Gillberg Neuropsychiatry Centre
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# Concept of ESSENCE and Support for Children and Adults with Developmental Disabilities


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# ESSENCE (neurodevelopmental/psychiatric disorders)

- **ESSENCE - Early Symptomatic Syndromes Eliciting Neurodevelopmental Clinical Examinations**
- **Predictors of *academic failure*, other school adjustment problems, social exclusion, *substance use and abuse*, psychiatric disorder (depression/GAD/PD/psychosis), eating disorders including obesity, accidents, *empathy problems*, antisocial lifestyle and *criminality* later in life, persistent autistic features "only", early death through accidents, criminality, and physical health problems (including difficulty-to-treat in diabetes)**
  - **ADHD** with or without **ODD/CD** (Oppositional Defiant Disorder/Conduct Disorder) 5-7%
  - **SLI** (Language Disorder inlcuding antecedents of Dyslexia) 5%
  - **DCD** (Developmental Coordination Disorder) 5%
  - **IDD (**Intellectual Disability/Intellectual Developmental Disorder) 2%
  - **ASD** (Autism Spectrum Disorder) 1.2%
  - **TD/TS/OCD** (Tic disorders/Tourette syndrome/OCD) 1%
  - **RAD** (Reactive Attachment Disorder/Disinhibited Social Engagement Disorder) 0.5-1.5%
  - **(BPS** (Behavioural Phenotype Syndromes, including FAS and VAS) 2%)
  - **(EP/NEUROMUSC** (Epilepsy syndromes and other neurological/neuromuscular disorders (HC, CP, Duchenne, myotonic dystrophy, neurometabolic): Landau-Kleffner Syndrome, CSWS, FS+, FS? 0.6%)
  - **(PANS** (Pediatric Acute-onset Neuropsychiatric Syndrome)? 0.1%)

## What are the "symptoms" of ESSENCE?

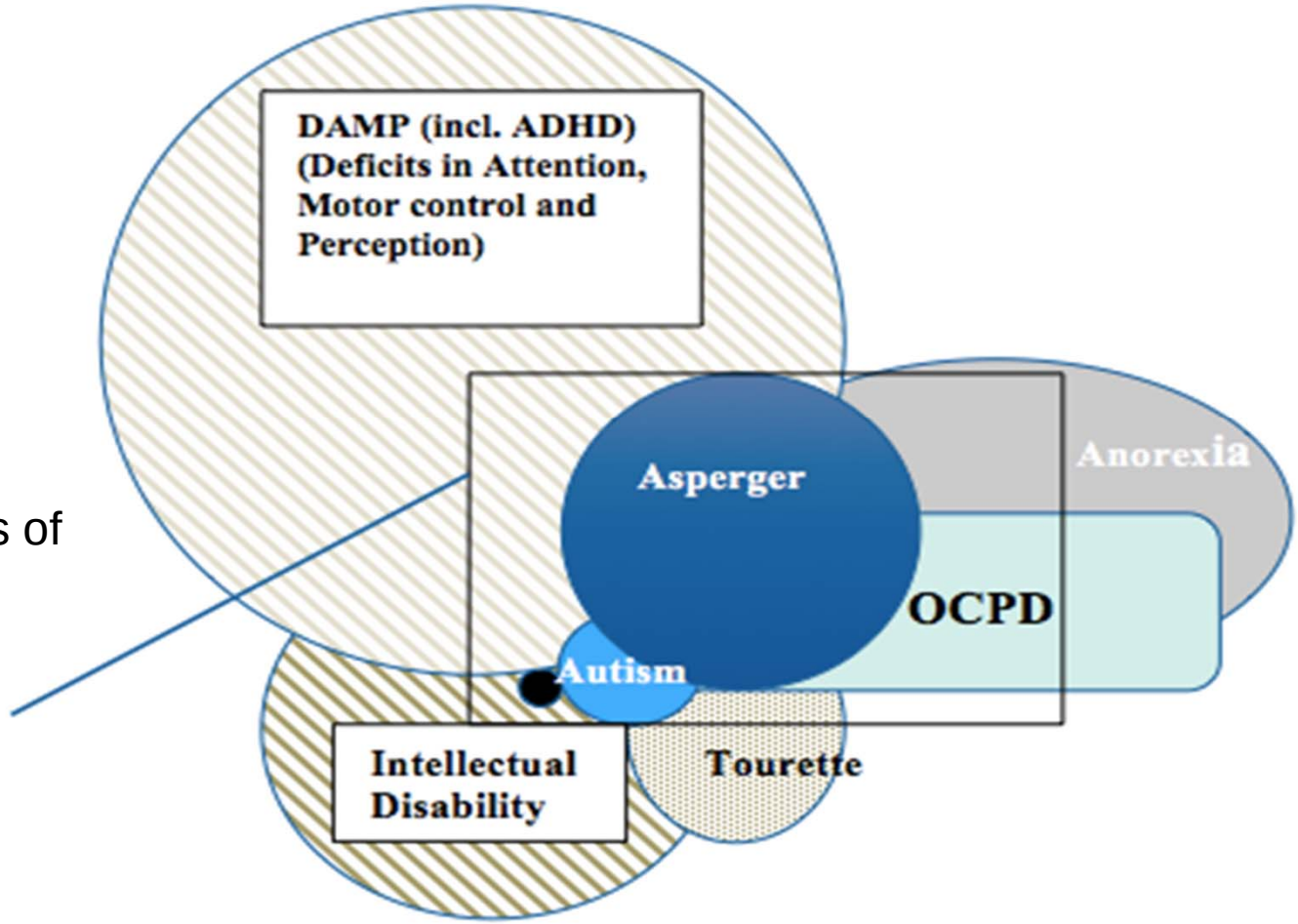
- **Major childhood onset symptoms either lasting more than 6 months or of extremely abrupt onset from one or more of the following domains are the markers of developmental disorder/ESSENCE; the symptoms lead to concern and "specialist" consultation**
  - **General development – delayed or very "patchy" mental development**
  - **Motor coordination – delayed gross or fine motor development**
  - **Perception/Sensory – hyper- or hyposensitivity to sensory stimuli**
  - **Communication/Language – delayed speech, few or no gestures**
  - **Activity/Impulsivity – too active or too passive, extremely impulsive**
  - **Attention – inattention, not listening, "not hearing", distractable**
  - **Social interaction/Reciprocity – little interest in adults, children, play, no response**
  - **Behaviour including stereotypic, insistence on sameness, tics, and OCD**
  - **Mood swings/emotional dysregulation – inability to control temper**
  - **Sleep – disrupted sleep-wake cycle, pavor, sleep onset, night waking problems**
  - **Feeding – food fads, selective/restrictive or consistent food refusal**

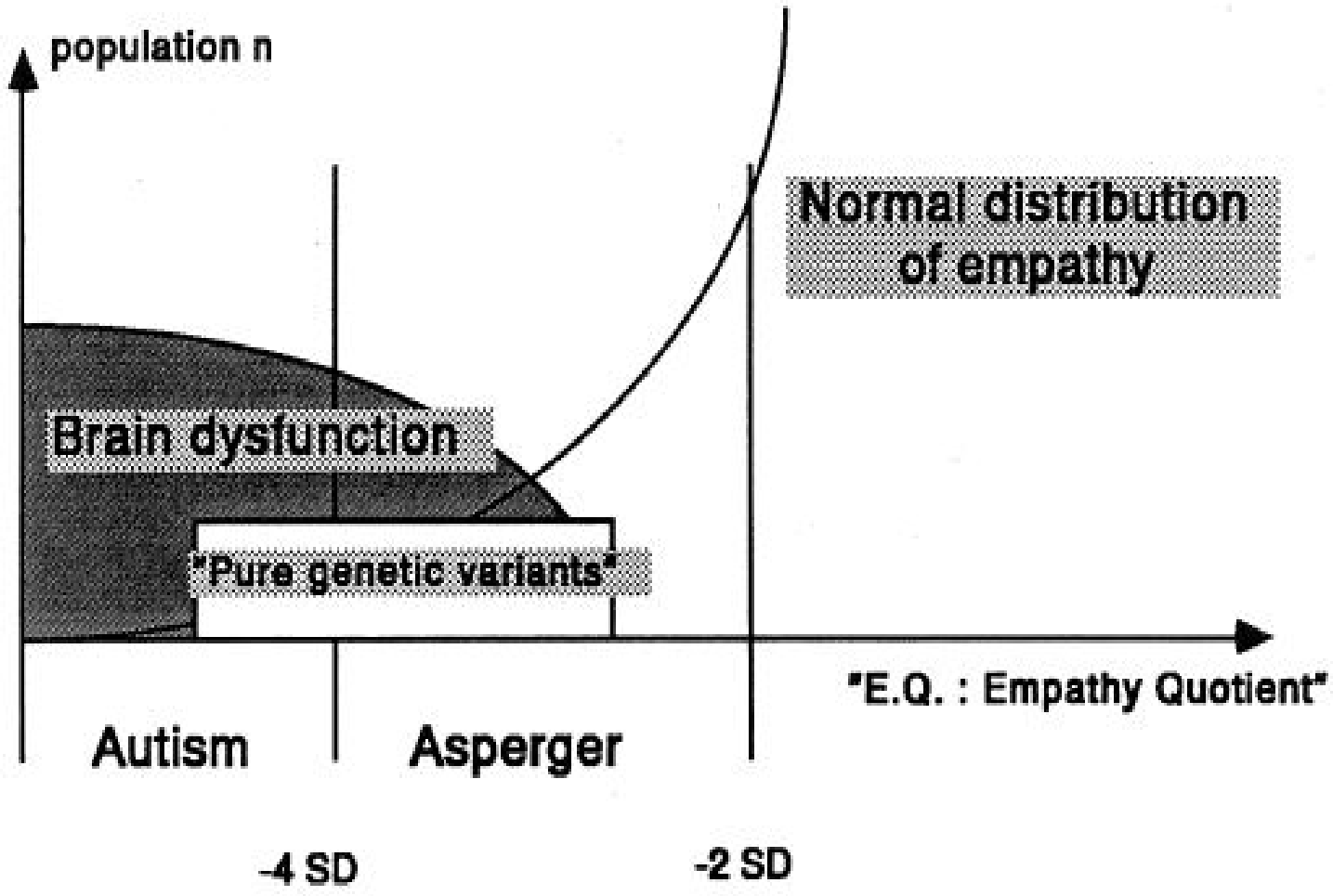
- Gillberg 2010, revised Gillberg 2013

# ASD: The Autisms

- ***The autisms*** are a group of multifactorially determined conditions. They ***ALWAYS coexist with other developmental/neurological problems in cases with early impairment (SLI, DCD, ADHD, IDD, tics, "OCD", epilepsy, other medical disorders). There are almost as many causes as there are cases*****. Cases with no comorbidity at all are not recognized or impairing early in life, or may be acknowledged as "loners", "nerds", "weirdos", "geniuses". *The prevalence of the phenotype is not increasing!*** Synapse and clock genes play a role in cases with impairment, but environmental factors (prematurity, fetal drug and toxin exposure, infections, trauma, cholesterol??, vitamin D deficiency?) contribute to or are associated with the clinical presentation in many cases and can cause autism in some instances. Variations of default network and unusual connectivity common finding. Impaired social facial perception in subgroup, related to specific brain areas. Abnormally high activation in subcortical system when constrained to look in the eye. Arousal and sleep problems important in subgroup. ***No sharp boundary between ASD and autistic traits or between autistic traits and "normality". You do not grow out of it, but impairment may increase or decrease and is, at least partly, an "effect" of comorbidities. No good evidence that base rate of autism symptoms has increased in the population, but diagnosis has gone through the roof (heavily overdiagnosed in some regions). No evidence that IBT changes long-term outcome.***
  - Iacoboni 2006, Buckner and Vincent 2007, Bourgeron 2007, Monk et al 2009, Gillberg 2010, Dinnstein et al 2010, Fernell et al 2011, Coleman and Gillberg 2012, Lundström et al 2012, Leblond et al 2012, Delorme et al 2013, Kocovska et al 2013, Zürcher et al 2013, Lundström and Gillberg 2014, Toro et al 2014, Lundström et al 2015; Posserud et al 2016, Hadjikhani et al 2016

Disorders of empathy




population n
Normal distribution of empathy
Brain dysfunction
"Pure genetic variants"
"E.Q. : Empathy Quotient"
Autism
Asperger
-4 SD
-2 SD

# Early symptoms of ASD (<5 years)

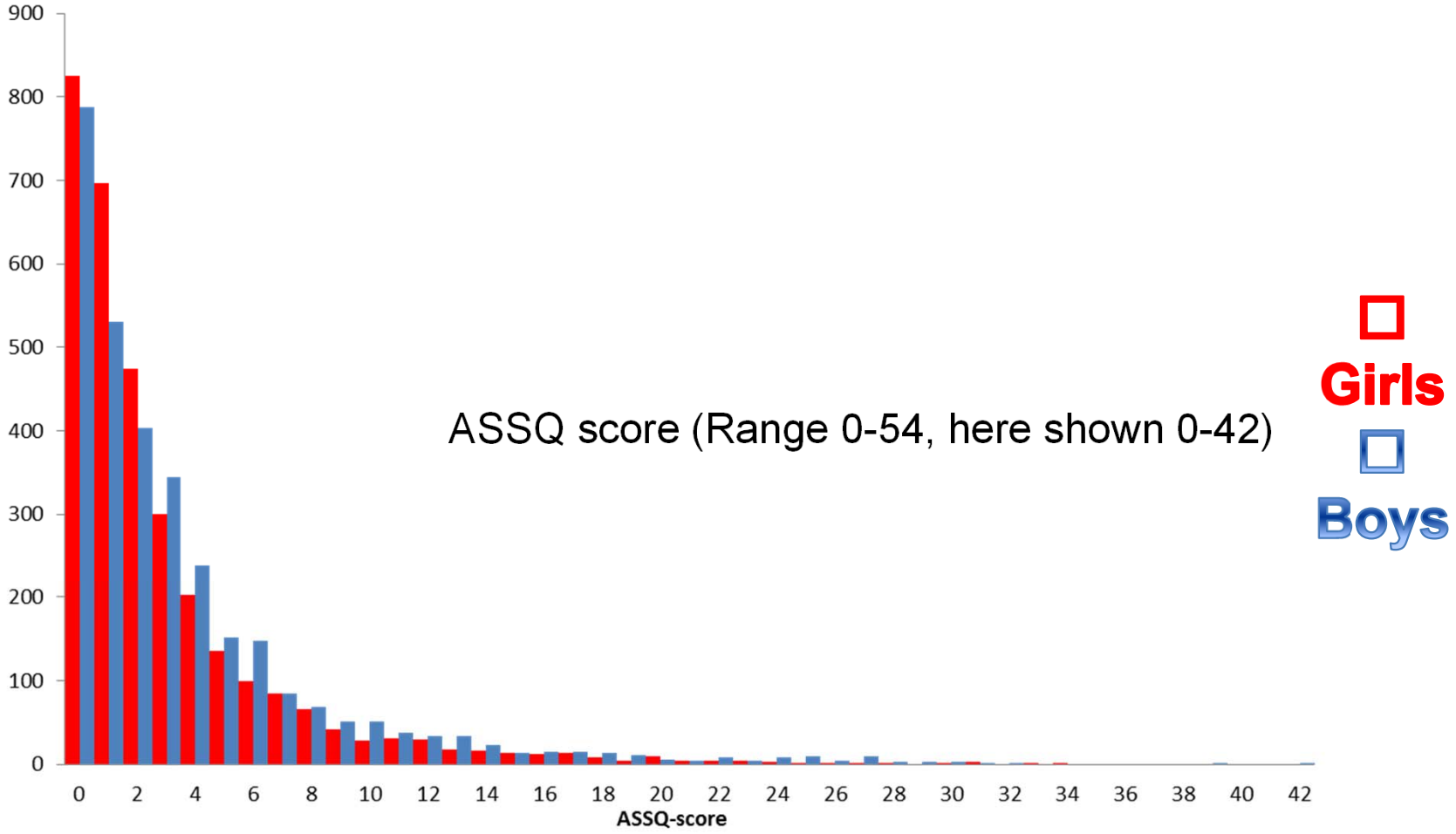
- **Motor control problems first year of life ("serious" face, relatively little smiling (but social smile can be elicited), strange movements from back to front, compartmentalised motor development, limpness, partial hypotonia) 50-100%**
- **Sensory-perceptual abnormalities/unusual preferences in 90-100%**
- **Behaviour problems (including insistence on sameness) in 90-100%**
- **Repetitive movements in 80-100%**
- **Language problems/pragmatic problems/strange voice in 90-100%**
- **No/little reaction to own name 30-100%**
- **No or limited initiation of joint attention ( => major social interaction problems), no pointing to attract attention 80-100%**
- **Hyperactivity and impulsivity (often extreme) in 40-50%**
- **Hypoactivity in 10-25%**
- **Sleep problems in 40%**
- **Food fads and other feeding problems in 50%**
- **Delayed general development in 20%**
- **Major mood swings in 10%**
- **One or several of the above could be presenting complaint**
  - Coleman and Gillberg 2012, Allely et al 2013, Höglund-Carlsson et al 2013, Barnevik-Olsson et al 2013, 2014, Hatakenaka et al 2016, Höglund-Carlsson et al 2016

# How many people are affected by ESSENCE?

- **At least 10% of school age children are or have been affected by "neuropsychiatric/neurodevelopmental disorders" (ESSENCE) (12% of boys, 8% of girls) - including ADHD, ASD, TS, CD, DCD, IDD – half this group "discovered" by age 6 years; many more than half this group will have persistent problems in adult life**
- **Overlap/"*Comorbidity*"/Co-existence *is the rule*; almost never "one problem only"**
- **When looking back: vast majority had symptoms <5 years**
- **Girls usually are not recognized until adolescence/adult age (and usually as non-ESSENCE)**
- **Half or (many?) more of all "chronic" adult psychiatric patients have had ESSENCE?**
- **Very large proportion of all frequent clinic attenders**

Gillberg 1983, Nylander et al 2009, Gillberg 2010, Kopp et al 2010, Gillberg 2013, Gillberg 2016

# PARENT REPORTS ON AUTISM SYMPTOMS (ASSQ) IN 6200 CHILDREN AGED 7-9 YEARS DATA FROM (LARGE GENERAL POPULATION) BERGEN CHILD STUDY



# Autism

- **Once (Kanner 1943, Rutter 1994), autism was considered a discrete disorder – the best and most clearly delineated in child psychiatry, also the most severe**
- **Gillberg (1983) found autistic traits to be very common in ADHD with DCD and found ASD in 0.7% of 7-year-olds in the mid 1970s**
- **Wing and Gillberg in the 1980s proposed a continuum/spectrum of autism**
- **Coleman and Gillberg proposed several different autism spectra (later "many different varieties of autisms") in the 1980s**
- **Gillberg (1991 and 1992) proposed that autism was on a spectrum with normally distributed empathy skills and that some variants even of the "disorder" could be considered mild, others moderate, yet others severe**
- **Gillberg (2010) proposed that autism is "hundreds of spectra" and a subgroup of ESSENCE**
- **Some (=marked) scorn because of this proposition**
- **Gillberg proposed Autism Plus as a clinically meaningful category (2013)**
- **DSM-5 diagnosis requires specification of IDD, SLD, medical, and severity**
  - Kanner 1943, Gillberg 1983, Coleman and Gillberg 1985, Wing 1979, Gillberg 1983, Gillberg 1992, Rutter 1994, Gillberg 2010, Coleman and Gillberg 2012, APA 2013, Gillberg and Fernell 2014

# Autism

- **L v Beethoven**
- **A Bruckner**
- **B Bartok**
- **E Satie**
- **HC Andersen**
- **S Kierkegaard**
- **A Conan Doyle**
- **I Kant**
- **L Wittgenstein**
- **A Einstein**
- **A Robbe-Grillet**
- **W Kandinskij**
- **P Klee**
- **E Hopper**
- **G Garbo**
- **D Springfield**
- **S Kubrick**
- **“ALL THE LONELY PEOPLE – WHERE DO THEY ALL COME FROM”**
  - **Gillberg 1992, Baron-Cohen 2003, Fitzgerald 2005, Wing 2011**

# From preschool to school and into adult life: what predicts what in autism?

- In virtually all studies of the outcome of autism, **language** disorder/problems/delay and low **IQ** predict poor outcome
- **Medical disorders**, including **epilepsy**, predict poor outcome
- **ADHD**/EF dysfunction in ASD predicts poor outcome
- Persistent NVLD in ASD predicts poor outcome
- Intervention may or may not predict the very long-term outcome, the jury is out, but we know it helps to do something (diagnosis + info) in the early years as regards intermediate-term outcomes
- ***But autism preschool or early school "load" in itself does not predict long-term outcome, maybe later persistence does***
- ***SO IT IS AUTISM PLUS THAT MATTERS***
  - Gillberg and Steffenburg 1987, Billstedt et al 2007, Cededrlund et al 2008, Fernell et al 2011, Eriksson et al 2013, Hagberg et al 2013, Helles et al 2014, 2016, Gillberg et al 2016

# Gender issues

- **Males are overrepresented but not as much as believed**
- **Autistic traits significantly more common in males in the population**
- **Autistic traits possibly much more common in gender dysphoria**
- **No strong evidence of link between autism and homosexuality**

- **IT IS LIKELY THAT ADOLESCENT AND ADULT FEMALES (AND SOME MALES) WHO HAVE HAD ASD AND/OR OTHER ESSENCE ALL THEIR LIVES ARE OFTEN MISDIAGNOSED AS SUFFERING FROM (ONLY):**
  - **"DEPRESSION",**
  - **"EATING DISORDER" AND/OR**
  - **"ANXIETY"**
  - **"BORDERLINE/OTHER PERSONALITY DISORDER/SELF-HARM"**
    - Kopp et al 2010, Lundström et al 2015, Shumer et al 2015, Davidsson et al 2016

# How should we proceed if we suspect AUTISM PLUS (but not if we suspect AUTISM ONLY)?

- **Observation** inside and outside clinic (if at all possible)
- Parent (and teacher) **questionnaires** plus follow-up interview – e.g. FTF (Five To Fifteen) or TTF (Two To Five), ATAC, SDQ, SNAP, ASSQ, most of these can probably be used for adults retrospectively
- **Parent interview** by doctor/psychologist
- **Medical/neurologic/psychiatric examination of child**
- **Hearing, vision, height, weight, head circumference, MPA screen**, genetic discussion, screening for thyroid and metabolic disorders, EEG sometimes (more often than currently), more if needed
- ***Assessment of intellectual functioning/neuropsychological/speech and language strengths and weaknesses, global assessment of adaptive functioning***
  - Kadesjö et al 2004, Hansson et al 2005, Coleman and Gillberg 2012, Miniscalco et al 2013, Gillberg 2013, Marinopoulou et al 2016

# How should we plan for best intervention in ASD PLUS, i.e. Autism with comorbidity (and ESSENCE more generally)?

- We need to recognize **all** the problems - **not just "the autism"**, "the ADHD", "the DCD", "the Tourette syndrome", "the IDD", "the SLI" and **all** interventions **must** be **individually** tailored **THROUGHOUT THE LIFESPAN**
- Parent "training" and education plan perhaps most important of all ("understanding the condition"), but parent ESSENCE problem needs to be taken into account (!)
- ADHD – whether or not combined with ASD, tic disorders, epilepsy or IDD - is ***usually*** responsive to treatment (meds and computer/cognitive training, possibly small effect of Omega-3)
- DCD is ***usually*** responsive to focused motor training regardless of comorbidity
- Epilepsy (possibly including "subclinical"), when present, ***should*** be treated as a top priority in all ESSENCE
- Sleep disorders ***sometimes*** responsive to melatonin or dose adjustment of other meds
- Violent behaviours/SIB ***can*** be responsive to low-dose neuroleptics or mood stabilizers
- Do not treat tics per se unless extreme
- Do not treat autism per se with meds (Bumetanide? Oxytocin?), overprescription of neuroleptics should be fought
- **Psychoeducation, communication enhancement, ESSENCE-friendly environment ("understanding the condition"), school and work place "adjustment", and behavioural approaches - sometimes only possible with medication - first and foremost throughout life**

# AUTISM PLUS and ESSENCE preliminary conclusions

- ***ESSENCE (not autism per se) is an extreme risk factor*** for adolescent/adult social exclusion, academic failure, other school adjustment problems, problems in the work-place antisocial personality disorder (and depression/anxiety, drug abuse, and criminality) – and for "non-handicapping" autistic traits?
- We still know VERY little about early intervention
- ***The* OVERFOCUS *on ASD ONLY in young children is possibly a big mistake; but AUTISM PLUS HAS HUGE IMPLICATIONS***
- For some ESSENCE we can screen and intervene early
- All advanced societies need to increase/spread knowledge about ESSENCE, including ADHD and IDD, **not just ASD**
- In research following children over time ***all aspects of ESSENCE need to be taken into account – with screeners such as ESSENCE-Q, A-TAC or TTF***

# Final conclusions: What we need to think about when setting up assessment routines

- **AUTISM PLUS is but one of a group of ESSENCE that overlap *genetically*, *symptomatically* and as regards *brain* dysfunction/*variation*, *environmental* factors also play a role, but it is unclear how much of the variance they account for**
- **AUTISM PLUS (i.e. with comorbidity) is a severe disorder, AUTISM ONLY?**
- **ASD persists into adult life (as do most other ESSENCE), re-assessments needed**
- **ADHD is common (c. 5%), ASD is relatively common (c. 1%)**
- **Other psychiatric disorders/problems/academic failure emerge or become "diagnosable" over time – these are the diagnoses that adult psychiatrists will make**
- **Autism *in itself* has *different* outcome, not necessarily poor, current focus on autism *only* in screening, assessment and intervention programs a big mistake**
- **IDD has "poor" outcome, ADHD probably has worse outcome (including obesity, pain syndrome, substance use, MCI?) than ASD "in itself", SLI may also have partly "poor outcome"**
- **Gender dysphoria needs to receive more attention in clinical practice**
- **Girls still usually missed or misdiagnosed**

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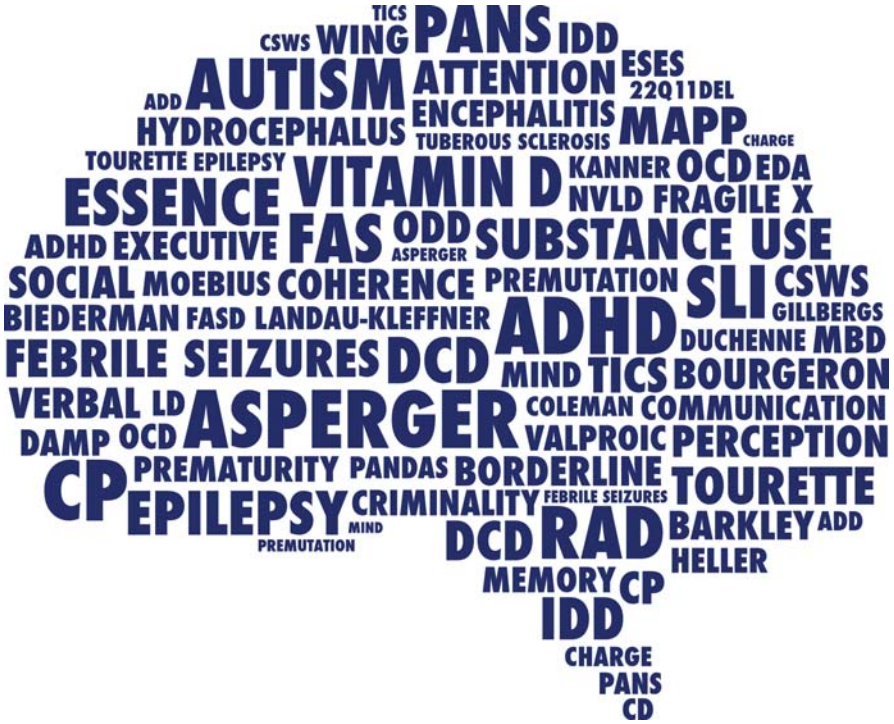
# ESSENCE CONFERENCE

## 10-11 APRIL 2018

## GOTHENBURG, SWEDEN

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