How severe is autism – really?

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How severe is autism – really?

• *Nature never draws a line without smudging it*
  – One of Lorna Wing’s favourite quotes
TEACHER REPORTS ON AUTISM SYMPTOMS (ASSQ)
N= 9500+ CHILDREN AGED 7-9 YEARS DATA FROM Bergen Child Study (BCS)

ASSQ score (Range 0-54, here shown 0-42)
PARENT REPORTS ON AUTISM SYMPTOMS (ASSQ)
N= 6200+ CHILDREN AGED 7-9 YEARS DATA FROM BCS

ASSQ score (Range 0-54, here shown 0-42)
Parent A-TAC Swedish twins n=19131

A-TAC score (Range 0-18, here shown 0- ≥8,5)
Autism

• Once (Kanner 1943, Rutter 1994), autism was seen to be 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
• Wing and Gillberg proposed a continuum/spectrum of autism
• Coleman and Gillberg proposed several different autism spectra (later “many different varieties of autisms”)
• Gillberg (1991) 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”
• Some scorn because of this proposition (Rutter in particular)
Autism

• HC Andersen
• A Conan Doyle
• A Robbe-Grillet
• I Kant
• L Wittgenstein
• A Einstein
• B Bartok
• E Satie
• A Bruckner
• W Kandinskij
• P Klee
• E Hopper
• S Kubrick

• “ALL THE LONELY PEOPLE – WHERE DO THEY ALL COME FROM”
Neurodevelopmental disorders/ESSENCE

• **ESSENCE - Early Symptomatic Syndromes Eliciting Neurodevelopmental Clinical Examinations**
  
• Predictors of academic failure, substance misuse, psychiatric disorder, empathy problems, and antisocial lifestyle later in life
  
  – ADHD with or without **ODD/CD** (Oppositional Defiant Disorder/Conduct Disorder) 5-7%
  
  – SLI (Language disorder) 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%
  
  – BPS (Behavioural Phenotype Syndromes, including FAS and VAS) 1%
  
  – EP (Epilepsy syndromes and other neurological disorders (HC, CP, neurometabolic): Landau-Kleffner Syndrome, CSWS, FS+, FS? 0.6%
  
  – RAD (Reactive Attachment Disorder)? 0.5-1%
  
  – PANS (Pediatric Acute-onset Neuropsychiatric Syndrome)? 0.1%
“The autisms”: What is ASD? What is autism?

• The autisms are a group of multifactorially determined conditions, that **ALWAYS coexist with other developmental/neurological problems in cases with impairment** (SLI, DCD, ADHD, IDD, tics, “OCD”, epilepsy, other medical disorders), **and 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”. Synapse and clock genes play a major role in cases with impairment, but environmental factors (prematurity, fetal drug and toxin exposure, infections, trauma, vitamin D deficiency) contribute to or are associated with the clinical presentation in many cases and can themselves cause autism in some instances. Abnormalities/variations of default network and decreased connectivity almost universal finding. Impaired social facial perception in large subgroup, related to specific brain areas. 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 usually an “effect” of comorbidities**

What is ADHD?

• ADHD is largely genetic, and ALMOST ALWAYS co-existing with other problems (ODD, DCD, IDD, tics, “OCD”, ASD, anxiety, epilepsy) in cases with clinical impairment; similar symptoms result after various types of environmentally caused brain dysfunction with or without genetic predisposition ( prematurity, fetal drugs and toxins, infections, vitamin D deficiency??). Atypical brain development in children with ADHD. Growing evidence that dopamine-dependent reward systems in the brain deviant in ADHD. Default network activity and connectivity variation/abnormality implicated. Executive function poor, but may not be caught on specific tests. Arousal and sleep problems important in subgroup. Failure to perceive errors typical of many. ADHD and ASD are related in some families. Bipolar disorder and ADHD related in other (?) families. Tourette syndrome and ADHD related in some (yet other??) families. No sharp boundary between ADHD and “normality”. You do not grow out of it, but impairment may increase or decrease

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
  – Motor coordination/Perception-Sensory
  – Communication/Language
  – Activity/Impulsivity
  – Attention
  – Social interaction/Reciprocity
  – Behaviour including insistence on sameness, tics, and OCD
  – Mood swings/emotional dysregulation
  – Sleep
  – Feeding

  - Gillberg 2010, revised Gillberg 2013
Early symptoms of autism (<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/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
Early symptoms of ADHD/ODD (<5 years)

- **Motor** control problems first year of life 50-100% (but possibly different from those in ASD?)
- Sensory-perceptual abnormalities in 50-100%
- **Language** problems/pragmatic problems in 50%
- Inability to control temper and other **mood** problems in 50-100%
- **Behaviour** problems in 50-100%
- Inattention/brief focused **attention**/non-purposeful attention 100%?
- Hyperactivity (rarely extreme) in 10-30%
- Hypoactivity problems and inattention in 25%
- Major **sleep** problems in 40%
- Delayed general development in 15%
- Major mood swings in 5%
- One or several of the above could be presenting complaint
  — Wilson et al 2013, Gillberg 2013
How many people are affected by ESSENCE?

• At least 10% of children under 18 years of age 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; 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?

From preschool to adult life

• If at least 10% of children under 18 years of age are or have been affected by ESSENCE (12% of boys, 8% of girls), then this is

• **Clearly a public health problem**

• For some of the subgroups we think we “know what to do” (ADHD, autism, IDD, SLI)

• For a few of the subgroups we “know that it helps” (ADHD, autism)

• For some of the subgroups there are good screening instruments that we know work in real life, with high PPV and sensitivity (autism, SLI)

• So, really, for some of these, should we not screen? (ADHD/ODD, ASD, SLI)
From preschool to 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 in ASD predicts poor outcome
• NVLD in ASD predicts poor outcome
• Intervention may or may not predict outcome, the jury is out

• But, in most studies, autism “load” in itself does not predict outcome
• Two thirds of males with Asperger syndrome have “medium to good outcomes”

If we screen for ESSENCE: what will happen after?

• After screening, is there a treatment? Is there a cure?
• Not really, except in rare circumstances/cases
• *Even with no cure, there is help to be had in almost all cases*
• Psychoeducation, parent expertise (“parent training”), adaptive skills training, ESSENCE-friendly environment (including “everybody has to understand the basics of ESSENCE”, early reading training, executive function training/martial arts?), individualised programmes, remedial education, and, sometimes medication (ADHD, for “comorbidities” of ASD, but not really for ASD “per se”)*
How should we proceed if we suspect ESSENCE?

• Observation outside clinic, if at all possible (pre-school/school)
• Observation – inside and outside clinic - of “joint attention”, “attention/listening”, “behaviour”, “communication”, “activity”, “impulsivity”, “emotional regulation”
• Parent (and teacher) questionnaires plus follow-up interview – e.g. FTF (Five To Fifteen) or TTF (Two To Five), A-TAC, SDQ, SNAP, ASSQ
• Parent interview by doctor/psychologist
• Medical/neurologic/psychiatric examination of child
• Hearing, vision, height, weight, head circumference, genetic discussion, screening for thyroid and metabolic disorders, EEG sometimes (more often than currently)
• Assessment of intellectual functioning/neuropsychological strengths and weaknesses
How should we plan for best intervention in ESSENCE?

- We need to recognize all the problems, not just “the autism”, “the ADHD”, “the DCD”, “the Tourette syndrome”, and all interventions must be individually tailored
- Parent “training” and education plan perhaps most important of all (“understanding the condition”), but parent ESSENCE problem (whenever present) needs to be taken into account (!)
- ADHD “per se” - or when combined with ASD, tic disorders, epilepsy or IDD - is usually responsive to treatment (meds and computer 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
- Psychoeducation, communication enhancement, ESSENCE-friendly environment (“understanding the condition”) and behavioural approaches – sometimes only possible with meds - first and foremost throughout life
100 girls with social and/or attention deficits

• Two of 100 girls (3-18 years) referred for social and/or attention deficits to a specialized neuropsychiatric clinic had received a diagnosis of ASD before coming to the clinic. In the other 98, a diagnosis of ASD or ADHD had not been considered
• 47 of the girls had ASD (many of whom also met criteria for ADHD)
• 47 other girls had ADHD ”only”
• Virtually all of these girls had been diagnosed as having depression, anxiety or ”family relationship problems”
  – Kopp et al 2010
Autism, ADHD and tics/OCD in anorexia, bulimia and obesity

• More than half of all adult anorexia/bulimia patients in a specialist clinic had one or more of ASD, ADHD or Tourette’s (0% had been considered from the point of view of autism, ADHD or tic disorders)

• ADHD extremely common in obesity in children – and ADHD treatment sometimes helps both conditions, obesity treatments help neither
ASD (and ADHD) in schizophrenia and PD

• Many with diagnosis of schizophrenia have developmental and symptom history consistent with ASD (with or without ADHD)
• Large proportion of men with Asperger syndrome meet criteria for personality disorder
• It is likely that adult psychiatric patients with ASD will get diagnoses of schizophrenia and personality disorder (particularly “paranoid”?) rather than ASD (with or without ADHD)
Conclusions

• ASD and ADHD are but two of a group of ESSENCE that overlap genetically, symptomatically and as regards brain dysfunction/variation

• ASD and ADHD (and other ESSENCE) are clearly genetically based in many cases, but environmental factors play an important role (to be studied: how important)

• Both disorders persist into adult life (as do most other ESSENCE)

• 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 early screening and intervention programmes probably a big mistake

• IDD has “poor” outcome, SLI may also have partly “poor outcome” ADHD probably has worse outcome (including obesity) than ASD “in itself”,

• Girls missed or misdiagnosed

• Early diagnosis makes a difference
Some early “warning signals”

• **Example: delayed language at 2.5 years**
  – Screen takes no more than 5 minutes (deviance if comprehension difficulties, fewer than 50 communicative words, major dysarticulation)
  – About 2-6% of all children screen positive and have “confirmed” language delay at 2.5 years
  – **Screen positive and confirmed language delay at 2.5 years => 70% have developmental disorder/ESSENCE at age 7 years (ADHD, ASD, IDD, DCD), virtually all have remaining speech-language problems or dyslexia**
  – i.e. all children with language delay at 2.5 years need to be followed carefully and vast majority will need services
  – **Language delay will not “show up” without screening**
Some other early warning signals: ASD?

- **Example: suspected ASD under age 3 years**
  - 28 children followed from under age 3 years with suspected autism: 75% met criteria for autistic disorder at age 6 years, and remainder had other neuropsychiatric diagnosis (other ASD, ADHD, IDD)
    - Gillberg et al 1990
  - 208 children with autism diagnosis made by clinicians at age 0-4 years: 52% met criteria for autistic disorder at follow-up, 39% met criteria for other autism, 9% had other ESSENCE diagnosis (ADHD, IDD) - prevalence of autism in this age group 0.6%
    - Fernell et al 2009, 2011
  - **Autism diagnosis around age 2-4 years highly stable in 90% of cases, virtually no “over-diagnosis”, many Asperger cases missed**
Yet another early warning signal: ADHD?

• Example: suspected ADHD under age 5 years
  - 131 children 3-7 years of age with ADHD: 60% met criteria for oppositional defiant disorder (ODD) at age 3, 4, 5, 6 and 7 years, only 10% of all with ADHD had no symptom of ODD; ODD symptom (often loses temper) most common individual symptom of all in ADHD-group
  - ADHD diagnosis around age 3-7 years confers highly stable risk of ODD in 60% of cases – risk of violence, aggression criminal, antisocial behaviour, and SUD in adult age much increased in this group
  - ADHD with DCD predicts language problems, autistic features and academic failure
    - Kadesjö & Gillberg 2000, Kadesjö et al 2003
ESSENCE final conclusions part I

• **ESSENCE is not a diagnosis!!!**

• ADHD, ASD, Tourette syndrome, IDD, DCD etc. overlap to a marked degree throughout life and are often not clearly separable under age 5 years

• All children presenting with major and impairing ESSENCE symptoms need to be followed up – ESSENCE clinics of the essence!

• Even though *refined diagnosis is needed in all cases*, at early stages ESSENCE may be the only “safe” label **BUT IT IS NOT A DIAGNOSIS**

• Never proclaim: “He/she will grow out of it” - no evidence that this is likely to happen in more than a small minority of cases
ESSENCE final conclusions part II

• **ESSENCE (not autism per se) is an extreme risk factor** for adolescent/adult social exclusion, academic failure, 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**

• For some ESSENCE we can screen and intervene early

• Therefore, all advanced societies need to increase and spread knowledge about ESSENCE, including ADHD and IDD, **not just about 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/FTF**
ESSENCE and ASD

• Autism per se is probably not a ”severe disorder”; autism makes people ”different”
• Together with other ESSENCE it can sometimes be a devastating problem unless intervened for at an early stage
• Autism ”+” is a problem for child health, psychiatry, pediatrics, general medicine, and neurology
• Autism ”per se” is worth thinking about, but maybe more as a matter of enormous theoretical interest than as a clinically impairing problem in its own ”right”