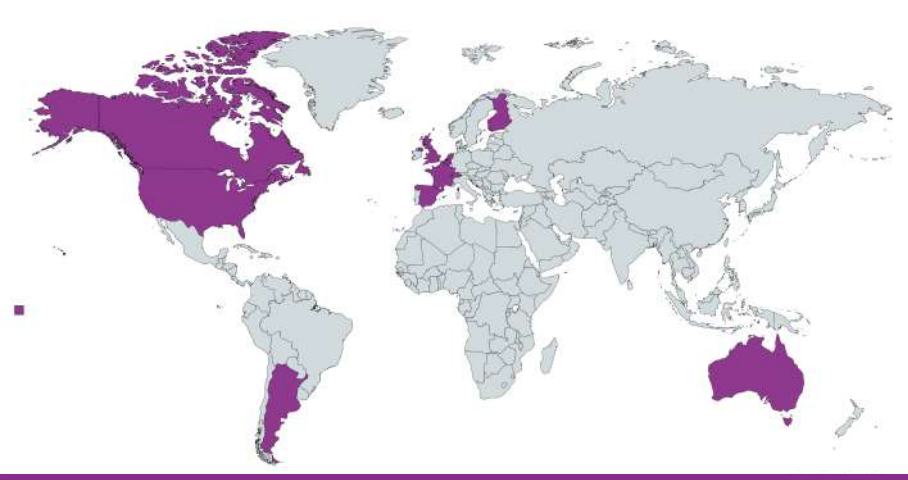


PACS1

The Natural History Project

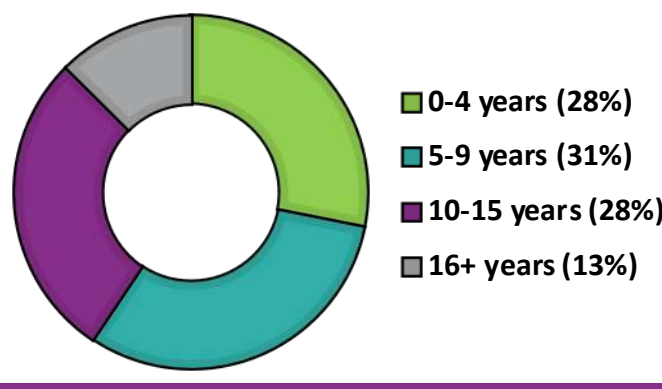
34 Parents from North America, Europe, Australia, and South America



Sex

50% ♀ **50%** ♂

Ages



INITIAL PRESENTATION

First sign of concern that brought child to medical attention :

50% - Delayed developmental milestones **18%** - Other
29% - Seizures or suspected seizures **3%** - Dysmorphic features

4.5 Months – Median age when first evaluated / **90%** by 12 Months

4.5 Years – Median age at PACS1 diagnosis / **90%** by 14 years

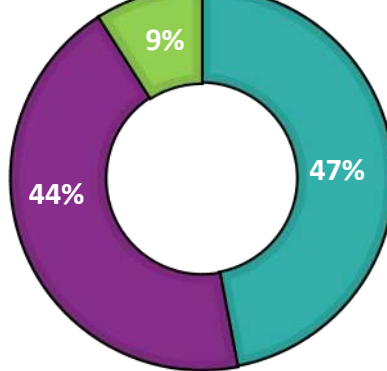
EPILEPSY

47% had an epilepsy diagnosis

- **7 Months** – Median age at seizure onset
- **17%** had seizures in the past 6 months

44% did **not** have an epilepsy diagnosis

9% were unsure



COMMUNICATION

84% have some form of symbolic language

74% can communicate with words

- **35%** can speak clearly
- **58%** can combine ≥ 3 words
- **55%** have a vocabulary of ≥ 100 words

58% can understand ≥ 100 words or pictures when said or shown

39% Effectively communicate even with people they don't know

Primary methods of communication:

68% Spoken Words **3%** Sign language
10% Gestures **3%** Sounds
3% Gaze or eye pointing

Common speech errors:

29% Substitutions
35% Omissions
32% Distortions
10% Stuttering

ACADEMICS*

53% some academic skills (reading, writing, counting)

43% Write a single letter

21% Copy words

50% Point to a specific letter

21% Write own name

32% Know what sounds letters make

32% Read signs

18% Read simple books

46% Count to 10

7% Count money

*Ages 3 and older

BEHAVIOR

Autism

32% Have features
35% Have a diagnosis

38% Frustration
17% Disobedience
14% Physical aggression
21% Temper tantrums
14% Verbal Aggression

Typical Behaviors

24% Sudden mood changes
72% Repetitive behaviors
45% Eyes-on supervision
28% Approach strangers
17% Self-injurious behavior

Behavioral Therapies**

35% Applied Behavior Analysis (ABA)

Parents rating therapy as having a good to excellent effect

67%

29% Sensory Integration Therapy

Parents rating therapy as having a good to excellent effect

80%

**Current and past

Positive Behaviors

86% Happy/Affectionate
62% Showing concern
55% Get along with adults

Activities Enjoyed

68% Listening to music **46%** Singing songs
71% Playing with water **43%** Puzzles
14% Playing instrument(s) **39%** Being sung to
36% Playing w/ friends **54%** Being read to
50% Building w/ blocks **57%** Coloring

FAMILY IMPACT

32% experience substantial financial challenges

43% find it hard to attend to their other children's' needs

39% say life feels like a rollercoaster

61% of parents experience moderate to severe fatigue

DAILY FUNCTIONS

16% dress themselves

61% use spoon correctly

71% mark or scribble

55% turn a page in a book

26% completely independent for toilet use

48% completes 4-5 piece puzzle

39% zip and unzip zippers

48% screw container lids on/off

48% walks independently in the community

16% runs well

13% rides a tricycle or bike with training wheels

77% picks up small objects with thumb and forefinger

3% can kick a ball

MEDICAL CONCERNS

Muscular

41% Hypotonia
18% Hypertonia
9% Ataxia

Neonatal

53% Feeding difficulty
15% Failure to thrive (FTT)
18% Jaundice
18% Neonatal seizures
24% NICU stay

Vision

26% Difficulty judging distance
21% Impaired depth perception

Gastrointestinal

41% Constipation
3% GERD

68% Report balance/steadiness problems

47% Due to poor muscle tone
44% Due to visual-motor integration difficulties

MRI

79% Have had a brain MRI
 • **15%** Normal
 • **26%** Equivocal
 • **35%** Abnormal

HEALTHCARE TEAM

Therapists Seen

75% Speech
75% Occupational
57% Physical

36% Behavioral
25% Psychologist
18% Developmental Peds.

Specialists Seen

57% Neurologist
32% Orthopedist
21% Otolaryngologist
61% Geneticist

25% Endocrinologist
36% Ophthalmologist
25% Special Needs Dentist
21% Gastroenterologist

6 – Median # of different specialist and therapists involved in care

64% Of children have a therapy appointment at least 2x per week

36% Of parents feel care is not well coordinated across specialists

* Throughout, "recent" refers to in the past 6 months

* Participants who can do the task independently of with occasional verbal prompts and reminders

The survey was designed in and administered through CLIRINX, Dublin Ireland

