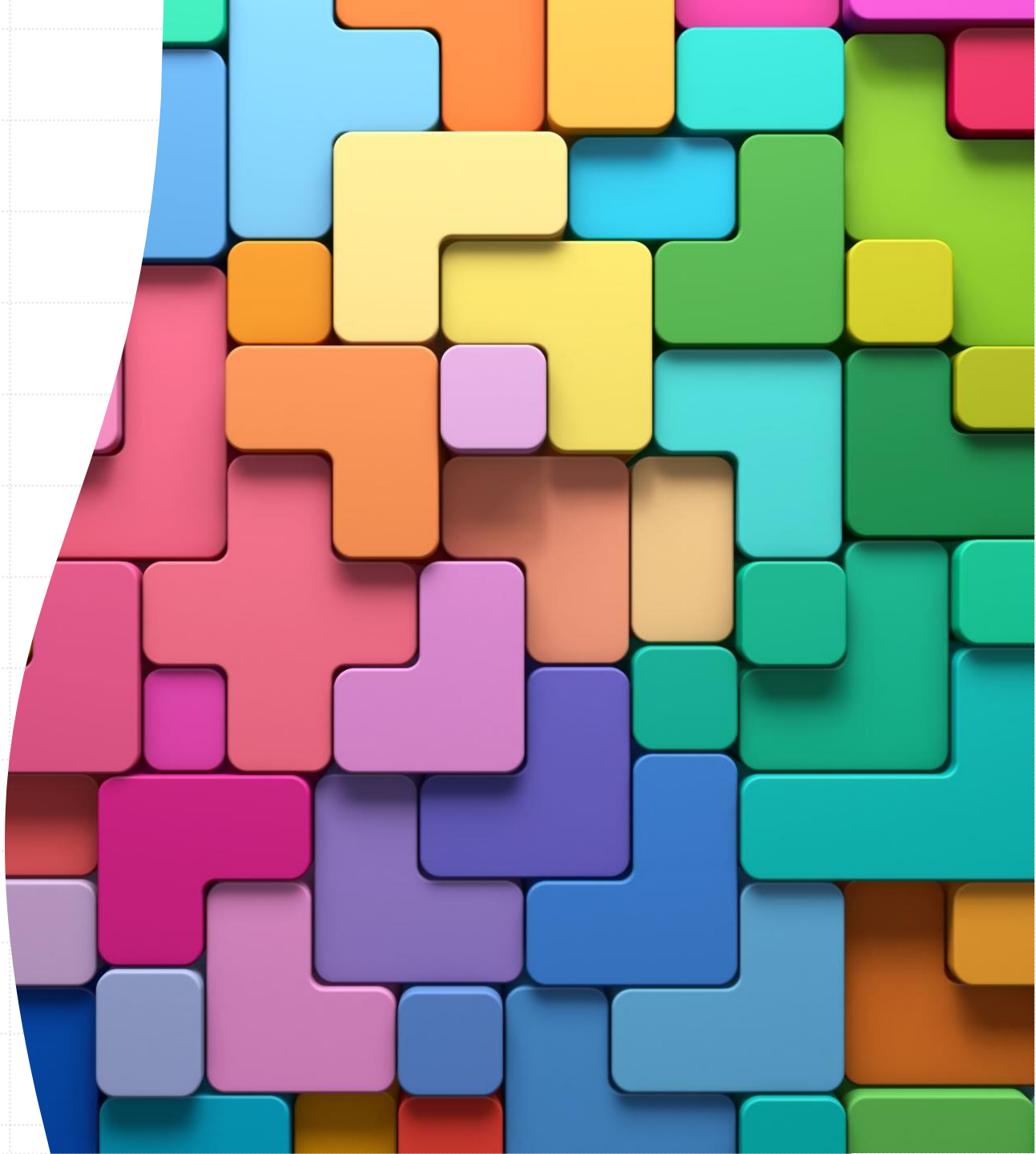
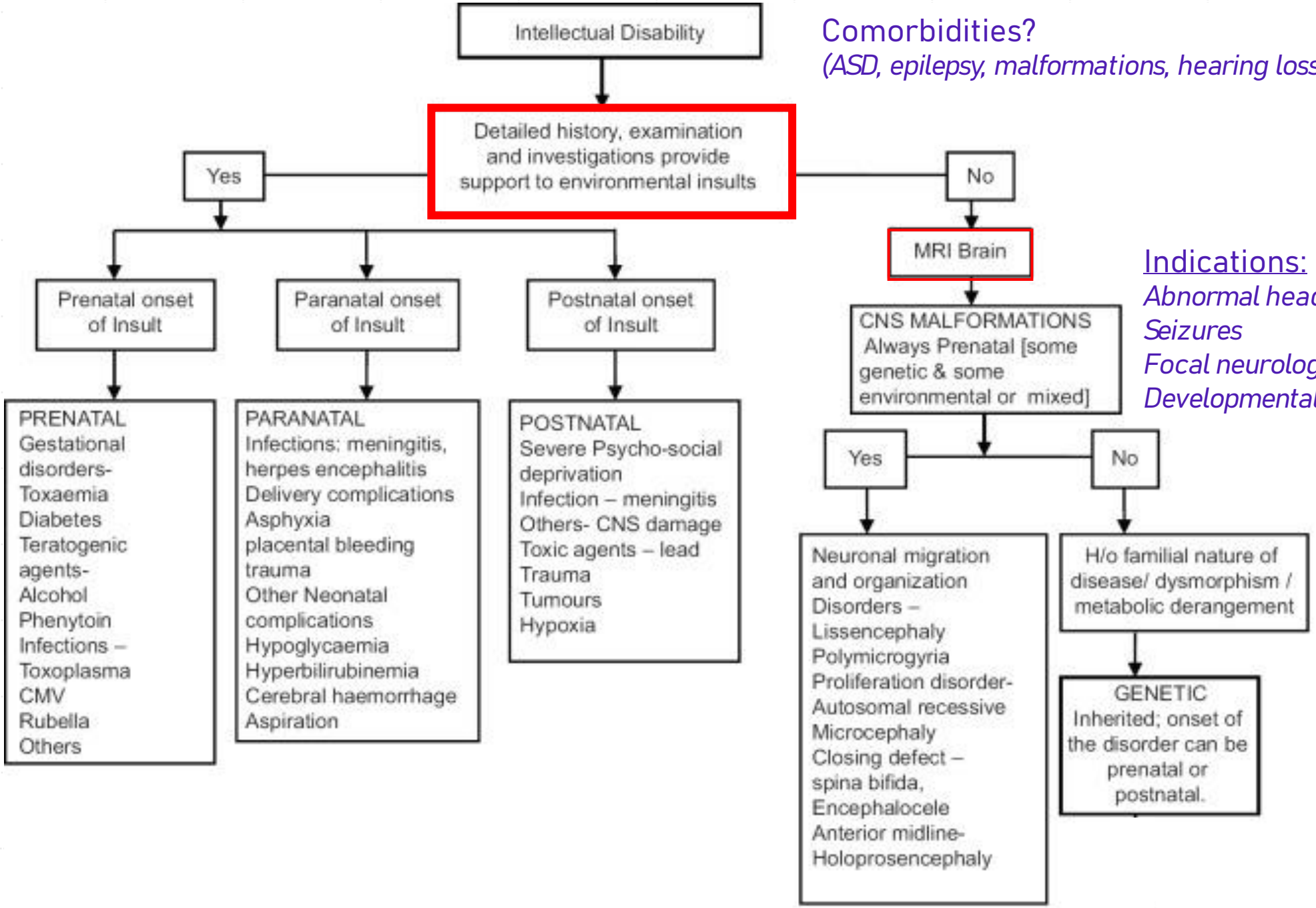
ID assessment and management

Maystadt I.

MANAMA

11.03.2024





Comorbidities?

(ASD, epilepsy, malformations, hearing loss, visual impairment,…)

Indications:

Abnormal head circumference
Seizures
Focal neurological signs
Developmental regression

Metabolic testing

Kishore MT et al, Indian J Psychiatr 2019

1st Tier: Non-Targeted screening to identify 54 (60%) treatable IEMs

Blood:

- ammonia, lactate
- plasma amino acids
- total homocysteine
- acylcarnitine profile
- copper, ceruloplasmin

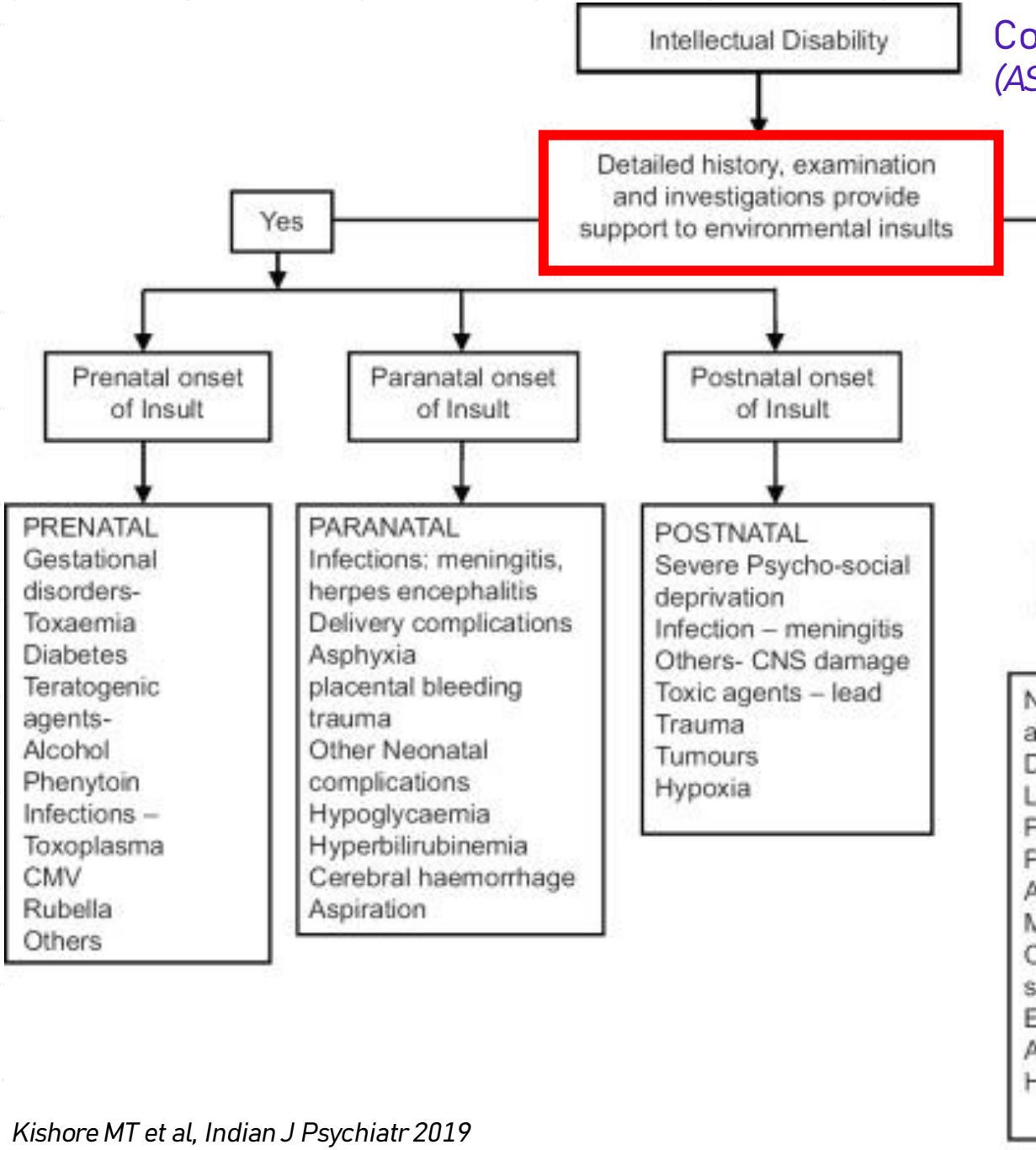
Urine:

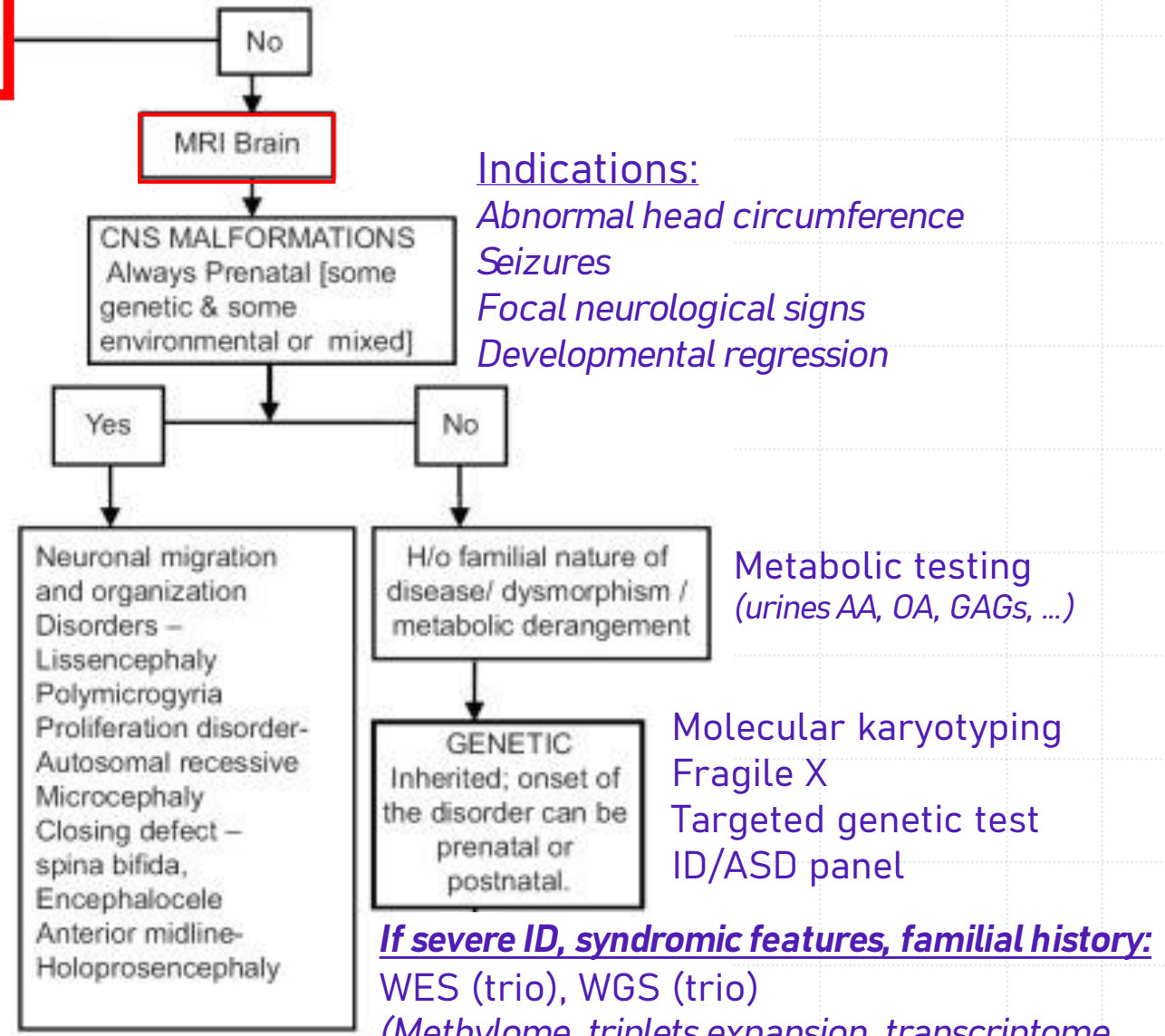
- organic acids
- purines & pyrimidines
- creatine metabolites
- oligosaccharides
- glycosaminoglycans

2nd Tier: Targeted testing to identify 35 (40%) treatable IEMs requiring 'specific testing'

- **according to patient's symptomatology patient (Table 4) & clinician's expertise**
- **utilization of textbooks & digital resources** (WebApp: *www.treatable-ID.org*)
- **consider the following biochemical / molecular analyses:**
  - whole blood manganese
  - plasma cholestanol
  - plasma 7-dehydroxy-cholesterol:cholesterol ratio
  - plasma pipecolic acid & urine AASA
  - plasma very long chain fatty acids
  - plasma vitamin B12 & folate
  - serum & CSF lactate:pyruvate ratio
  - enzyme activities (leucocytes): arylsulphatase A, biotinidase, glucocerebrosidase, fatty aldehyde dehydrogenase
  - urine deoxypyridonoline
  - CSF amino acids
  - CSF neurotransmitters
  - CSF: plasma glucose ratio
  - CoQ measurement fibcroblasts
  - molecular: *CA5A, NPC1, NPC2, SC4MOL, SLC18A2, SLC19A3, SLC30A10, SLC52A2, SLC52A3, PDHA1, DLAT, PDHX, SPR, TH*

Van Karnebeek CDM et al, Mol Genet Metab 2014





Comorbidities?

(ASD, epilepsy, malformations, hearing loss, visual impairment,…)

Indications:

Abnormal head circumference
Seizures
Focal neurological signs
Developmental regression

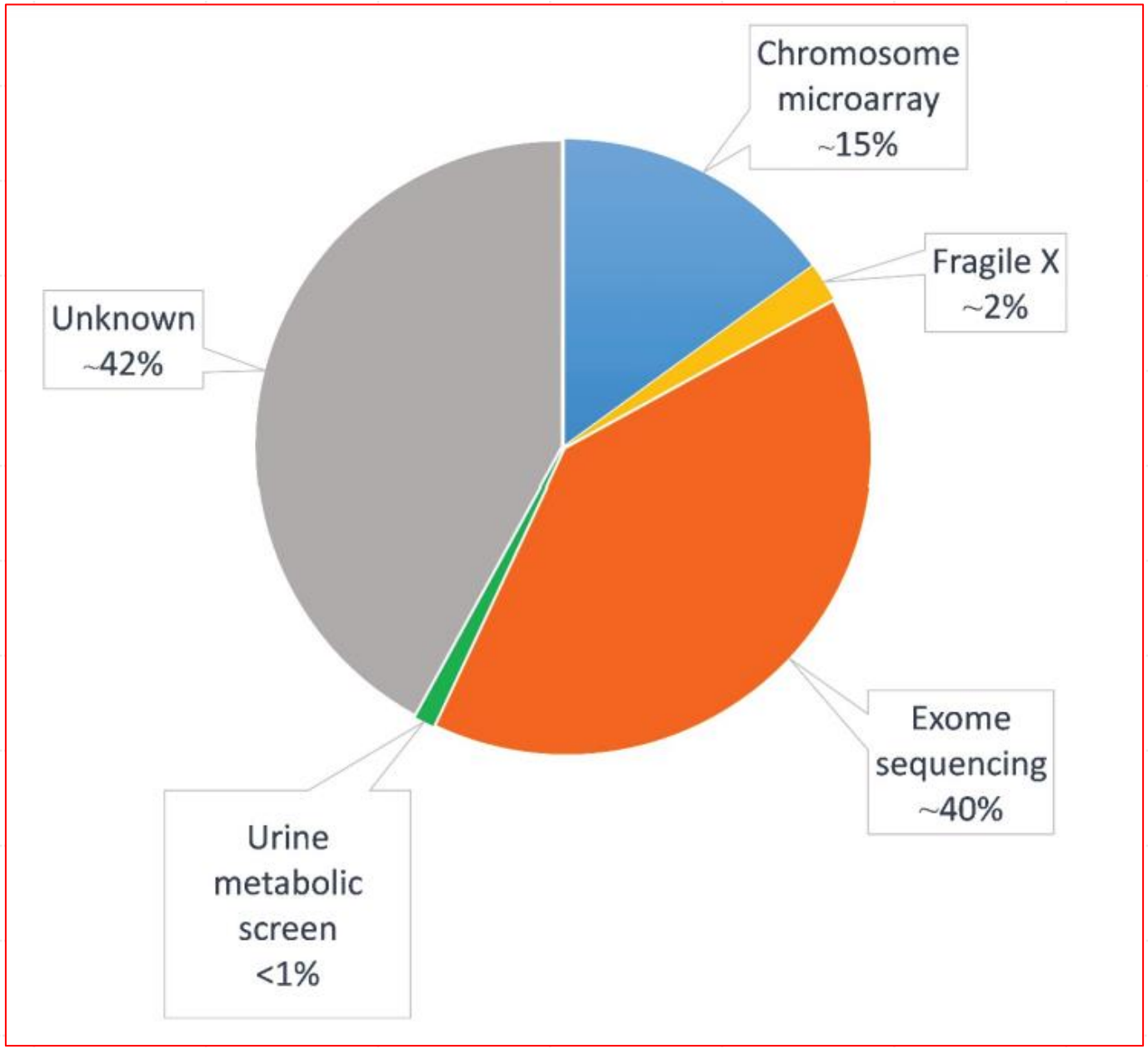
Metabolic testing
(urines AA, OA, GAGs, …)

Molecular karyotyping
Fragile X
Targeted genetic test
ID/ASD panel

If severe ID, syndromic features, familial history:

WES (trio), WGS (trio)
(Methylome, triplets expansion, transcriptome, proteome,…)

Kishore MT et al, Indian J Psychiatr 2019
Amor DJ et al, J Paediatr Child Health 2018



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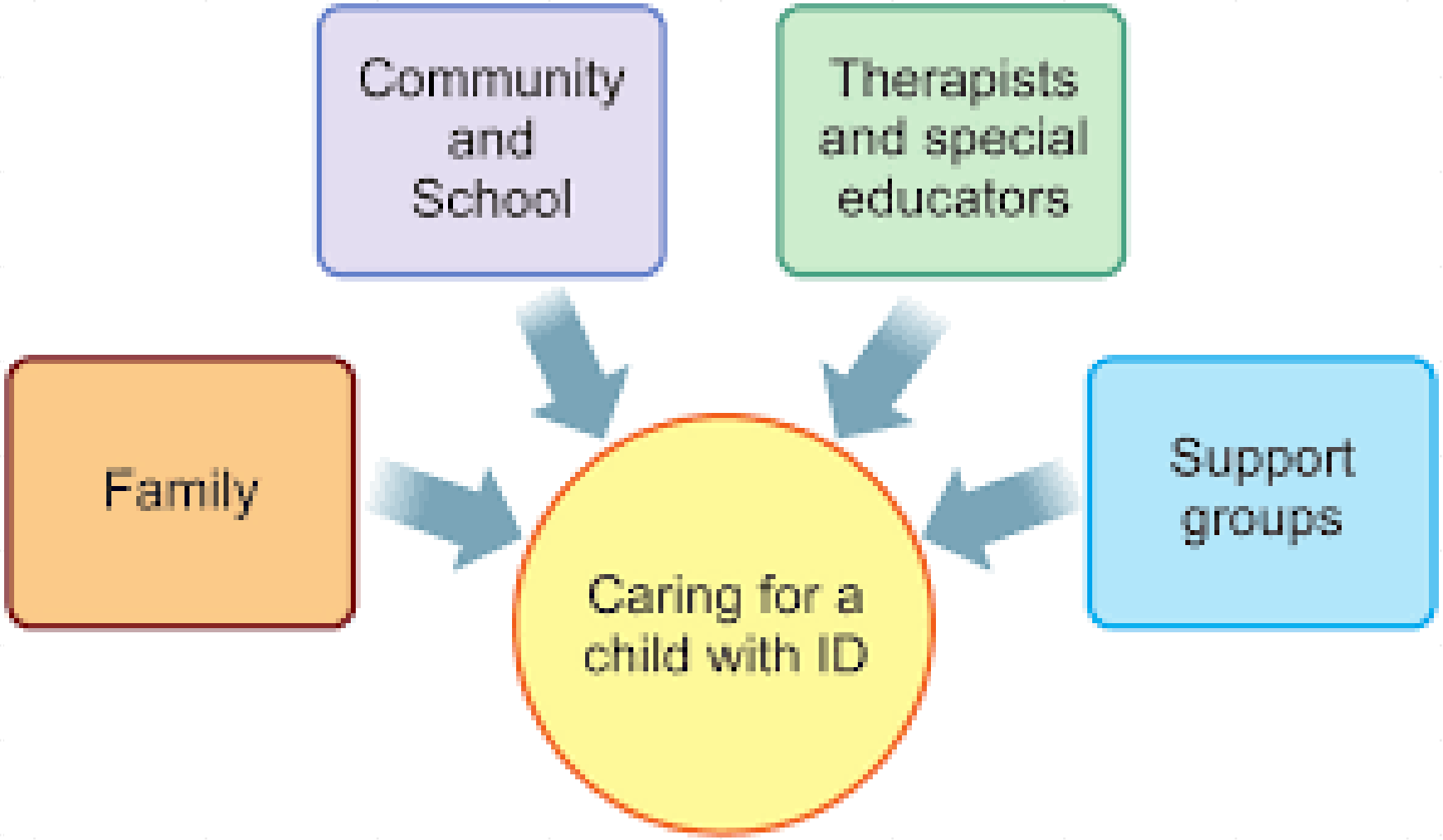
Community and School
Therapists and special educators
Family
Support groups
Caring for a child with ID


Physiotherapist
Clinical Psychologist
Occupational therapist
Pediatric Neurologist
Development Pediatrician
Speech Therapist
Special Educator

Special school:

- **type 1** : déficience intellectuelle légère - lichte verstandelijke handicap
- **type 2** : déficience intellectuelle modérée ou sévère
  matige of ernstige verstandelijke handicap
- **type 3** : troubles du comportement et/ou de la personnalité
  gedragsproblemen en/of emotionele problemen
- **type 4** : déficiences physiques - lichamelijke handicaps
- **type 5** : maladies ou élèves convalescents - zieke of herstellende leerlingen
- **type 6** : déficiences visuelles - visuele handicaps
- **type 7** : déficiences auditives - auditieve handicaps
- **type 8** : troubles de l'apprentissage - leerstoornissen
- **type 9** : autismespectrumstoornis, maar zonder verstandelijke beperking (enkel in de Vlaamse Gemeenschap)

https://www.belgium.be/fr/formation/enseignement/specialise
https://www.belgium.be/nl/Leren/onderwijs/buitengewoon_onderwijs