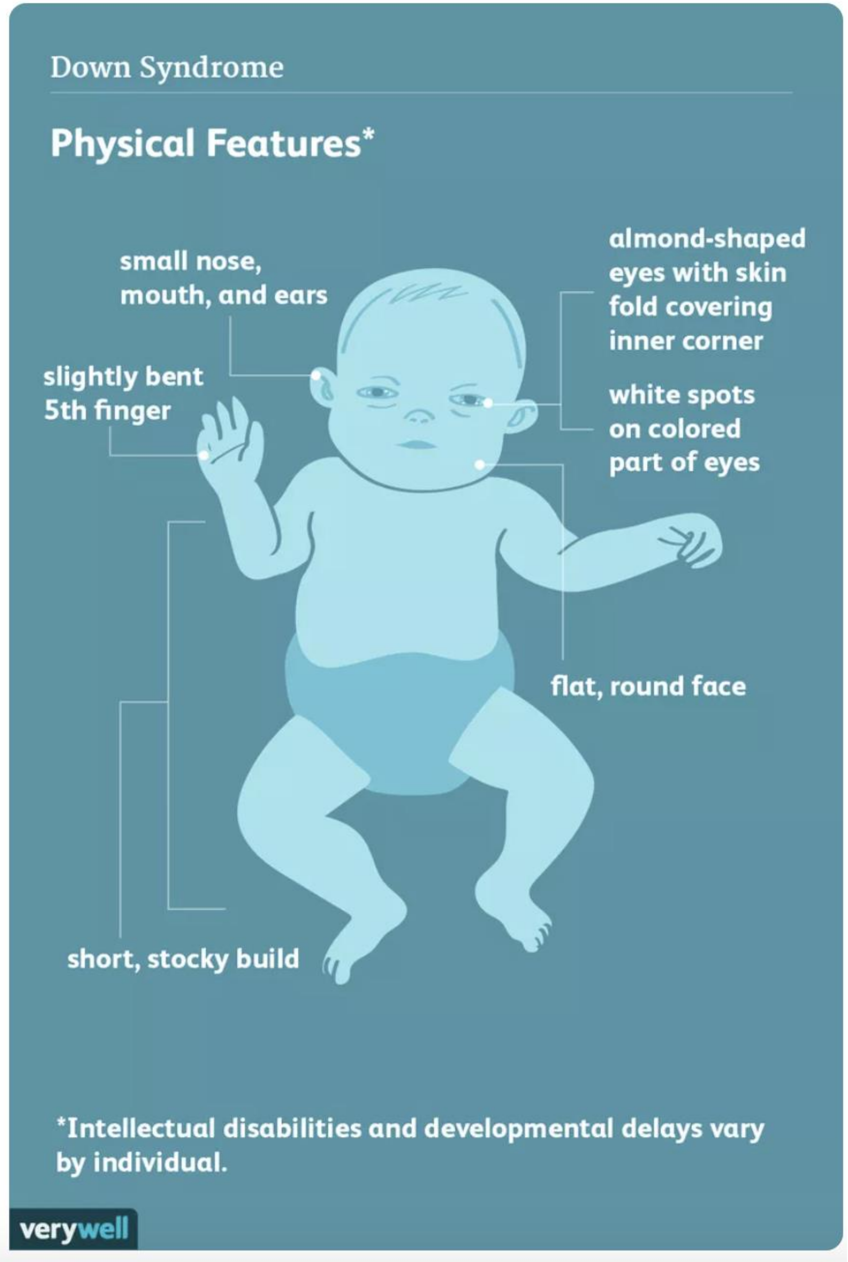
Down syndrome

Professor Sue Buckley OBE
CPsychol AFBPsS
Director for Science and Research
Down Syndrome Education International
Emeritus Professor of Developmental Disability
Dept Psychology, University of Portsmouth, UK.

	March 2022 • BBC ticks all the boxes for babies with Down syndrome. • Babies with Down syndrome have needs in speech and language. • There are not enough SLTs in the UK. • BBC is ideal because of parent training and telehealth – can be scaled up very easily

Sue Buckley is one of the world's leading researchers in the education and development of children with Down syndrome. Sue's research in the early 1980s led to the founding of the charity, Down Syndrome Education International (DSE).

https://gigisplayhouse.org/westchester/sue-buckley-seminar/



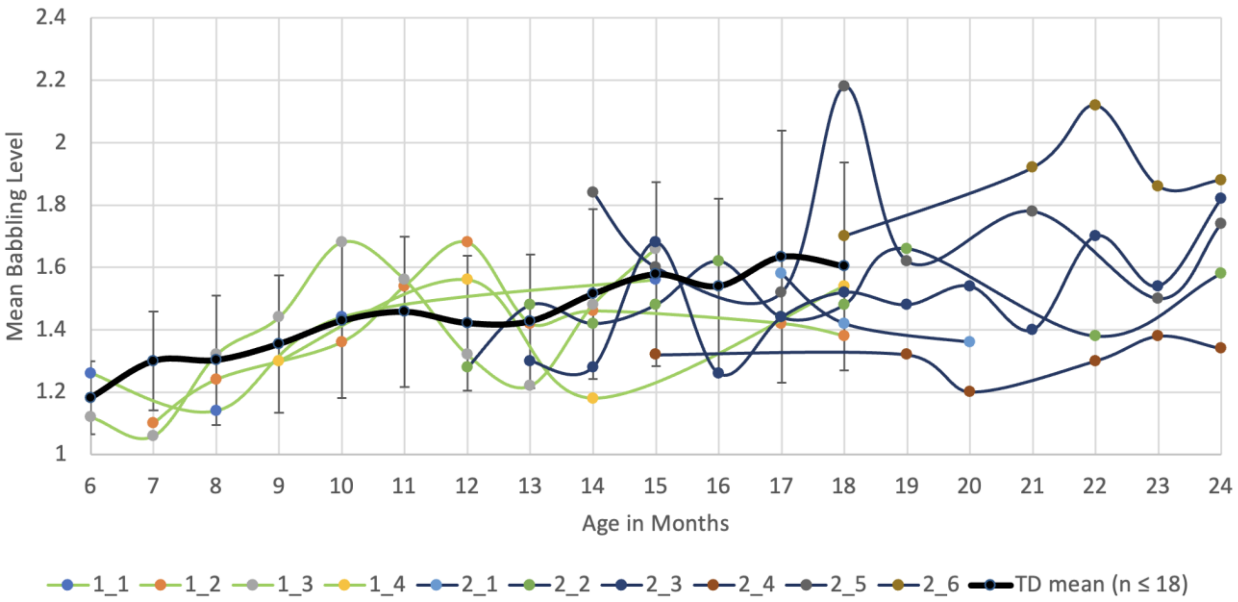
https://www.verywellhealth.com/symptoms-of-down-syndrome-1120463

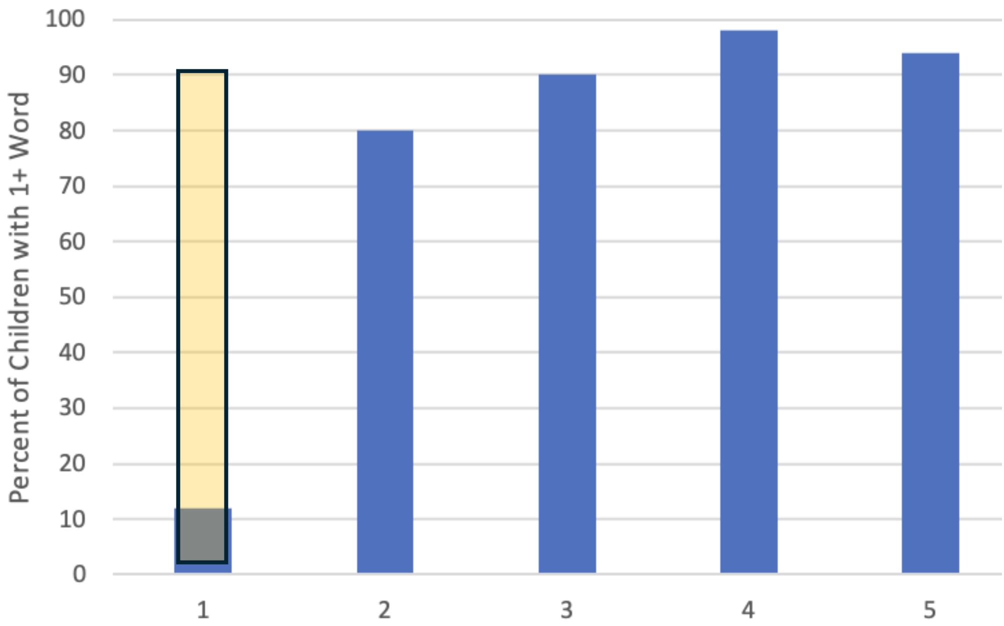
Developmental features across the lifespan

- Cardiac anomalies at birth (surgery)
- Gastrointestinal issues (surgery)
- Hearing loss
- Vision impairments
- Hypotonia
- Motor coordination
- Intellectual disability
- Speech delays
  - Speech difficult to understand
  - Speech sound errors, acoustic differences
- Language delays
  - First words around 2 years, small vocabulary, morphosyntax
  - Difficulty extracting linguistic regularities from ambient language
- Math delays
- Elevated risk for Alzheimer disease in adulthood
- Strong interpersonal skills
- Strong joint attention skills

Piloting BBC with 10 infants with Down syndrome

4 infants 4-5 m
6 infants 10-16 m
Weekly sessions for 10 months





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Bainbridge-Ropers Syndrome

Published symptoms and clinical features

Bainbridge-Ropers Syndrome (ASXL3)

ARRE
ASXL RARE RESEARCH ENDOWMENT

Communication
Speech delay/absent
Decreased expressive communication
Nonverbal communication
Alternative communication methods

Neurological
Microcephaly
Hypotonia
Seizures
Abnormal MRI

Motor
Motor delay
Stereotypies
Unsteady, broad gait
Oral motor impairment

Cognition
Intellectual delay
ADHD
Global developmental delay

Behavior/Emotional
Autism or autistic traits
Behavior disturbance
Self injurious/aggressive behavior
Repetitive movements
Happy demeanor
Hand flapping
Obsessive compulsive disorder
Poor eye contact
Maladaptive behavior
Anxiety

Musculoskeletal
Ulnar deviation of both wrists
Deep palmar creases
Scoliosis/other skeletal defects
Micro/retrognathia
Craniofacial abnormalities
Eye appearance abnormalities
Nose abnormalities
Hand and feet abnormalities
Ear appearance abnormalities
Dental and oral abnormalities
Delayed bone age

Gastrointestinal
Feeding difficulties
Reflux/vomiting
Tube feeding
Underweight
Constipation
Laryngomalacia
Other GI surgeries

Visual
Visual Impairment
Strabismus
Periorbital edema (swelling around eyes)
Nystagmus
Convergent or divergent squint

Sleep
Sleep apnea
Sleep disturbance
Bruxism

Cardio-pulmonary
Cardiac abnormalities
Respiratory distress and infections

Skin/Hair
Abnormal hair growth
Acanthuses Nigricans (dark patches of skin)
Nevus flammeus (forehead)
Poor temperature regulation

Other
Failure to thrive
Precocious puberty
Genital abnormalities
IUGR and pregnancy complications

This data was summarized from 109 individuals with Bainbridge-Ropers Syndrome reported in the medical literature as of October 2023 (Bacrot 2018; Bainbridge 2013; Balasubramanian 2017, 2020; Chinen 2018; Cuddapah 2021; Dinwiddie 2013; Dad 2017; Fu 2019; Gou 2019; Hori 2016; Koboldt 2018; Kuechler 2017; Li 2020; Meyers 2018; Qiao 2019; Schirwani 2020, 2021, 2023; Srivastava 2016; Verhoeven 2018; Wang 2022; Wayhelova 2019; Yang 2020; Yu 2021; Zhang 2018).

The syndrome is named after Matthew Bainbridge and H. Hilger Ropers, two doctors who described the similar clinical characteristics of people with a variation on the ASXL3 gene in 2013.

Age of diagnosis:
4 to 7 years, not newborn screening!

Absent speech	Complete lack of development of speech and language abilities. **Synonyms:** Absent speech development; Lack of language development; Lack of speech; No speech development; No speech or language development; Nonverbal	Frequency Frequent Uncommon — Always

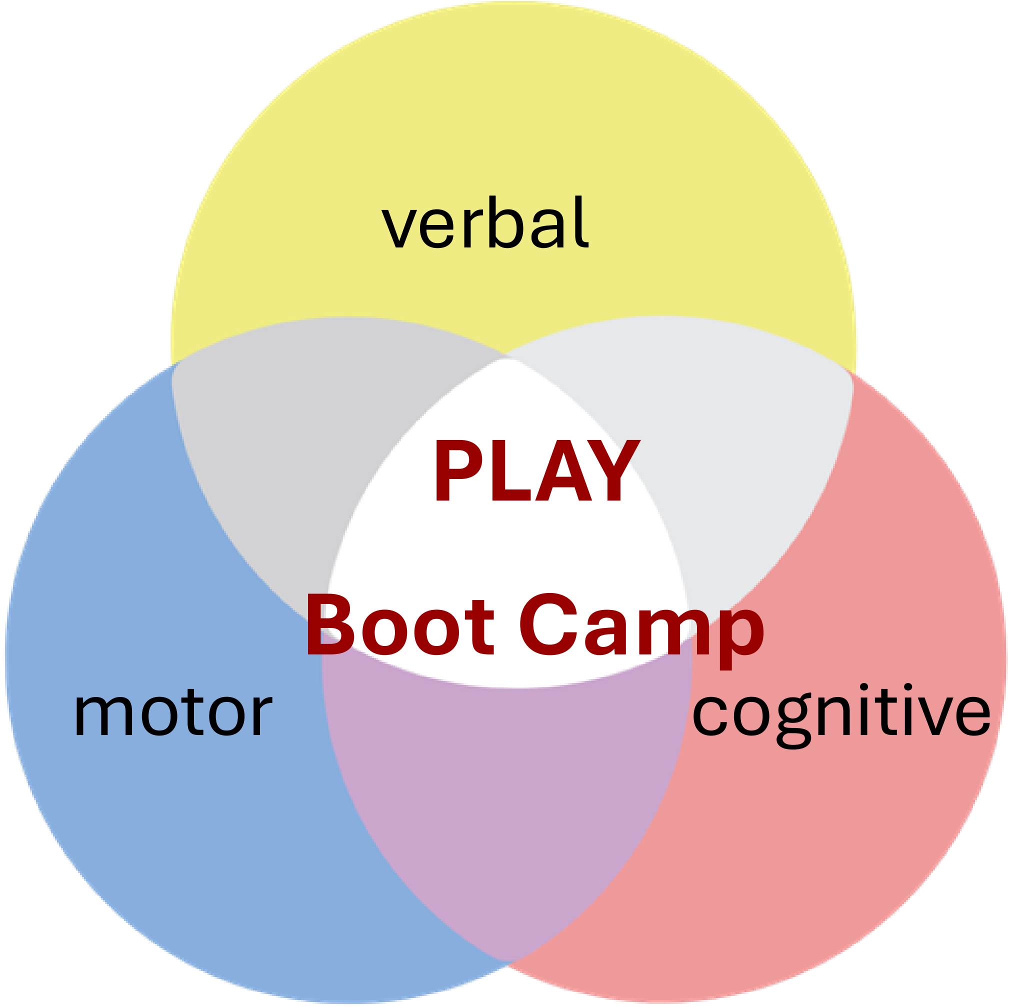
Initially, extremely rare
- 4 cases in 2013
- 12 new cases in 2016
- 300 in 2020
- Now new cases every week

https://www.arrefoundation.org/bainbridge-ropers

Branching out:
Broader intervention targets

An **Integrated** Approach to Support Children with DS

Play Boot Camp©



Based on an Established Model (BBC)

Developed by an interdisciplinary, international team

Known synergies among these developmental domains – an integrated approach would predict maximal progress.

The many faces of dyslexia: Clinical focus on the endophenotype

Example: Diminished neural adaptation

ARTICLE · Volume 92, Issue 6, P1383-1397, December 21, 2016 · *Open Archive*

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Dysfunction of Rapid Neural Adaptation in Dyslexia

Tyler K. Perrachione [1,3,4,6] · Stephanie N. Del Tufo [1,3] · Rebecca Winter [3] · … · Satrajit S. Ghosh [2,3] · Joanna A. Christodoulou [1,5] · John D.E. Gabrieli [1,3] … Show more

Affiliations & Notes · Article Info

Contents lists available at ScienceDirect
Clinical Neurophysiology
journal homepage: www.elsevier.com/locate/clinph

Auditory gating in adults with dyslexia: An ERP account of diminished rapid neural adaptation

Beate Peter[a,b,*], Hunter McCollum[a], Ayoub Daliri[a], Heracles Panagiotides[c]

[a] Speech and Hearing Science, Arizona State University, Tempe, AZ, USA
[b] Department of Communication Sciences and Disorders, Saint Louis University, Saint Louis, MO, USA
[c] Jackson School of International Studies, University of Washington, Seattle, WA, USA

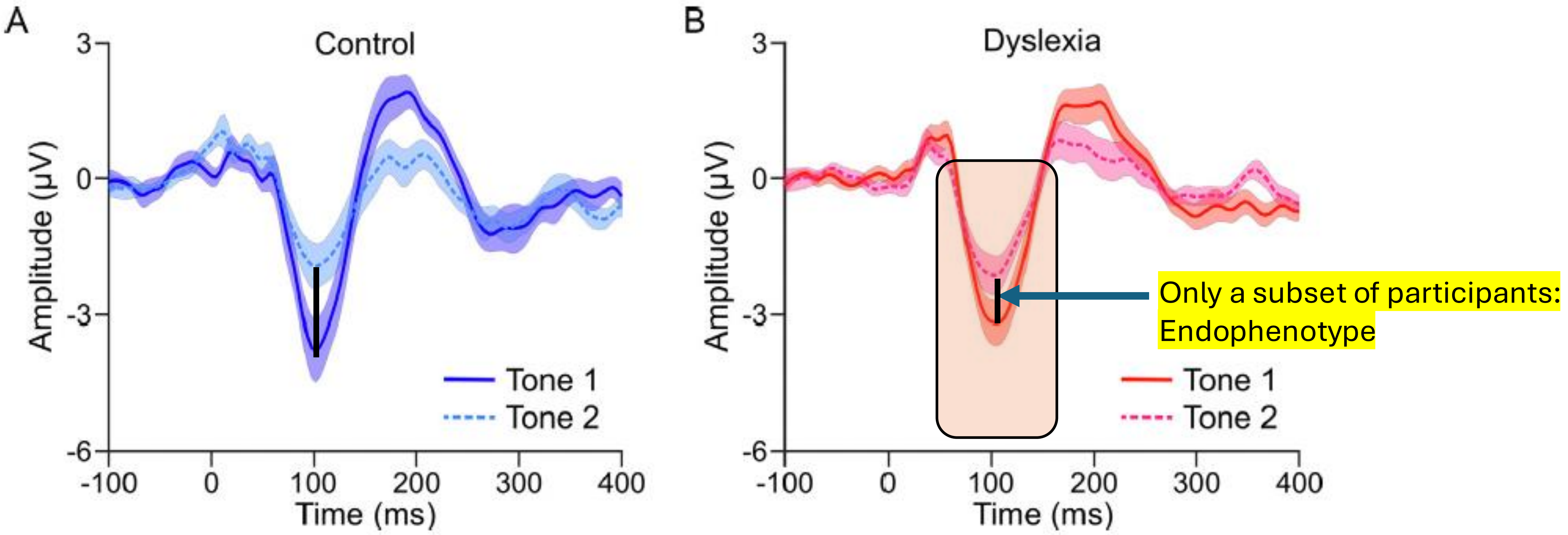
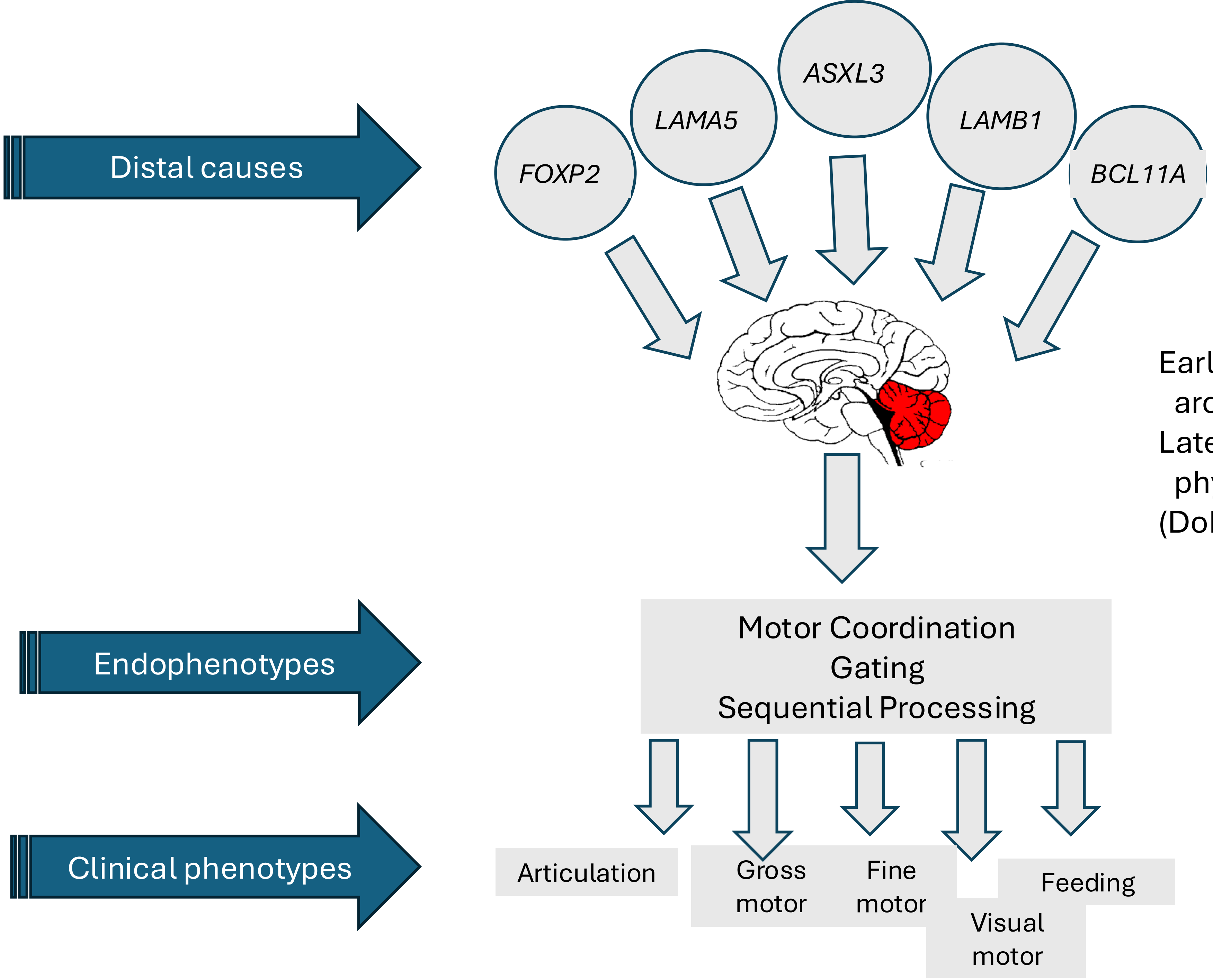


Fig. 1. Waveforms. Grand average ERP waveforms in response to the first and the second tones for the frontocentral electrodes in control participants (A) and participants with dyslexia (B). The shaded area corresponds to standard error of the mean in each time point.



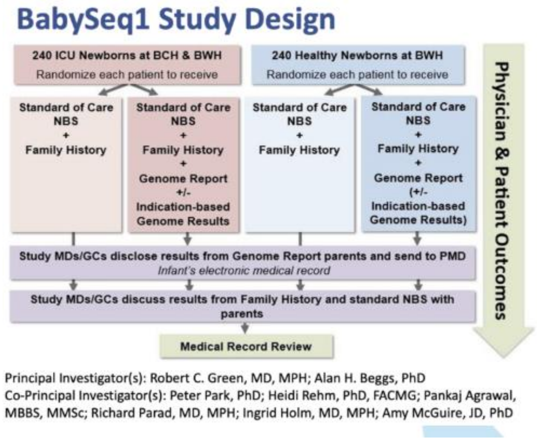
... plus many more genes!

Early developing brain:
architecture
Later developing brain:
physiology
(Dobrynin et al., 2025)

What is needed: New ways to identify children at risk early

Is whole genome sequencing at newborn screening a good idea?

The BabySeq Project is a research study exploring the use of genome sequencing in newborns.



The BabySeq Project

Implementation of Whole Genome Sequencing as Screening in a Diverse Cohort of Healthy Infants

Principal Investigator(s): Robert C. Green, MD, MPH; Ingrid A. Holm, MD, MPH
Co-Principal Investigator(s): Alan Beggs, PhD; Clement Bottino, MD, MPH; Kurt Christensen, PhD; Joy Dean, MD; Kelly East, MS; Bruce Gelb, MD; Carol R. Horowitz, MD, MPH; Bruce R. Korf, MD, PhD; Neil Lamb, PhD; Matt Lebo, PhD; Amy McGuire, JD, PhD; Stacey Pereira, PhD; Heidi Rehm, PhD; Jill O. Robinson, MA; Hadley S. Smith, PhD, MPSA; Hana Zouk, PhD

https://www.genomes2people.org/research/babyseq/

https://answers.childrenshospital.org/babyseq-2/

The future of genetics in our field: Leveraging precision-based approaches

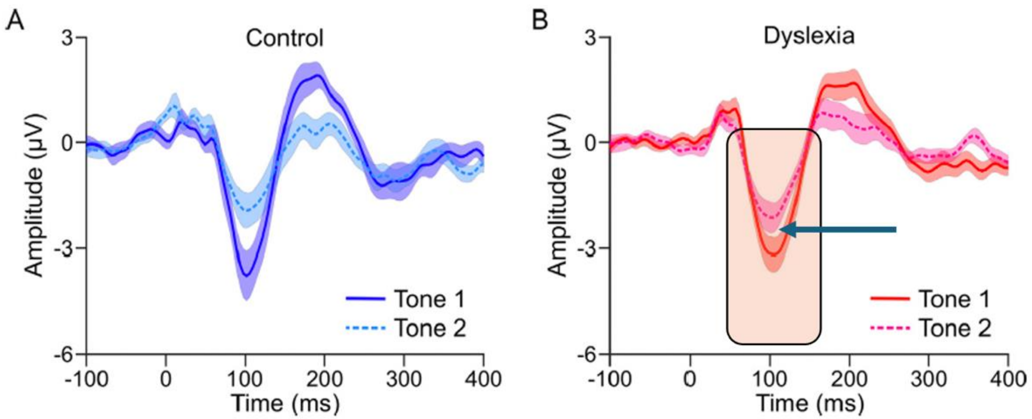
Prevention

Babble Boot Camp©

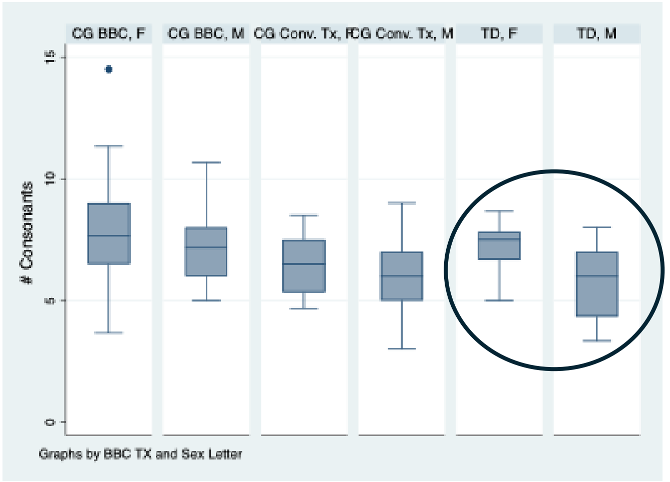
Play Boot Camp©

The next thing

Personalization by …



… endophenotype



… gender

… etc. … etc. … etc. … etc.

Genetic diagnosis: Opportunity instead of liability

If a child has a speech disorder due to a genetic cause, we don't treat that child because we can't change the DNA.

School district representative
Personal communication, Barabara Lewis

Many thanks

Collaborators

- Amy Armstrong-Heimsoth, NAU
- Laurel Bruce, ASU
- Sue Buckley, Down Syndrome Education International, UK
- Jenny Davis, ASU
- Linda Eng, ASU
- Lizbeth Finestack, UM
- Yookyung Kim, ASU
- Susan Loveall, UNL
- Nancy Potter, WSU
- Nancy Scherer, ASU
- Lauren Thompson, WSU
- Mark VanDam, WSU
- ... and many more

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- ASU Foundation Gift Account “**Babble Boot Camp**”
- ASU Social Science Research Institute

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