

EPV0275

Behavioral phenotype of Noonan-like syndrome with loose anagen hair

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Introduction: Noonan-like syndrome with loose anagen hair (NSLH MIM 607721) is associated to mutations in PTPN11, RAF1, BRAF and SHOC2 genes.

Objectives: Here, we report behavioral phenotype of a child suspected to have NSLH.

Methods: A 2-years-old Tunisian child harboring severe pulmonic valvular stenosis was referred to our genetic counselling for genetic assessment. Medical dysmorphism, cytogenetic analysis as well as genetic exploration of RAS-MAPK pathway genes were conducted.

Results: The child had short stature and ectodermal features including ichthyotic skin and thin-soft nails. He has specific hair appearance associated to NS features. In fact, he had a small nasal tip, thick lips and sticking-out rotated ears. He harbored typical nasal voice and loose anagen hair with ungrowing thin hair, sparse and pale scalp hair and eyebrows. He showed cognitive deficits with mental retardation and hyperactive behavior. Considered as having NSLH, cytogenetic analysis revealed a 46,XY formula, but molecular screening of PTPN11, RAF1, BRAF, RIT1 and SHOC2 genes was negative.

Conclusions: Mutations within the RAS-MAPK signaling pathway affect neurophysiologic activity in brain regions underlying attention and executive functions. Children with rasopathies demonstrated higher rates of attention deficit-hyperactivity (ADHD) and autism spectrum disorders. However, no studies have examined specifically the aspects of behavioral attention in the various types of Rasopathies. A recent study demonstrated that ADHD seems to be higher in children with NSLH and SHOC2 mutation, which is the case of our patient. We suggest that assessment of inattentive and hyperactivity symptoms in children should consider Rasopathies with specific molecular screening.

Disclosure: No significant relationships.

Keywords: Noonan-like syndrome with loose anagen hair; RAS-MAPK; attention deficit-hyperactivity disorder

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Psychiatric comorbidity in epilepsy and difficulties in treatment: A case report.

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Introduction: We present the case of a 36-year-old male with depressive states, impulsive traits and fits of anger (including episodes of self and heteroaggressiveness) since early childhood, in the context of traumatic family history and the beginning of an epileptic disease. These symptoms have been maintained over the

years, in addition to other variable and recurrent symptoms, such as severe anxiety, somatizations or serious depressive symptoms.

Objectives: To highlight the possible influence of epilepsy in the course of mental illnesses, especially depression, as well as the increased difficulty in management.

Methods: We collected the complete medical history of a patient with an important history of mental health in addition to epileptic disease since childhood and we carried out a review of the comorbidity between these diseases and their treatment.

Results: The epileptic disease of our patient may have influenced the behavioural alterations and the depressive symptoms since childhood, as well as the personality traits with aggressiveness and impulsiveness. There is an added difficulty in treating this case given the possible interactions between antiepileptic and antidepressant medications.

Conclusions: This case highlights the importance of taking into account the influence of this comorbidity on the prognosis of patients. Knowing the interactions and side effects of drugs is essential for good clinical management.

Disclosure: No significant relationships.

Keywords: epilepsy; comorbidity; Depression; Impulsivity

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Somatic Comorbidity of Anxious and Depressed Miners

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Introduction: Depression is the leading cause of disability worldwide and is a major contributor to the overall global burden of disease. The prevalence of depression is rising and it often co-occurs with other physical diseases.

Objectives: The research aims to determine the comorbidity of depression and anxiety disorders with chronic physical diseases among employees of the „Brown coal mine Banovici“.

Methods: We conducted a retrospective study that included 117 employees from the disease registry who are being under the treatment of depression and anxiety disorder. We collected data from medical records of patients about sex, age, marital status, smoking status, physical diseases, types of antidepressants, and the other drugs they use.

Results: The study showed that there are 117 employees of the „Brown coal mine Banovici“ who are under treatment of depression and anxiety-depressive disorder. 22 (18,8%) of them are females and 95 (81,2%) males in an average life span of 48,3 years. The most commonly used antidepressant is Escitalopram. 62 (53%) out of 117 patients with depression have comorbidity with diseases of the circulatory system, 24 (20,51%) have comorbidity with diseases of the musculoskeletal system and connective tissue, 16 (13,68%) have comorbidity with endocrine, nutritional and metabolic diseases. 25 (21,37%) patients are not suffering from any other chronic physical disease. The most commonly used drugs besides antidepressants are antihypertensives.

Conclusions: The comorbidity rate of depression and anxiety disorders with cardiovascular diseases among employees of the