

# Autism in Infants:

## Infant Day: Whānau Ora and Infant Wellbeing

June 11<sup>th</sup> 2010

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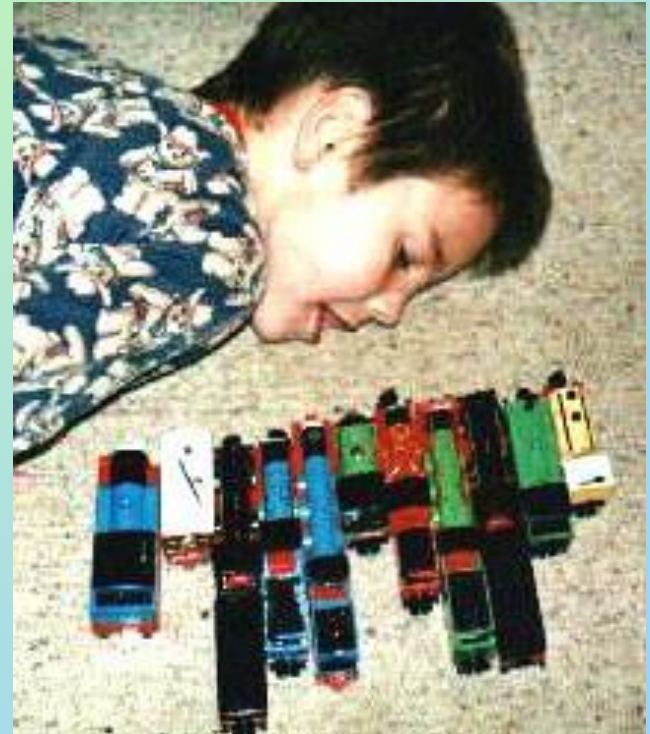
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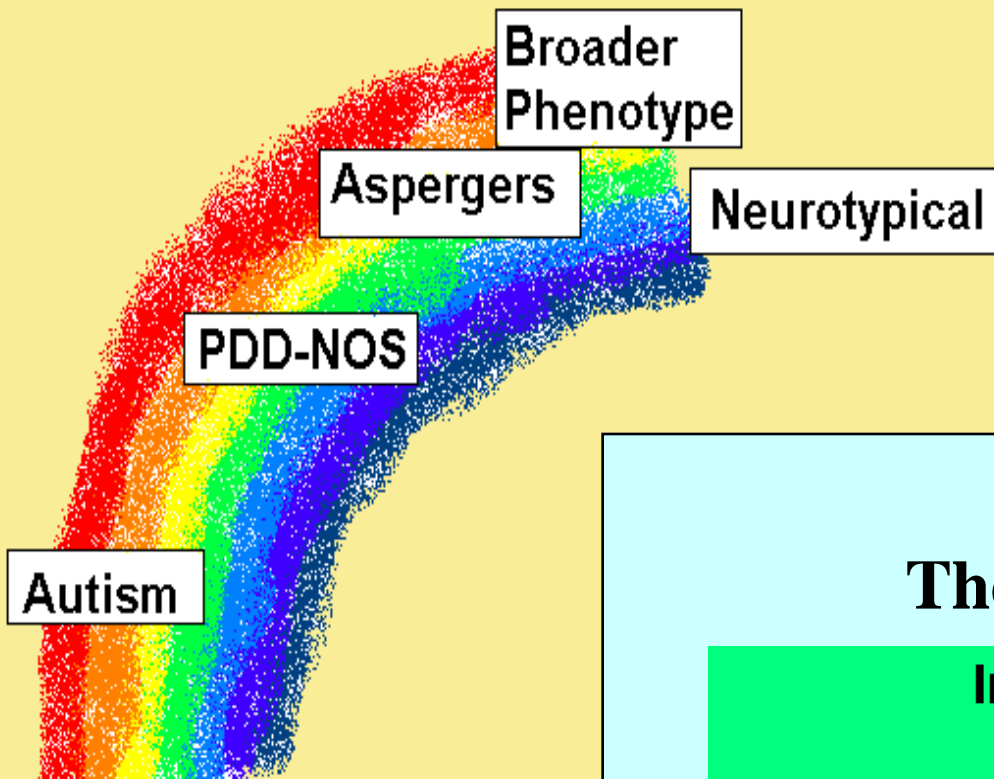


# Overview

- **Diagnostic Categories**
- **Conceptual Framework for Neuromaturational Disorders**
- **Why is Early Diagnosis important?**
- **Early Identification of ASD**
  - Family History
  - Social History
  - Biological Markers
  - Developmental Markers
- **Early correct diagnosis - trends in NZ**
- **Questions**

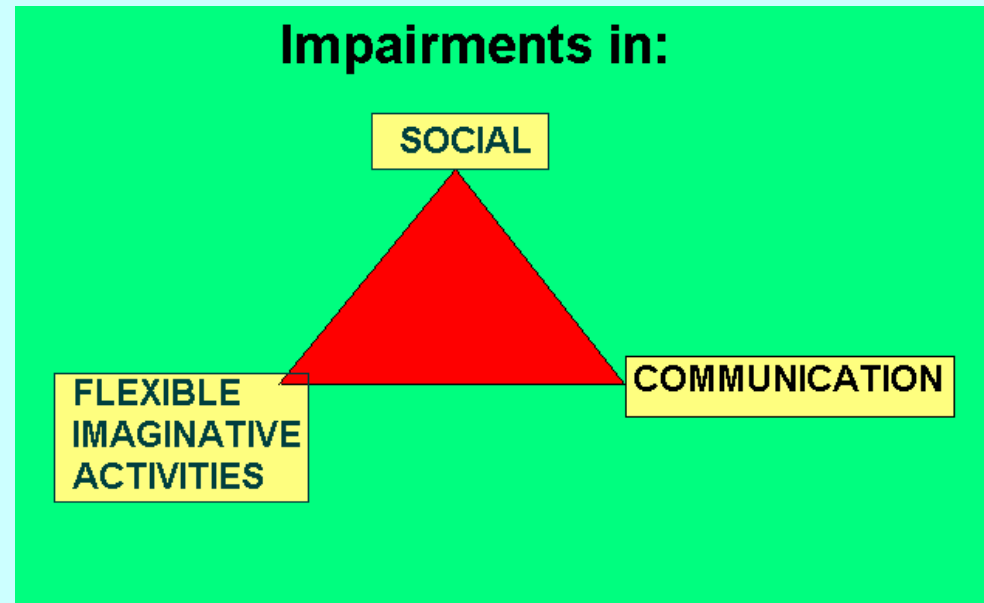


# Diagnostic Categories



## The Autism Triad

Impairments in:



- **Incidence Autism 1/1000**
- **Incidence all ASD up to 1/100?**
- **4:1 M:F ratio**

# Diagnostic Criteria: Autism (DSM-IV)

## 1) Qualitative impairment in social interaction, *as manifested by at least two of the following:*

- marked impairment in the use of multiple nonverbal behaviours such as eye-to-eye gaze, facial expression, body postures, and gestures to regulate social interaction.
- failure to develop peer relationships appropriate to developmental level.
- a lack of spontaneous seeking to share enjoyment, interests, or achievements with other people (eg, by a lack of showing, bringing or pointing out objects of interest).
- Lack of social and emotional reciprocity

# Diagnostic Criteria: Autism (DSM-IV)

2) Qualitative impairment in communication, *as manifested by at least one of the following:*

- delay in, or total lack of, the development of spoken language (not accompanied by an attempt to compensate through alternative modes of communication such as gesture or mime).
- in individuals with adequate speech, marked impairment in the ability to initiate or sustain conversation with others.
- stereotyped and repetitive use of language or idiosyncratic language.
- lack of varied, spontaneous make-believe play or social imitative play appropriate to developmental level.

# Diagnostic Criteria: Autism (DSM-IV)

3) Restricted repetitive and stereotyped patterns of behaviour, interests, and activities, *as manifested by at least one of the following:*

- encompassing preoccupation with one or more stereotyped and restricted patterns of interest that is abnormal either in intensity or focus.
- apparently inflexible adherence to specific, non-functional routines or rituals
- stereotyped and repetitive motor mannerisms (eg, hand or finger flapping or twisting, or complex whole-body movements).
- persistent preoccupation with parts of objects.

# Common Features of Autism

(not part of diagnostic criteria)

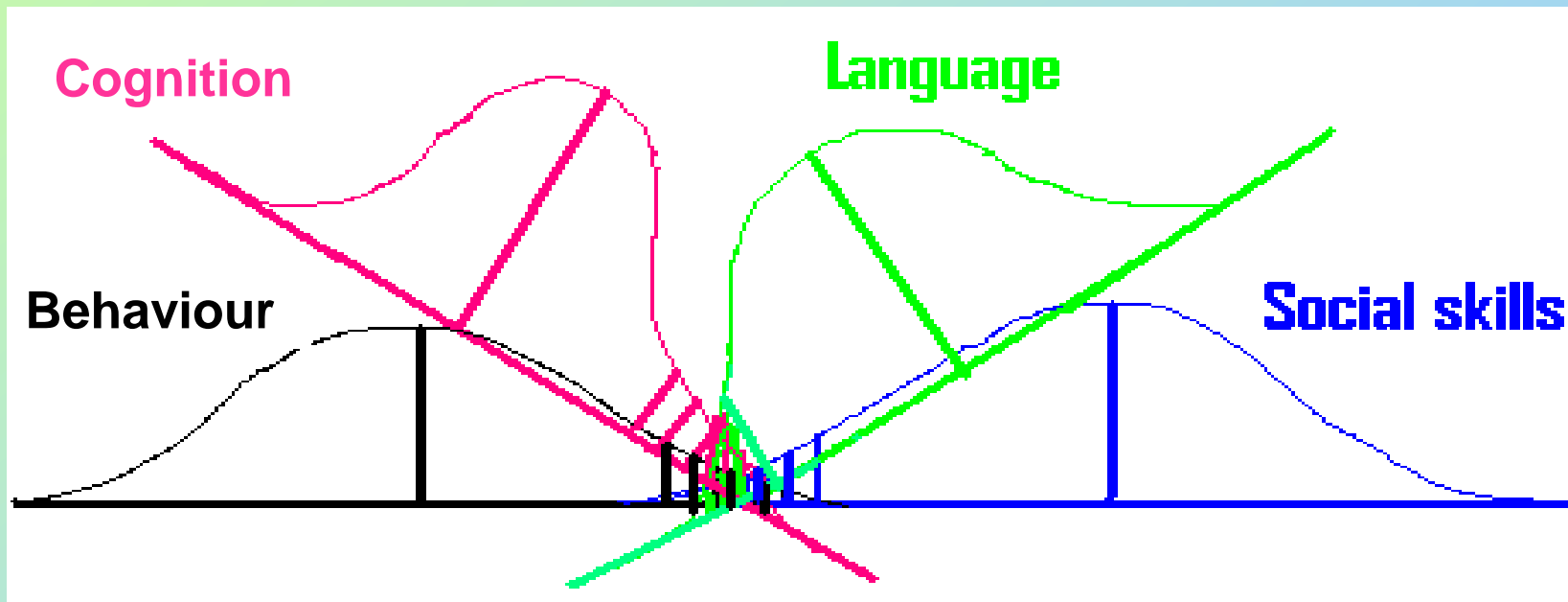
- Sensory Sensitivity
- Motor Clumsiness
- Anxiety
- Behaviour problems including aggression
- Sleeping and Eating problems
- Auditory Processing Disorder
- Attentional problems



# Conceptual Framework for Neuro-Maturational Disorders

## Concept 1:

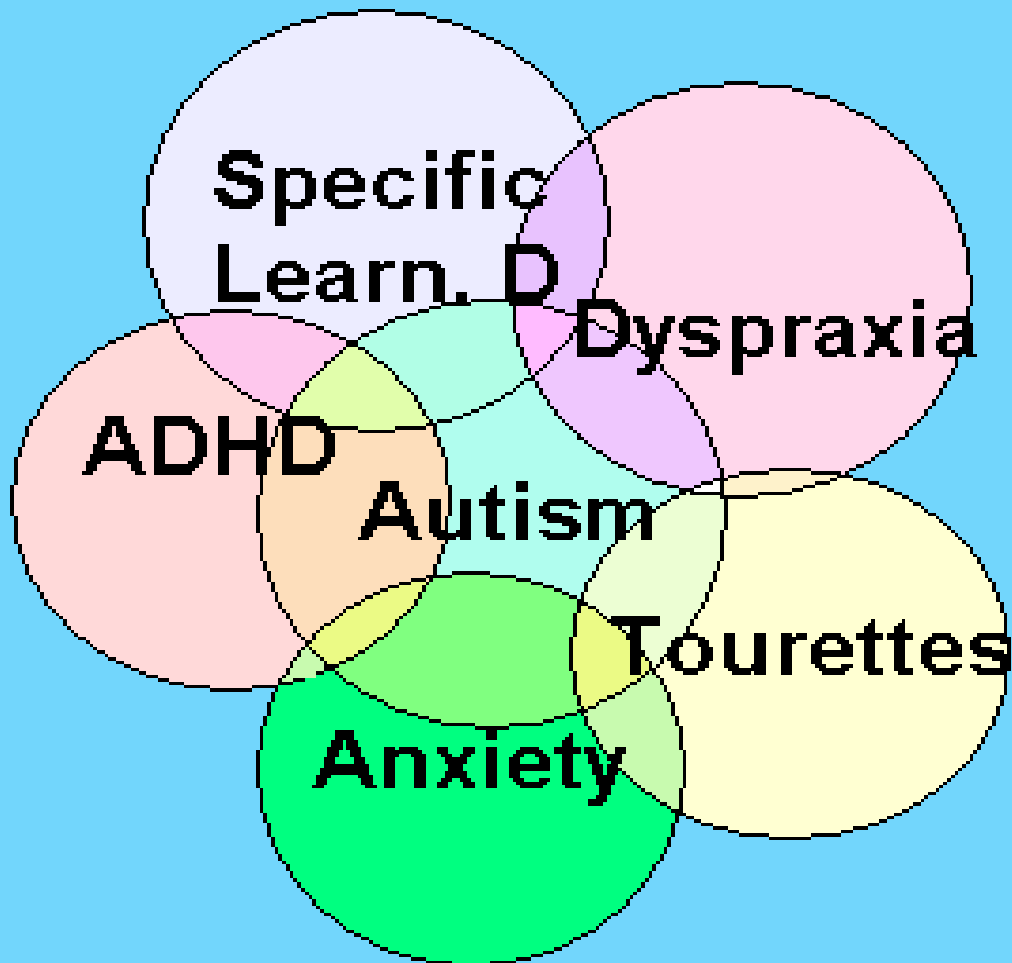
Deficits are in a continuum with the normal range



# Conceptual Framework for Neuro-Maturational Disorders

## Concept 2:

### Neuromaturational Disorder Overlap

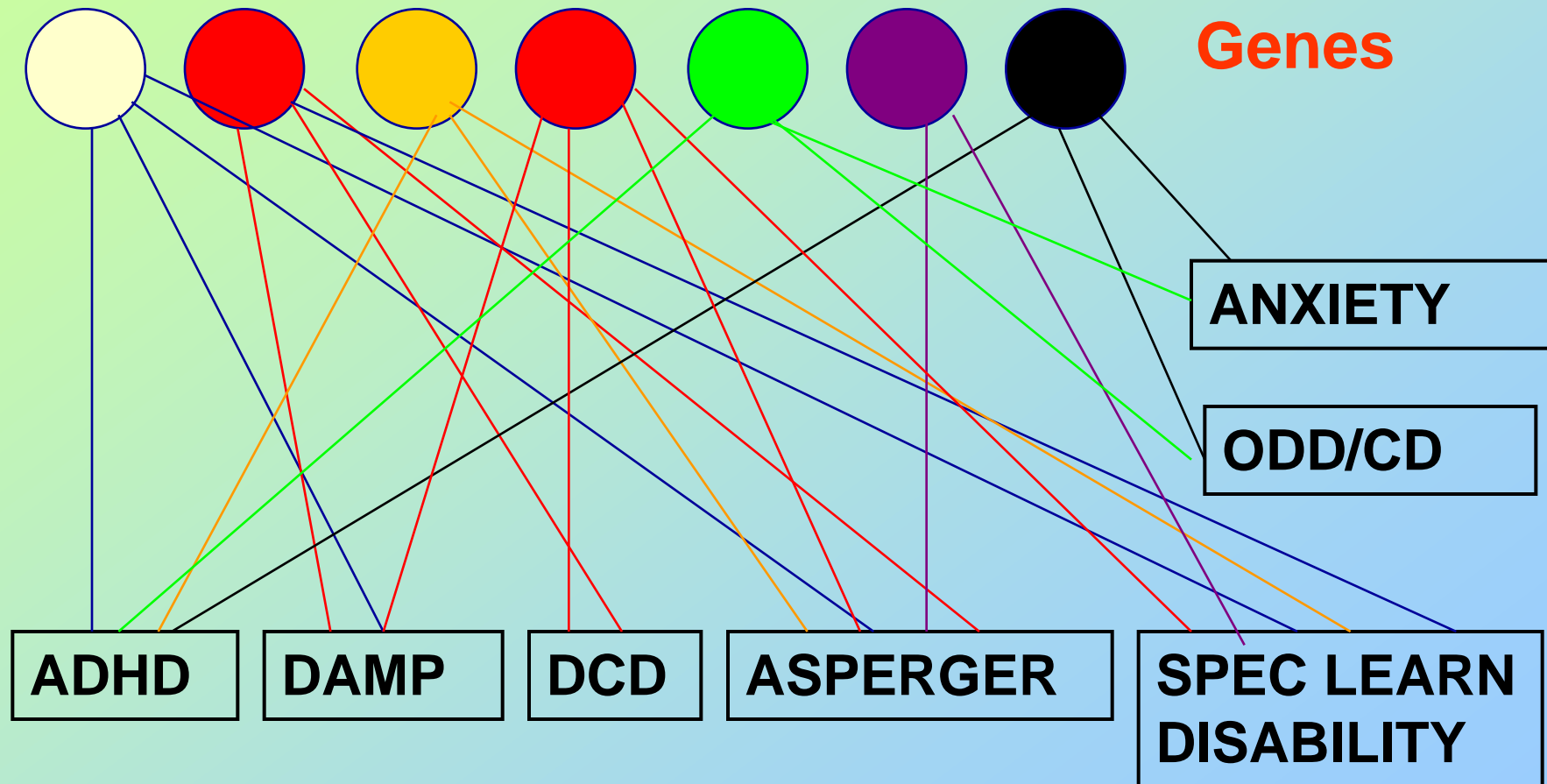


*Clumsiness, attention deficits, hyperactivity, social difficulties, anxiety and learning disabilities often cluster together*

*Always consider the Environment (Abuse / Neglect / Parental Mental Health) as potent contributors or causes of all these symptoms*

# Conceptual Framework for Neuro-Maturation Disorders

## Concept 3: Cause is largely polygenic



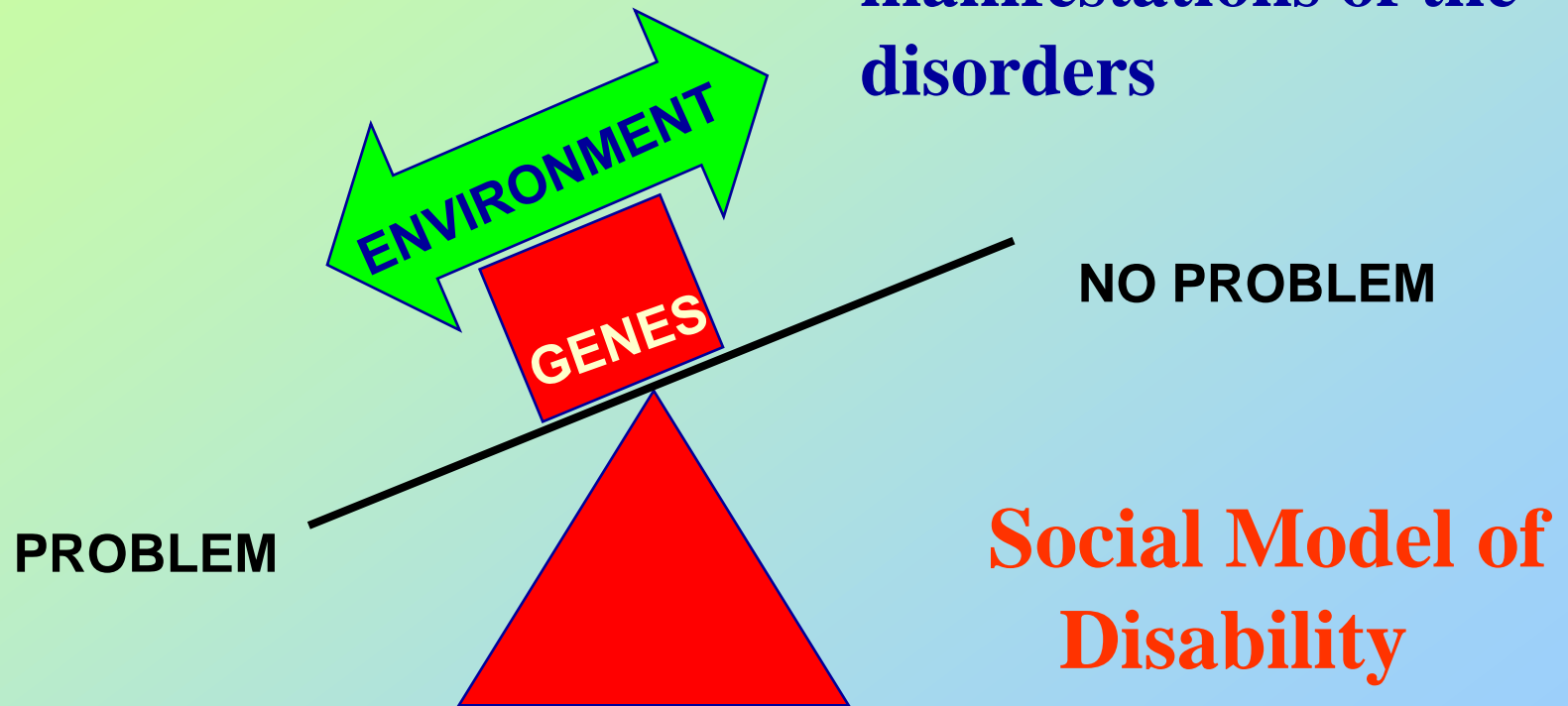
# Conceptual Framework for Neuro-Maturational Disorders

## Concept 3: Cause is largely polygenic

- All these conditions share genetic predisposition
- Autism twin studies 70-80% genetic
  - 50% monozygotic concordance, 5% dizygotic concordance
  - Siblings 50-fold risk compared to population risk
- Multiple interacting genes
- Family history invariably present
- In 2001, 4.7 % of the workforce and 3.65 % of the male NZ European workforce was employed in the IT industry
- In children with Autism, 8/26 = 31% Dads worked in IT and telecommunications (*in 2 yr local cohort of 68*)
- *Evidence that engineering, science and accountancy are over-represented in fathers of children with Autism*
- *Evidence of a ‘peak and trough’ cognitive profile in Autism relatives (better rote-learned and spatial abilities and difficulties with executive function)*

# Environment/Gene Interaction

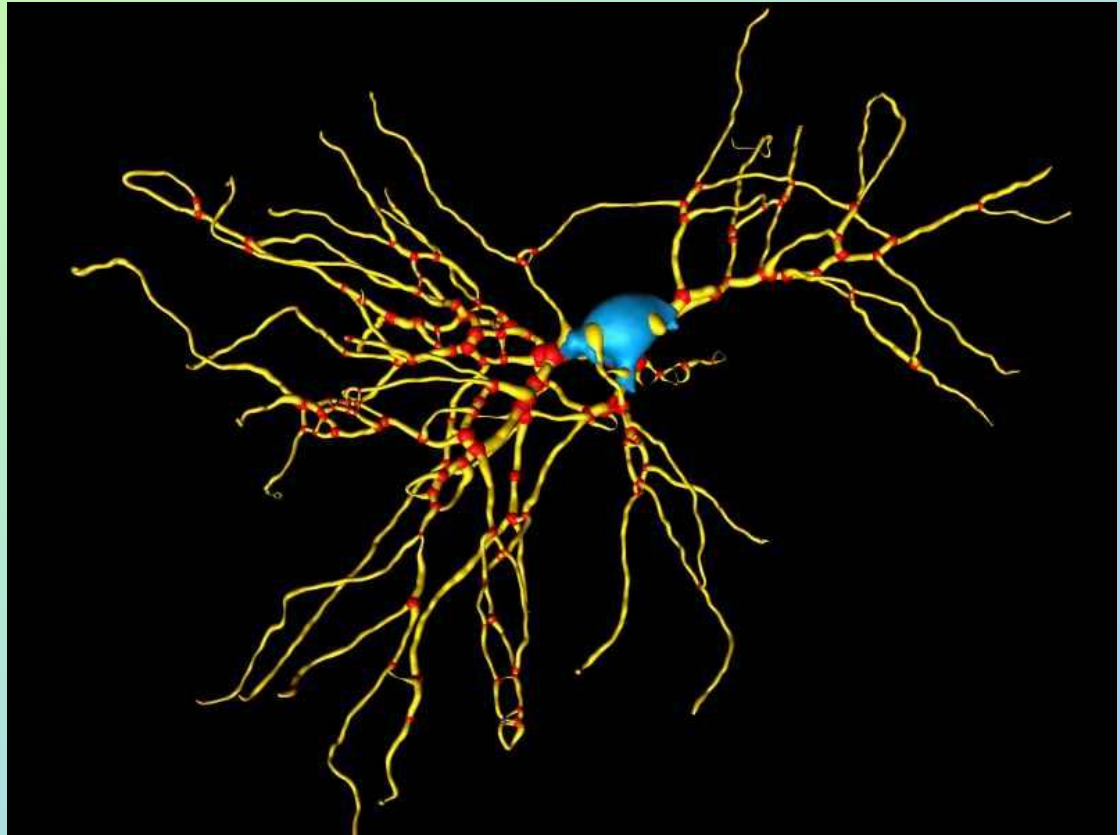
Concept 4: Environment plays a significant part in the manifestations of the disorders



An impairment is only disabling if the environment is not adapted to it

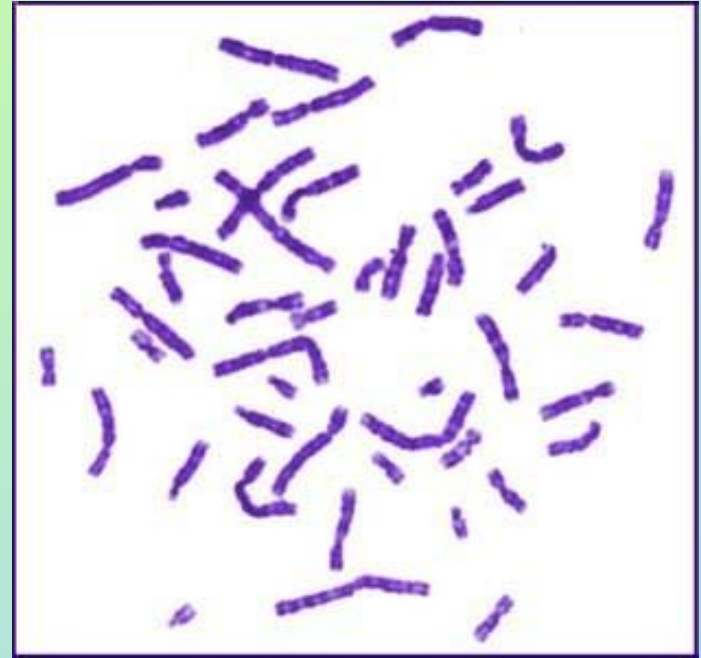
# Early Diagnosis - Why is it important?

- Most (85%) of a baby's brain development occurs after the birth - in the first 3 years
- Child's experiences during these years that enable brain to grow.
- Relationally-rich experiences provide children with the 'brain-food' they need to grow into happy, secure and well functioning adults



# Early Diagnosis – Why is it important?

- A child's developmental course is determined genetically, in combination with their environment
- Children are “wired” to learn, and will do so unless in a deprived environment
- Evidence that Early Educational Intervention improves long-term outcome



# Early Diagnosis - Why is it important?

- Parental concerns about their child's development need recognition/validation
- Early intervention does not require a diagnosis
- However, a diagnosis serves as a “short-hand” to direct the most appropriate action at the best time
- A correct diagnosis empowers by enabling understanding of needs and what the future may hold. It defines both strengths and difficulties
- Beware the dangers of a diagnosis – restricting future possibilities
- Accurate diagnosis must be timely, high quality and collaborative

# Early Identification of ASD

- **Family History**
  - Neuro-Developmental or Mental Health Disorders
  - Occupational History and Parental Personality Traits
- **Social History**
  - Environmental deprivation, neglect or abuse
  - Parental mental illness
  - Exposure to toxins in pregnancy
- **Biological Markers**
  - No genetic testing available
    - (except for single gene causes such as Fragile X, TS)
  - Increasing head size from 6 months
- **Developmental Markers: Screening**
  - CHAT

# Biological Markers

## Evidence of brain overgrowth in the first year of life in autism.

JAMA 2003 Courchesne E, Carper R, Akshoomoff N.

- Reduced head size at birth and a sudden and excessive increase in head size between 1 to 2 months and 6 to 14 months in 1/2 autistic children
- 59% of infants with autistic disorder showed accelerated HC growth trajectories ( $>2.0$  SDs from birth to 6 to 14 months) compared to 6% of healthy infants
- Brain overgrowth begins earlier than the first noticeable behavioural symptoms
- Head circumference predicts brain volume under 6 years ( $r = 0.93$ )
- Brain abnormally large in Autism
  - *average male 1450g in 8000 autopsy cases, largest adult brain reported 1750g, but 2 kg in a 3 yr old with autism.*
- Birth HC in infants with ASD was significantly smaller ( $n=48$ )
- After birth, HC increased 1.67 SDs and mean HC was at the 84th percentile by 6 to 14 months

# Developmental Markers: Screening

- Prospective UK study (Baron-Cohen 1996)
- Screened 16 000 children using CHAT
- Identified risk groups for developmental delay or autism
- Autism Discriminators:  
**Protodeclarative pointing,**  
**Gaze monitoring**  
**Pretend play**
- Risk groups administered ADI-R

## Autism Discriminators:

- **At 20 months:**
  - Limited range facial expression
  - Lack of interest in other children
  - Lack of pointing to express interest
  - Limited use of conventional gestures
- **At 42 months:**
  - Lack of seeking to share enjoyment
  - Lack of offering comfort
  - Lack of pointing to express interest
  - Limited use of conventional gestures
  - Lack of nodding
  - Lack of imaginative play

# Developmental Markers: Screening

- **Same cohort: screening procedures at age 3 and 5 years, and assessed at age 7.**
- **At follow-up a total of 50 cases of Autism were identified via all surveillance methods.**
- **19 Autism were successfully identified by the CHAT at 18 months.**
- **5 more at 42 months,**
- **4 between 42 months and 7 years, and**
- **25 at 7 years.**
- **Thus, the CHAT has a sensitivity of 38% and a specificity of 98% for identifying Childhood Autism.**

# Developmental Markers: Screening

- **Research using high risk infants (siblings of children with Autism) compared to low risk and looking for differences in behaviour:**
  - by 12 months of age, siblings who are later diagnosed with autism may be distinguished from other siblings and low-risk controls on the basis of:
    - (1) several specific behavioural markers, including atypicalities in eye contact, visual tracking, disengagement of visual attention, orienting to name, imitation, social smiling, reactivity, social interest and affect, and sensory-oriented behaviours;
    - (2) prolonged latency to disengage visual attention;
    - (3) a characteristic pattern of early temperament, with marked passivity and decreased activity level at 6 months, followed by extreme distress reactions, a tendency to fixate on particular objects in the environment, and decreased expression of positive affect by 12 months; and
    - (4) delayed expressive and receptive language.
  - Int J Dev Neurosci. 2005 Apr-May;23(2-3):143-52. Behavioral manifestations of autism in the first year of life. Zwaigenbaum L, Bryson S, Rogers T, Roberts W, Brian J, Szatmari P.

# Developmental Markers: Screening

- **Research using high risk infants (siblings of children with Autism) compared to low risk and looking for differences in behaviour:**
  - 108 infants at high and 18 at low risk for autism were tested from age 14 to 36 months.
  - Outcome diagnostic classification at 30 or 36 months:
    - ASD - early diagnosis, n = 16
    - ASD - later diagnosis, n = 14
    - broader autism phenotype n = 19
    - normal n = 58.
  - Social, communication, and play behaviour in the early-diagnosis group differed from that in all other groups by 14 months of age.
  - By 24 months, the later-diagnosis group differed from the non-autism spectrum disorder groups in social and communication behaviour, but not from the early-diagnosis group.
  - Examination of growth trajectories suggests that autism may involve developmental arrest, slowing, or even regression..
    - Arch Gen Psychiatry. 2007 Jul;64(7):853-64.Social and communication development in toddlers with early and later diagnosis of autism spectrum disorders.Landa RJ, Holman KC, Garrett-Mayer E.

# Developmental Markers: Screening

- **Research using high risk infants (siblings of children with Autism) compared to low risk and looking for differences in behaviour:**
  - 87 infants at high and at low risk for autism were tested at target ages 6, 14, and 24 months
  - Outcome diagnostic classification at 24 months:
    - ASD
    - Language Delay
    - Normal
  - No statistically significant group differences were detected at 6 months.
  - By 14 months of age, the ASD group performed significantly worse than the unaffected group on all scales except Visual Reception.
  - By 24 months of age, the ASD group performed significantly worse than the unaffected group in all domains,
  - The developmental trajectory of the ASD group was slower than the other groups', and showed a significant slowing in performance between 14 and 24 months of age in ASD
    - J Child Psychol Psychiatry. 2006 Jun;47(6):629-38. Development in infants with autism spectrum disorders: a prospective study. Landa R, Garrett-Mayer E.

# Developmental Markers: Screening

## Absolute Indicators for Immediate Evaluation:

- No babbling or pointing or other gesture by 12 months
- No single words by 16 months
- No 2-word spontaneous non-echolalic phrases by 24 months
- Any loss of ANY language or social skills at ANY age

*NZ Autism Guideline, based on Filipek, Accardo et al. The Screening and Diagnosis of Autistic Spectrum Disorders. Journal of Autism and Developmental Disorders 1999, 29 (6); 439-484*

- **Routine Developmental Surveillance:**

CDI                      ASQ  
PEDS                    BRIGANCE

- **Autism Screen:**

CHAT                    PDDST – Stage1

- **Diagnostic Assessments:**

ADI-R                    PDDST-Stage 3  
DISCO                   CARS  
GARS                    ADOS- G  
PIA

# Early Correct Diagnosis – Trends in NZ

- **Improving knowledge in Early Childhood Professionals (i.e. Plunket, Preschool Teachers) of normal developmental and of developmental disabilities → earlier recognition and referral**
- **Better accessible information sources (internet)**
- **Smaller, later families and reduced care giving networks may have decreased parent knowledge**
- **Improving diagnostic services nationally**
- **Shorter wait and high accuracy locally**



# Summary

- ASD presents as a social communication and behavioural disorder in young children
- Awareness of the genetic and environmental contributors allows earlier recognition of risk factors
- Developmental screening may pick up signs by 15-18 months
- The earlier the diagnosis is made and appropriate interventions are put in place the better the outcome

**Questions?**