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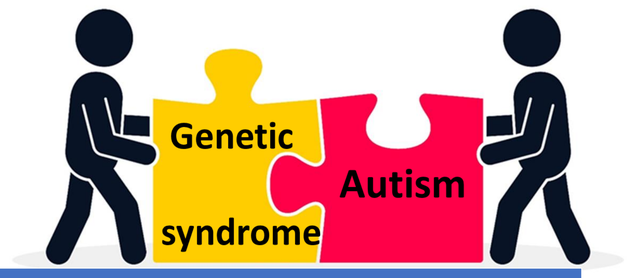

A broad exploration of clinical challenges in individuals with Phelan McDermid syndrome in Norway

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BACKGROUND

Phelan McDermid Syndrome (PHMDS) is a rare genetic disorder and represents a compound phenotype that may include a range of somatic, behavioural and psychiatric challenges.
Little is known about the Norwegian population of individuals with PHMDS.

METHOD

All known individuals with PHMDS throughout Norway were invited to participate.
A broad assessment was conducted via informant interview (family member or caregiver).
We used standardized instruments in addition to questions concerning level of functioning, language and communication, sleep, somatic problems, autism, psychiatric and behaviour problems, developmental/neuropsychiatric regression, etc.

RESULTS

N = 32, Males = 16 (50%) , Mean age = 21 years
Mean age at genetic diagnosis = 13

IQ

Mild ID	1
Moderate ID	11
Severe ID	18
Profound ID	1
Not ID	1

Autism
16 (50%) had an autism diagnosis
5 (16%) more suspected to have ASD after assessment

Communication:
10 (31%) primarily used spoken language
14 (44%) use ASC or gestures
7 (22%) had difficulties conveying basic needs

Psychiatric disorders
3 (10%) with bipolar before assessment
14 (44%) identified with suspicion of mental health disorder

Regression
20 (63%) had experienced regression/loss of skills

Somatic disease (e.g cancer, coeliac disease, NF2,…)	14
Sleep problems	27
Stomach pain	20
High pain threshold	21
Sweating problems (heat intolerance)	24

Many of the characteristics previously described in PHMDS were identified, but no characteristic appeared to be present in all participants.

CONCLUSION

The large variation across different domains highlights the need for broad and individualized clinical assessments for all individuals with PHMDS.

The heterogeneity underscores the need to avoid attributing co-occurring difficulties solely to PHMDS (genetic determinism), as this may lead to misinterpretation of symptoms and failure to adequately treat co-occurring difficulties.

Many individuals with severe somatic illness and limited speech calls attention to regular monitoring of somatic health.

Frequent reports of regression and suspected mental disorders (e.g., bipolar and catatonia) indicate that many participants need access to specialized mental health services.

The potential need for co-occurring mental health and somatic health services highlights the importance of collaboration between different parts of the health care system.

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