



# The power of diagnosis: thinking pragmatically

- Ginny Russell



Autism and Neurodiversity

# Exploring Diagnosis

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## Who we are

We are a research team based at Egenis, part of the University of Exeter in the UK. Our research explores the role that diagnosis plays in society and in medicine, using diagnosis of autism spectrum disorder as a case study.

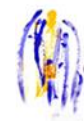
On the way, we aim to celebrate and value the abilities and

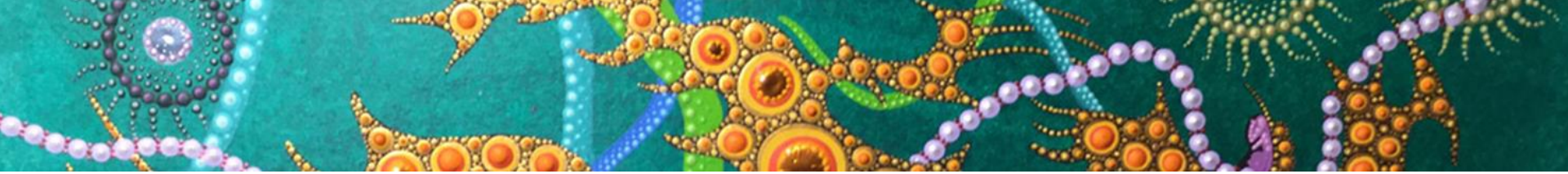




# Structure of talk

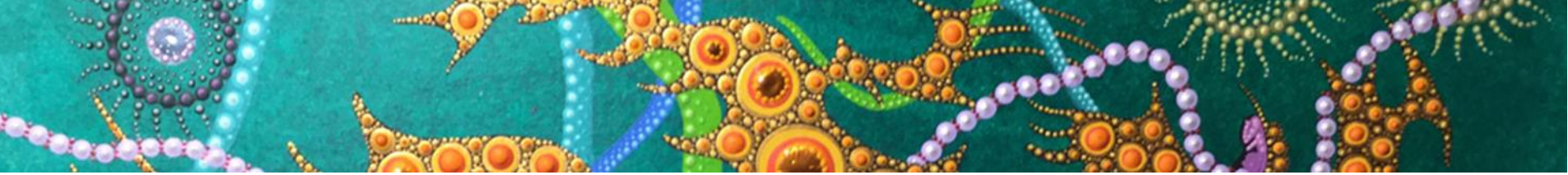
1. Sociology of diagnosis versus clinical perspectives
2. Assessing autism as a diagnosis
3. Autism as a 'pragmatic construct'



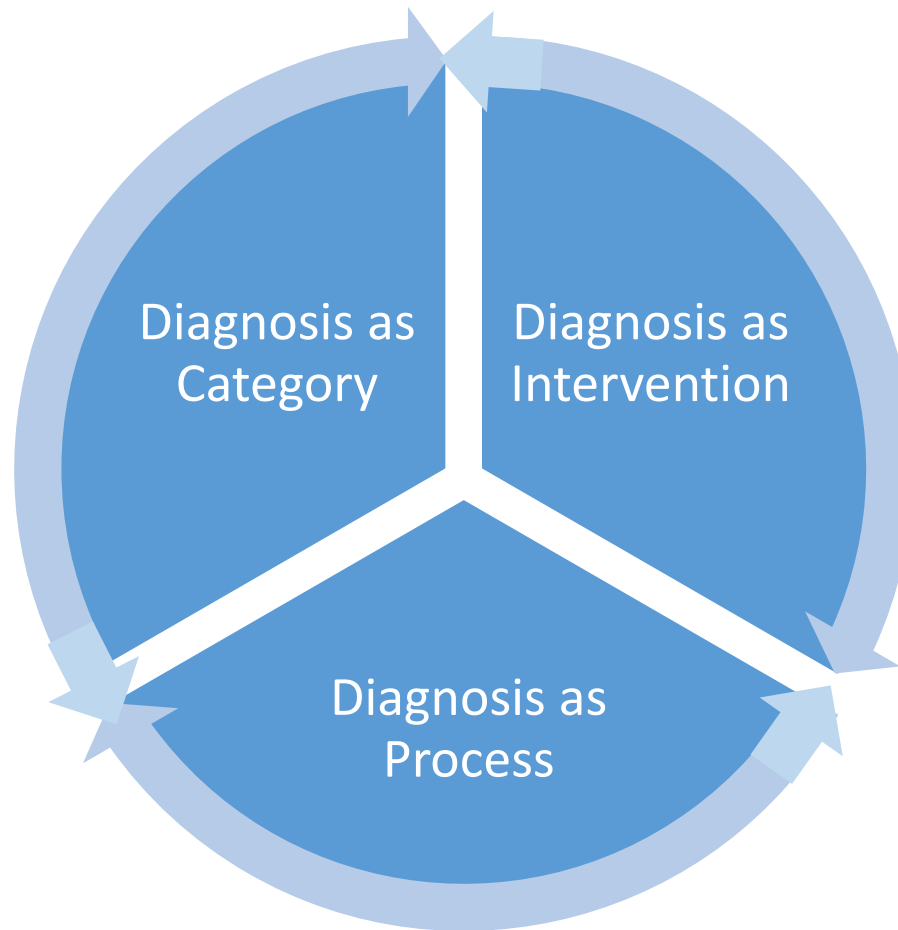


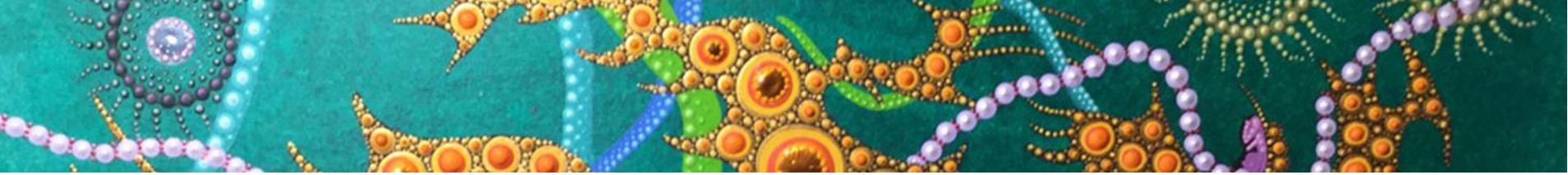
# Functions of diagnosis in society

1. Legal
2. Clinical communication
3. Determines treatment pathways
4. Social identity
5. Medical role and authority
6. Defines 'normal'
7. Determines service commissioning
8. Object for organising research, services, resistance



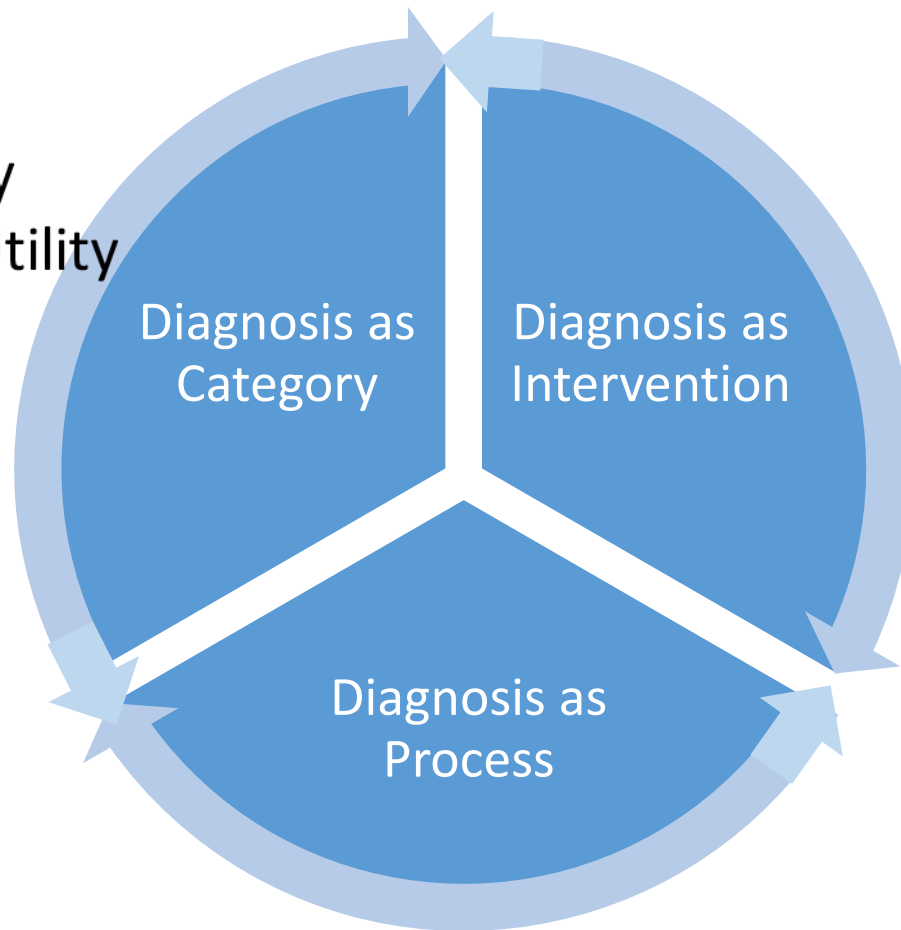
# Sociology of Diagnosis

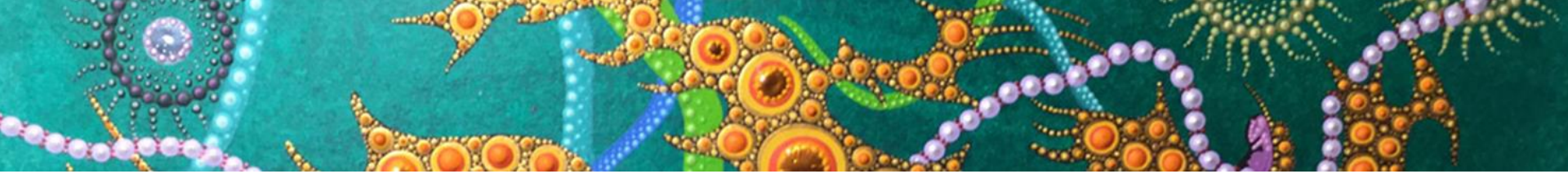




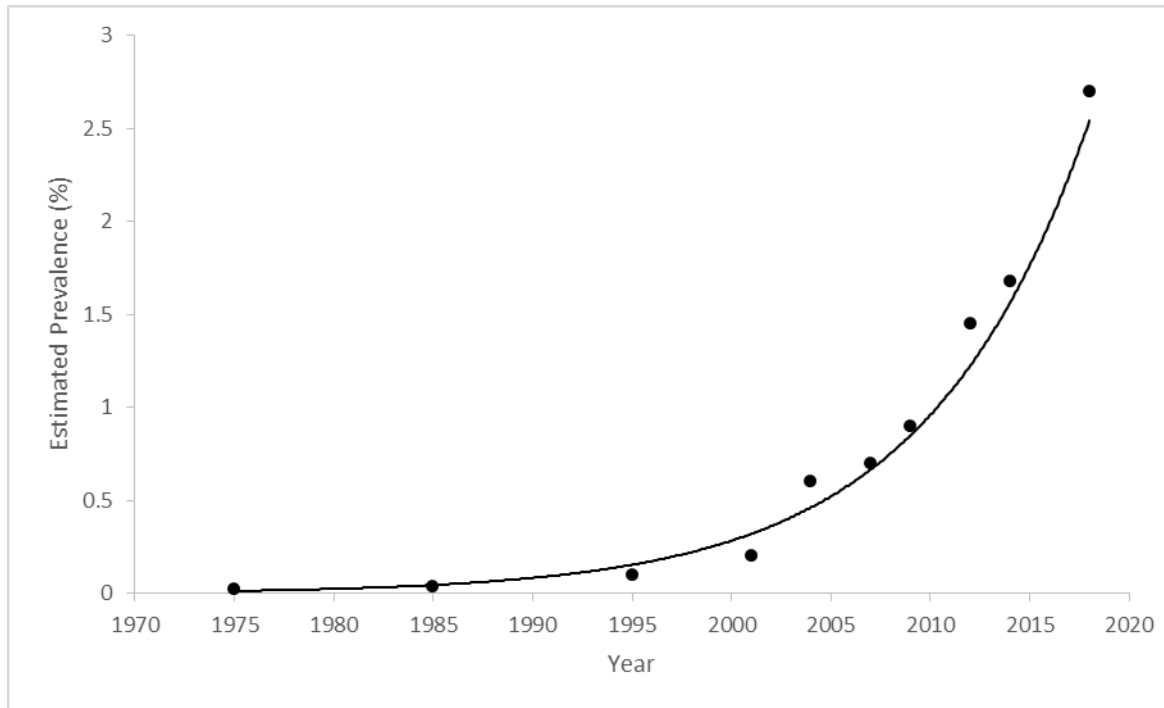
# Medical assessment of diagnosis

1. Validity
2. Reliability
3. Clinical Utility





# Validity of autism as a diagnostic category



Weintraub et al. (2011) The Autism Puzzle *Nature*, ; Center for Disease Control (2014) data



# Validity of autism as a diagnostic category

## 1. Challenges: Splitters (into subtypes)

By genetic profiles (no co-inheritance of core triad)

By genetic syndromes (idiopathic vs syndromic)

By cognitive traits

By neural differences

By IQ, HFA vs SA

By gender, (female autism phenotype)

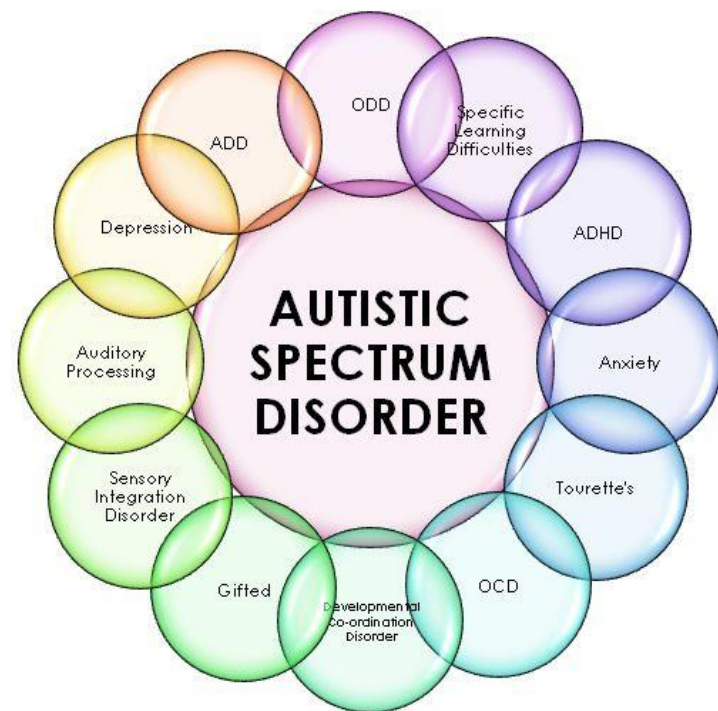
By sensory processing differences



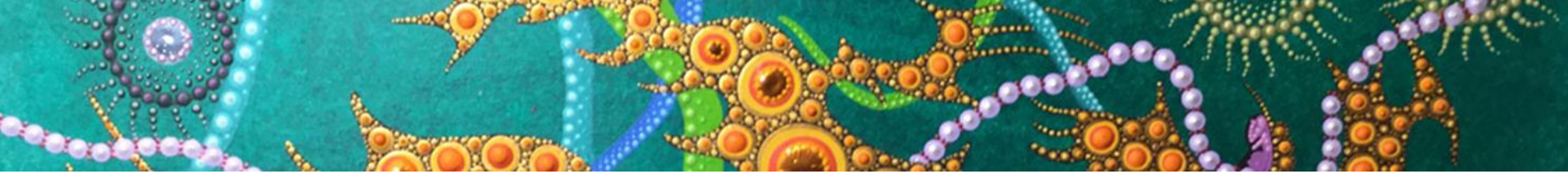


# Validity of autism as a diagnostic category

2. Lumpers: Co-occurring conditions
3. ID
4. Anxiety
5. SLD
6. PDD
7. Depression
8. ADHD
9. DCD
10. Epilepsy

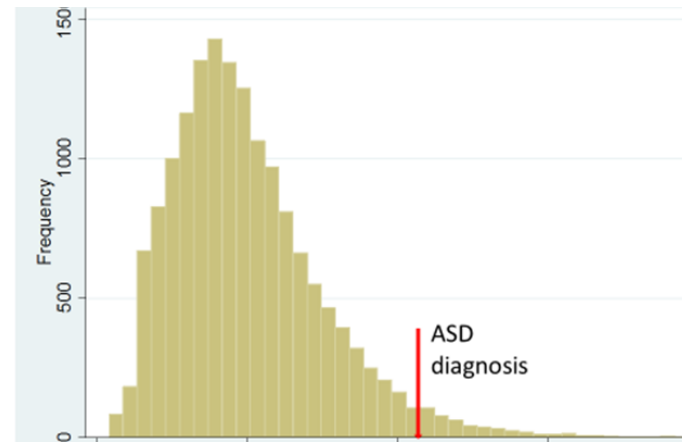


Gillberg, C. (2010) The ESSENCE in child psychiatry: *Research in Developmental Disabilities*  
Simonoff et al. (2008) Psychiatric Disorders in Children With Autism Spectrum Disorders *JAACAP*



# Borderline and uncertain cases

## 1. Dimensional



2. Autism may not be clearly distinguishable from 'normal' or atypical behaviours in children (and adults) without.
3. Co-occurring conditions mean differential diagnosis problematic

Russell et al. (2015) Changes in diagnosis rates of autism *BJPsyche Open*

Steer et al. (2010) Traits contributing to the autism spectrum *Plos-one*



# Reliability of autism as a diagnosis/ category

## 1. Inter-rater reliability good

Rater Groups	Autism vs. non-PDD		
	$\kappa$	No. of cases	Clinical significance
Experienced vs. Experienced	1.00	44	E
Psychologist vs. Psychiatrist	1.00	38	E
Inexperienced vs. Inexperienced	1.00	14	E
All reliability raters	0.95	103	E
Experienced vs. Inexperienced	0.89	42	E

## 2. Replicability

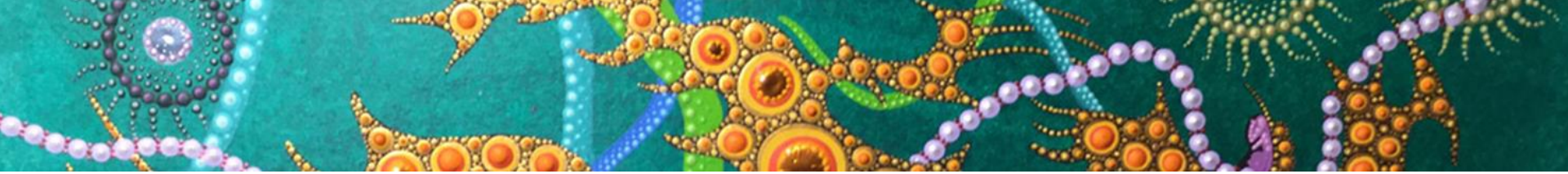
- Varies by region
- Varies by resources
- Varies by time

Elsabbagh et al. (2012) Global Prevalence of Autism and Other Pervasive Developmental Disorders Autism Research

Liu , King & Bearman (2010) Social Influence and the Autism Epidemic American Journal of Sociology

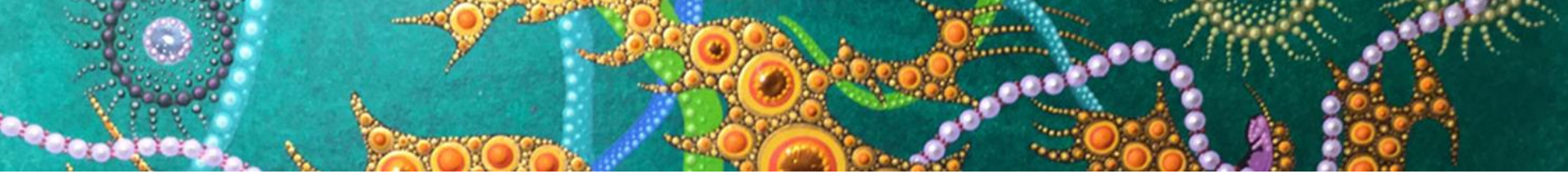
Mandell and Palmer (2005) **Differences Among States in the Identification of Autistic Spectrum Disorders** Arch Pediatr Adolesc Med.

Klin et al. (2000) Interrater Reliability of Clinical Diagnosis and DSM-IV Criteria for Autistic Disorder: JADD.



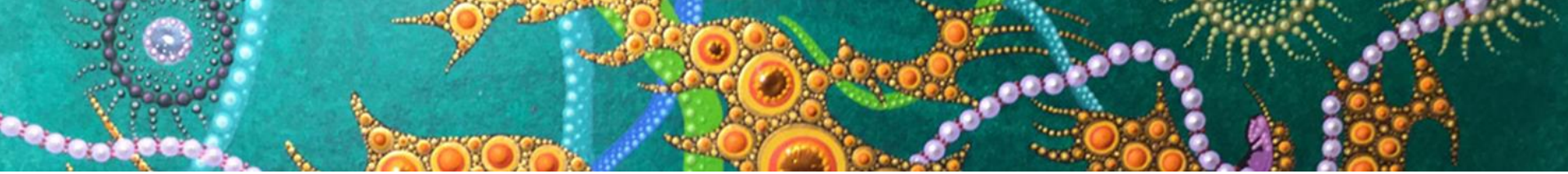
# Utility of autism as a diagnostic category

1. For clinicians
2. For researchers
3. For educators
4. For people so diagnosed
5. For parents
6. For peers
7. For insurers



# Medical assessment of diagnosis

1. Validity
2. Reliability
3. Clinical Utility
4. Personal Utility



# Disadvantage of diagnosis

1. Stigma
  - Varies by region
  - Varies by contact
  - Varies by time
  - Varies by awareness/education
2. Brain based explanation -exculpation
3. The 'new' normal
4. The autism 'lens'

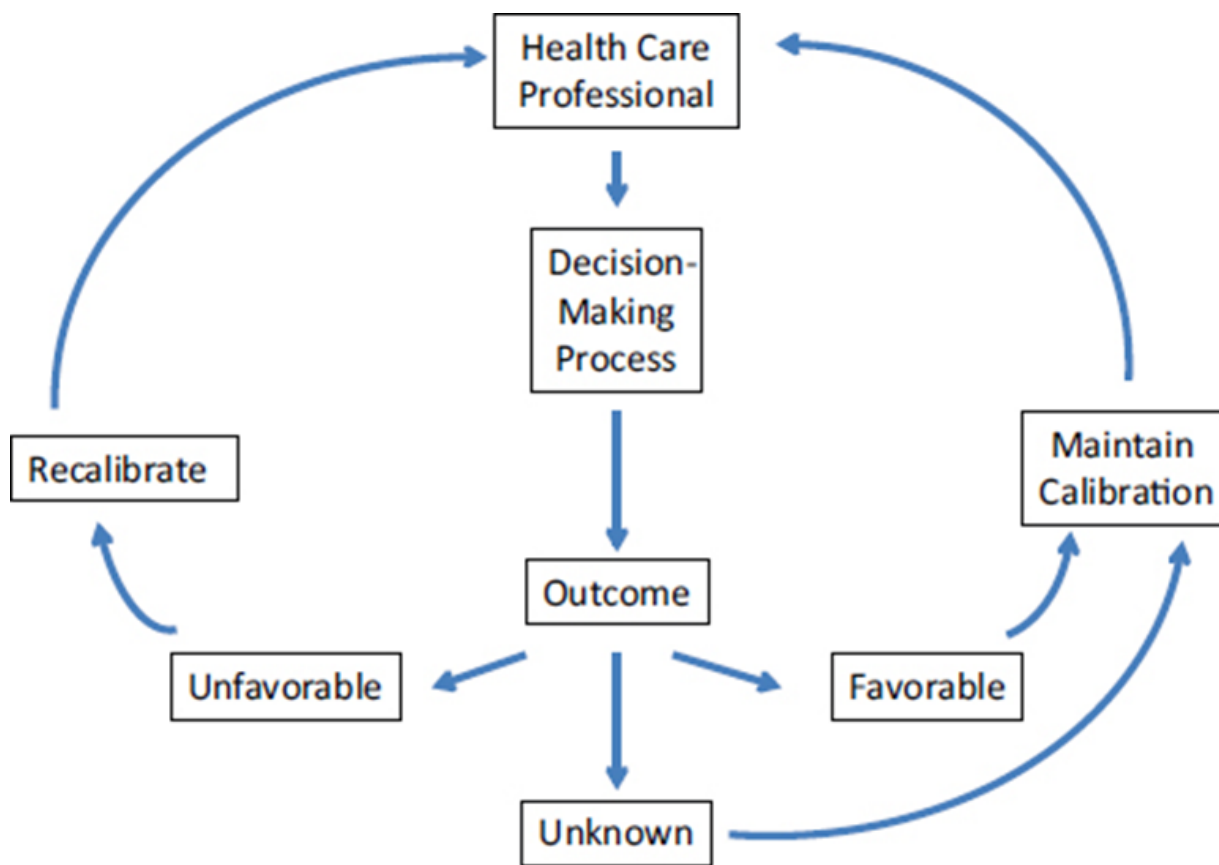


# Autism diagnosis as a pragmatic construct

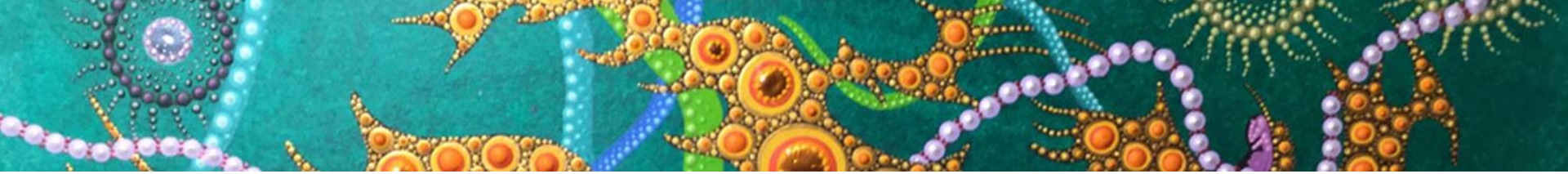
1. Borderline cases where another dx, or no dx is considered
2. We are never going to definitively know if a person does or does not have autism, as autism is a moving target.
3. Instead of trying to ask is it valid category- does this person really 'have autism' -- ask 'is this a useful diagnosis?'
4. Prioritise clinical utility, and 'personal utility'- does it promote best outcome for child (or adult)? For carers?
5. Who decides and who benefits?



# Autism diagnosis as a pragmatic construct







# Autism diagnosis as a pragmatic construct

Lorna Wing – ‘It is impossible to draw sharp lines between autistic and not really autistic’

Instead

‘ask what are impairments, difficulties and skills and what can we do to help him?’

Wing (1972)