TANNIS WIEBE, MD, FRCPC

FAMILY MEDICINE LECTURE SERIES

JANUARY 11, 2019

CONFLICT OF INTEREST STATEMENT

NO RELATIONSHIPS WITH COMMERCIAL INTERESTS

OBJECTIVES

- DESCRIBE THE CORE FEATURES OF AUTISM SPECTRUM DISORDER
- REVIEW PRESENTING SIGNS AND SYMPTOMS THAT MAY WARRANT REFERRAL FOR ASSESSMENT
- DISCUSS STEPS TO TAKE WHILE AWAITING ASSESSMENT
- REVIEW THE DIAGNOSTIC PROCESS
- OVERVIEW OF INTERVENTION AND ONGOING CARE

WHAT IS AUTISM?

- IMPAIRED SOCIAL INTERACTION AND COMMUNICATION SKILLS
- UNUSUAL BEHAVIOURS

AUTISM SPECTRUM DISORDER (DSM-5)

- A. **PERSISTENT DEFICITS IN SOCIAL COMMUNICATION AND SOCIAL INTERACTION ACROSS MULTIPLE CONTEXTS**, AS MANIFESTED BY THE FOLLOWING, CURRENTLY OR BY HISTORY (EXAMPLES ARE ILLUSTRATIVE, NOT EXHAUSTIVE, SEE TEXT):
  - **DEFICITS IN SOCIAL-EMOTIONAL RECIPROCITY**, RANGING, FOR EXAMPLE, FROM ABNORMAL SOCIAL APPROACH AND FAILURE OF NORMAL BACK-AND-FORTH CONVERSATION; TO REDUCED SHARING OF INTERESTS, EMOTIONS, OR AFFECT; TO FAILURE TO INITIATE OR RESPOND TO SOCIAL INTERACTIONS.
  - **DEFICITS IN NONVERBAL COMMUNICATIVE BEHAVIORS USED FOR SOCIAL INTERACTION**, RANGING, FOR EXAMPLE, FROM POORLY INTEGRATED VERBAL AND NONVERBAL COMMUNICATION; TO ABNORMALITIES IN EYE CONTACT AND BODY LANGUAGE OR DEFICITS IN UNDERSTANDING AND USE OF GESTURES; TO A TOTAL LACK OF FACIAL EXPRESSIONS AND NONVERBAL COMMUNICATION.
  - **DEFICITS IN DEVELOPING, MAINTAINING, AND UNDERSTANDING RELATIONSHIPS**, RANGING, FOR EXAMPLE, FROM DIFFICULTIES ADJUSTING BEHAVIOR TO SUIT VARIOUS SOCIAL CONTEXTS; TO DIFFICULTIES IN SHARING IMAGINATIVE PLAY OR IN MAKING FRIENDS; TO ABSENCE OF INTEREST IN PEERS.

- *SPECIFY* CURRENT SEVERITY: **SEVERITY IS BASED ON SOCIAL COMMUNICATION IMPAIRMENTS**

Diagnostic and statistical manual of mental disorders- 5th edition. American Psychiatric Association.

AUTISM SPECTRUM DISORDER (DSM-5)

- B. **RESTRICTED, REPETITIVE PATTERNS OF BEHAVIOR, INTERESTS, OR ACTIVITIES,** AS MANIFESTED BY AT LEAST TWO OF THE FOLLOWING, CURRENTLY OR BY HISTORY (EXAMPLES ARE ILLUSTRATIVE, NOT EXHAUSTIVE; SEE TEXT):
  - **STEREOTYPED OR REPETITIVE MOTOR MOVEMENTS, USE OF OBJECTS, OR SPEECH** (E.G., SIMPLE MOTOR STEREOTYPIES, LINING UP TOYS OR FLIPPING OBJECTS, ECHOLALIA, IDIOSYNCRATIC PHRASES).
  - **INSISTENCE ON SAMENESS, INFLEXIBLE ADHERENCE TO ROUTINES, OR RITUALIZED PATTERNS OF VERBAL OR NONVERBAL BEHAVIOR** (E.G., EXTREME DISTRESS AT SMALL CHANGES, DIFFICULTIES WITH TRANSITIONS, RIGID THINKING PATTERNS, GREETING RITUALS, NEED TO TAKE SAME ROUTE OR EAT SAME FOOD EVERY DAY).
  - **HIGHLY RESTRICTED, FIXATED INTERESTS THAT ARE ABNORMAL IN INTENSITY OR FOCUS** (E.G., STRONG ATTACHMENT TO OR PREOCCUPATION WITH UNUSUAL OBJECTS, EXCESSIVELY CIRCUMSCRIBED OR PERSEVERATIVE INTEREST).
  - **HYPER- OR HYPOREACTIVITY TO SENSORY INPUT OR UNUSUAL INTERESTS IN SENSORY ASPECTS OF THE ENVIRONMENT** (E.G., APPARENT INDIFFERENCE TO PAIN/TEMPERATURE, ADVERSE RESPONSE TO SPECIFIC SOUNDS OR TEXTURES, EXCESSIVE SMELLING OR TOUCHING OF OBJECTS, VISUAL FASCINATION WITH LIGHTS OR MOVEMENT).

- *SPECIFY* CURRENT SEVERITY: **SEVERITY IS BASED ON RESTRICTED REPETITIVE PATTERNS OF BEHAVIOR**

- **C. SYMPTOMS MUST BE PRESENT IN THE EARLY DEVELOPMENTAL PERIOD** (BUT MAY NOT BECOME FULLY MANIFEST UNTIL SOCIAL DEMANDS EXCEED LIMITED CAPACITIES, OR MAY BE MASKED BY LEARNED STRATEGIES IN LATER LIFE).

- **D. SYMPTOMS CAUSE CLINICALLY SIGNIFICANT IMPAIRMENT IN SOCIAL, OCCUPATIONAL, OR OTHER IMPORTANT AREAS OF CURRENT FUNCTIONING.**

- **E. THESE DISTURBANCES ARE NOT BETTER EXPLAINED BY INTELLECTUAL DISABILITY (INTELLECTUAL DEVELOPMENTAL DISORDER) OR GLOBAL DEVELOPMENTAL DELAY.** INTELLECTUAL DISABILITY AND AUTISM SPECTRUM DISORDER FREQUENTLY CO-OCCUR; TO MAKE COMORBID DIAGNOSES OF AUTISM SPECTRUM DISORDER AND INTELLECTUAL DISABILITY, SOCIAL COMMUNICATION SHOULD BE BELOW THAT EXPECTED FOR GENERAL DEVELOPMENTAL LEVEL.

- ***SPECIFY* IF:**
  - **WITH OR WITHOUT ACCOMPANYING INTELLECTUAL IMPAIRMENT**
  - **WITH OR WITHOUT ACCOMPANYING LANGUAGE IMPAIRMENT**
  - **ASSOCIATED WITH A KNOWN MEDICAL OR GENETIC CONDITION OR ENVIRONMENTAL FACTOR**

IMPAIRED SOCIAL SKILLS

- DEFICITS IN SOCIAL-EMOTIONAL RECIPROCITY
- SHOWING
- SHARING ENJOYMENT
- JOINT ATTENTION
- HAVING A CONVERSATION

IMPAIRED SOCIAL SKILLS

- IMPAIRED NONVERBAL COMMUNICATION
  - EYE CONTACT
  - GESTURES
  - AFFECT

Video “Ricky”

IMPAIRED SOCIAL SKILLS

- DIFFICULTY DEVELOPING RELATIONSHIPS
- PEER INTEREST
- SHARING IMAGINATIVE PLAY
- MAKING FRIENDS

ATYPICAL BEHAVIOURS

- STEREOTYPED OR REPETITIVE BEHAVIOURS
  - MOTOR STEREOTYPIES
  - REPETITIVE/STEREOTYPED PLAY
  - ECHOLALIA
  - OVERLY FORMAL OR OTHERWISE UNUSUAL LANGUAGE

VIDEO- CHARLY

ATYPICAL BEHAVIOURS

- RIGIDITY
  - OVER-RELIANCE ON ROUTINE
  - RITUALS
  - BLACK AND WHITE THINKING

ATYPICAL BEHAVIOURS

- HIGHLY RESTRICTED OR UNUSUAL INTERESTS

ATYPICAL BEHAVIOURS

- SENSORY SYMPTOMS
  - OVER- OR UNDER-REACTIVE
  - SENSORY-SEEKING BEHAVIOUR

VIDEO- 2YO REPETITIVE PLAY

DIFFERENTIAL DIAGNOSIS

ANXIETY

- POOR EYE CONTACT
- SOCIAL INHIBITION
- DIMINISHED PEER INTEREST
- SELF-SOOTHING BEHAVIOURS
- RIGIDITY
- DIFFICULTY WITH TRANSITIONS AND CHANGES IN ROUTINE

ADHD

- EYE CONTACT FLEETING
- POOR PEER RELATIONSHIPS
- REACTIVE TEMPERAMENT
- DIFFICULTY WITH TRANSITIONS
- STEREOTYPIC MOVEMENTS
- POOR PLAY SKILLS

LANGUAGE DISORDER

- POOR COMMUNICATION SKILLS
- ECHOLALIA
- STEREOTYPED SPEECH
- SOCIAL INHIBITION
- DIFFICULTY WITH CHANGES IN ROUTINE
- POOR PLAY SKILLS

OTHER

- INTELLECTUAL DISABILITY/GLOBAL DEVELOPMENTAL DELAY
- FASD
- MOOD DISORDER
- OCD
- TIC DISORDER
- SOCIAL (PRAGMATIC) COMMUNICATION DISORDER

WHAT IF I'M STILL NOT SURE?

- SCREENING CHECKLISTS
  - M-CHAT: MODIFIED CHECKLIST FOR AUTISM IN TODDLERS (18-30 MONTHS)
    - HTTPS://WWW.M-CHAT.ORG/
- REFER TO GENERAL PEDIATRICIAN

SO YOU'RE REFERRING TO CDC…

- REFER TO AUDIOLOGY
- REFER TO SLP
- REFER TO OT
  - PLAY SKILLS, SOCIAL SKILLS, ATTENTION AND REGULATION, PICKY EATING, TOILETING, SENSORY
- ENCOURAGE SOCIALIZATION OPPORTUNITIES: DAYCARE, DROP-IN GROUPS, PLAY DATES
- SCREEN TIME COUNSELING
- PLEASE MENTION CONCERN RE: ASD IN CONSULT LETTER
- PLEASE SHARE YOUR CONCERNS FRANKLY WITH PARENTS
  - CONSIDER RECOMMENDING A WEBSITE LIKE WWW.FIRSTSIGNS.ORG SO THEY CAN FAMILIARIZE THEMSELVES WITH ASD

DIAGNOSIS

ADOS AND ADI

- ADI-R: EXTENSIVE STANDARDIZED INTERVIEW FOCUSING ON 3 DOMAINS- RECIPROCAL SOCIAL INTERACTION, COMMUNICATION, AND RESTRICTIVE/REPETITIVE/STEREOTYPED BEHAVIOUR
- ADOS-2: COLLECTION OF STANDARDIZED PLAY AND INTERVIEW ACTIVITIES THAT PRODUCES SCORES IN DOMAINS OF SOCIAL AFFECT AND RESTRICTED/REPETITIVE BEHAVIOUR
  - 4 MODULES FOR DIFFERENT AGES/LANGUAGE LEVELS
- ADOS SHOWS GOOD INTERRATER AND TEST-RETEST RELIABILITY, INTERNAL CONSISTENCY, AND DIFFERENTIATION OF NONSPECTRUM INDIVIDUALS[1]
- SENSITIVITY AND SPECIFICITY >75-80% WHEN TOOLS COMBINED; SPECIFICITY LOWER WHEN USED ALONE[2]

1. Lord et al., J Autism Dev Disord. 2000 Jun;30(3):205-23.
2. Risi et al., J Am Acad Child Adolesc Psychiatry. 2006 Sep;45(9):1094-103

POST-DIAGNOSIS
CONSIDERATIONS

ETIOLOGY

- DIAGNOSTIC YIELD OF GENETIC WORK-UP 30-40%
  - CHROMOSOMAL MICROARRAY (10%)
  - FRAGILE X (1-5%)
  - MECP2, PTEN
- CONSIDER BRAIN MRI IF ASSOCIATED FEATURES EG. SEIZURES, MICROCEPHALY, ATYPICAL REGRESSION, NEURO FINDINGS
- RECURRENCE RISK 3-10%
  - 30% IF 2 AFFECTED SIBLINGS
- RISK FACTORS INCLUDE ADVANCED PARENTAL AGE, LBW, PREMATURITY, TWINS, PRENATAL VPA

CO-OCCURRING CONDITIONS

- INTELLECTUAL DISABILITY ~50%
- LANGUAGE DISORDER
- SPECIFIC LEARNING DISORDER, DCD
- 70% HAVE ≥1 COMORBID MH DISORDER; 40% HAVE ≥2
  - ANXIETY (INCLUDING SELECTIVE MUTISM), ADHD, OCD, TIC DISORDERS, MOOD DISORDER, PSYCHOTIC DISORDER, ARFID
- EPILEPSY, SENSORY IMPAIRMENTS, CEREBRAL PALSY
- GENETIC DISORDERS
  - T21, CHARGE, FRAGILE X, PWS, ANGELMAN, RETT, TUBEROUS SCLEROSIS, METABOLIC DISORDERS
- SLEEP DIFFICULTIES, GI SYMPTOMATOLOGY

INTERVENTION

BEHAVIORAL APPROACHES

- PRINCIPLES OF OPERANT CONDITIONING
  - FUNCTIONAL ANALYSIS, BEHAVIOR MODIFICATION, ENVIRONMENTAL CHANGES
- ABA
  - DISCRETE TRIAL INSTRUCTION
  - GAINS SHOWN, BUT RISK OF POOR MAINTENANCE AND GENERALIZATION

DEVELOPMENTAL APPROACHES

- PRINCIPLES OF DEVELOPMENTAL THEORIES, EMPHASIZE MORE NATURALISTIC, PLAY-BASED INTERACTIONS
- FLOORTIME
  - CHILD-LED PLAY, DAY-TO-DAY SKILLS
  - GAINS SHOWN, GOOD PARENTAL SATISFACTION

HABILITATIVE INTERVENTIONS

- SPEECH-LANGUAGE THERAPY
  - LANGUAGE, SIGNS, PECS
- SOCIAL SKILLS TRAINING
- OCCUPATIONAL THERAPY
  - BEHAVIOR, PLAY SKILLS, PICKY EATING, TOILETING

ONGOING CARE

- TREATMENT OF ASSOCIATED CONDITIONS
- MEDICATIONS FOR SPECIFIC SYMPTOMS
  - IRRITABILITY, HYPERACTIVITY, STEREOTYPIES
- SAFETY ISSUES
- TRANSITION PLANNING

OUTCOMES

ADULTHOOD

- PROGNOSIS LARGELY TIED TO IQ AND LANGUAGE
  - FUNCTIONAL LANGUAGE BY 5YO SIGNIFICANT POSITIVE PROGNOSTIC FACTOR
- SIGNIFICANT IMPACT OF EXECUTIVE FUNCTIONING AND PSYCHIATRIC IMPAIRMENTS
- LOWER RATES OF COLLEGE ATTENDANCE
- SOCIAL ISOLATION
- LOW RATES OF INDEPENDENT LIVING (EVEN WITH NO ID)

MCQ

- A 2 YO BOY IS BROUGHT TO YOUR OFFICE FOR A WELL CHILD CHECK UP. HIS MOTHER DESCRIBES HIM AS A CHALLENGING CHILD WHO IS VERY ACTIVE AND HAS FREQUENT TEMPER TANTRUMS. HE IS UPSET BY SMALL CHANGES IN HIS ROUTINE. HE FOLLOWS DIRECTIONS ON HIS TERMS AND OFTEN DOES NOT TURN WHEN HIS NAME IS CALLED. HE DOES NOT PLAY WITH OTHER CHILDREN AND ONLY SPEAKS IN SINGLE WORDS. HE IS A PICKY EATER. DURING THE VISIT, THE BOY IS PREOCCUPIED WITH WAVING A PENCIL BACK AND FORTH IN FRONT OF HIS EYES. THE MOST LIKELY CAUSE OF THIS CHILD'S BEHAVIORS IS
- 1. ASD
- 2.GLOBAL DEVELOPMENTAL DELAY
- 3. FRAGILE X
- 4. SPEECH AND LANGUAGE DELAY
- 5. DIFFICULT TEMPERMENT

CASES

1. 3YO BOY WITH 10 WORDS, ECHOLALIA, FREQUENT TANTRUMS, ONLY EATS 6 DIFFERENT FOODS, DIFFICULTY WITH TRANSITIONS, SOLITARY PLAY

2. 2YO GIRL NONVERBAL, POOR RESPONSE TO JOINT ATTENTION, DOESN'T RESPOND TO NAME, REPETITIVE PLAY, HAND STEREOTYPIES

3. 8YO BOY WITH POOR EYE CONTACT, RESTRICTED AFFECT, ODD, SENSORY SENSITIVITIES, NO FRIENDS, SELF-INJURIOUS BEHAVIORS, RIGIDITY AROUND ROUTINES AND ENVIRONMENT

RECOMMENDED REFERENCES

- WEBSITES
  - HTTP://WWW.AUTISMSPEAKS.CA/
  - HTTP://FIRSTSIGNS.ORG/
  - HTTPS://WWW.SFARI.ORG/
  - HTTP://AUTISM.SESAMESTREET.ORG/

- ARTICLES

  - ANAGNOSTOU ET AL. AUTISM SPECTRUM DISORDER: ADVANCES IN EVIDENCE-BASED PRACTICE. CMAJ VOL.186 NO.7 APR. 2014 HTTP://WWW.CMAJ.CA/CONTENT/186/7/509.FULL
  - HARRINGTON, ALLEN. THE CLINICIAN'S GUIDE TO AUTISM. PIR VOL.35 NO.2 FEB. 2014
  - SIMMS, JIN. AUTISM, LANGUAGE DISORDER, SOCIAL (PRAGMATIC) COMMUNICATION DISORDER. PIR VOL.36 NO.8 AUG. 2015
  - ZWAIGENBAUM ET AL. EARLY IDENTIFICATION AND INTERVENTION FOR AUTISM SPECTRUM DISORDER. PEDIATRICS VOL.136 SUPP.1 OCT. 2015
  - MAGLIONE ET AL. NONMEDICAL INTERVENTIONS FOR CHILDREN WITH ASD. PEDIATRICS VOL.130 SUPP.2 NOV. 2012