Results: The array CGH was normal. WES identified a pathogenic heterozygote variant c.5600G>A in the exon 31 of CREBBP gene, confirming MHS.

Conclusions: Overall, the features of our patient are consistent with those reported in the previous reports, including developmental and speech delay, autistic behavior, dysmorphic features, recurrent upper way infections, sensorineural hearing loss, and visual defects. Other common features, such as growth delay and microcephaly were not present in our patient. Our case contributes to the clinical characterisation of the new syndrome. Funding: The research leading to these results has received funding from the EEA Grant 2014-2021, under the project contract No 6/2019.

Disclosure: No significant relationships.
Keywords: developmental delay; Menke-Hennekam syndrome; dysmorphic features; autistic behavior

EPV0191
The Line between Psychois and Schizotypy: a case report.

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doi: 10.1192/j.eurpsy.2022.1111

Introduction: Since Kraepelin and Bleuler, schizotypy was understood as a mild expression of psychosis, a latent form with the same trajectory but different severity. They pointed characteristics such as being eccentric, unreasonable, superstitious or hypersensitive, interpersonal aversiveness (often related to suspiciousness and expectation of rejection), ambivalence, anhedonia, and psychosis-like features that don’t usually lead to help-seeking.

Objectives: To do a case review

Methods: We report a case of a 17 years old boy admitted in our department for developmental delay. The clinical examination revealed dysmorphic features; severe speech delay, mild intellectual disability, autistic behaviour. The patient had a personal history of recurrent respiratory infections, visual defect and bilateral sensorineural hearing loss. Other investigations included EEG, abdominal echography, and cerebral MRI all were normal. The genetic studies included array CGH and WES.

Results: The array CGH was normal. WES identified a pathogenic heterozygote variant c.5600G>A in the exon 31 of CREBBP gene, confirming MHS.

Conclusions: Overall, the features of our patient are consistent with those reported in the previous reports, including developmental and speech delay, autistic behavior, dysmorphic features, recurrent upper way infections, sensorineural hearing loss, and visual defects. Other common features, such as growth delay and microcephaly were not present in our patient. Our case contributes to the clinical characterisation of the new syndrome. Funding: The research leading to these results has received funding from the EEA Grant 2014-2021, under the project contract No 6/2019.

Disclosure: No significant relationships.
Keywords: developmental delay; Menke-Hennekam syndrome; dysmorphic features; autistic behavior

EPV0193
Self injuries in adolescence, an unusual clinical presentation of autism.

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doi: 10.1192/j.eurpsy.2022.1111

Introduction: Although autism is only twice more common in men than women in general population, in clinical samples women are underrepresented. This difference may be due to a poor sensitivity of current diagnostic criteria of autism related to females. We present a 13-year-old woman referred to the adolescent psychiatric unit for anxiety, self injuries and suicidal ideation. After careful assessment of current symptoms and neurodevelopmental milestones, deficits in emotional-comunicaional reciprocity, nonverbal communication and relationships emerged, as well as inflexible adherence to routines and restricted interests. The diagnose of autism spectrum disorder was made and the patient started a specific treatment.

Objectives: To review the clinical features of autism spectrum disorders in adolescent females and its differential diagnosis.

Methods: Review of the literature on autism spectrum disorders in female and its specific features.

Results: The “Female Autism Phenotype” is a group features that are more common in autistic women, as opposed to the classic symptoms of autism in men. Some of these differential characteristics are: fewer social impairments and higher levels of social motivation; more age and gender appropriate restricted and repetitive interests; more internalizing rather than externalizing symptoms; and a tendency towards camouflaging

Conclusions: - Autism in women is frequently underdiagnosed. - Females express autism in ways that not allways meet the current diagnostic criteria. - The “Female Autism Phenotype” has been proposed as an specific way of expression of autism in females.

Disclosure: No significant relationships.
Keywords: Adolescents; Autism Spectrum Disorder; Female autism phenotype