was limited to female participants, the genetic effect stayed significant only at the anger scale of the BPQ.
Conclusions: Family environment had pronounced effect on aggressive behavior and personality functioning, interaction with common monoaminergic genetic variants was detected only in women.
This study was supported by the National Research Development and Innovation Office grants NKFI K 129195 and NKFI K 135437.

Disclosure of Interest: None Declared

EPV0541

Pain and gain of predictive genetic testing: Particular case of fragile X syndrome

N. Bouayed Abdelmoula*, N. Ramma and B. Abdelmoula

Genomics of Signalopathies at the service of Precision Medicine - LR23ES07, Medical University of Sfax, Sfax, Tunisia
*Corresponding author.
doi: 10.1192/j.eurpsy.2024.1221

Introduction: The purpose of predictive genetic tests is to identify carriers or the onset of a disease in pre-symptomatic individuals. Prediction is linked to a negative psychological impact (anxiety, depression, etc.), depending on the perception of risk, the severity of the disease, and the availability and effectiveness of treatments.
Objectives: Here, we report on genetic counselling during predictive genetic testing offered to an Arab family affected by fragile X condition (FXS) caused by the unstable expansion of a CGG repeat (CGGR) in the FMR1 gene.
Methods: A 10-year-old boy who harbored a mental retardation was referred to our genetic counselling for genetic testing as he was suspected to be affected by FXS. Screening of FMR1 gene mutations was conducted for the index case and his mother. A predictive genetic testing for the family members (brothers, sisters and others) was offered, focusing on knowledge of genetics and medical risks of FXS.
Results: FMR1 molecular analysis showed a full mutation (300 to 2000 CGGR) for the boy and a large premutation (100 CGGR) for the mother. During genetic counselling, the family was informed about the significance of the genetic results. In FXS initiated by an expansion of over 200 CGGR. While mental retarded males usually harbor the full mutation, the mother carry a premutation (70 to 200 CGGR). The deficiency of FMR protein (FMRP) in the neurons of affected males leads to brain developmental abnormalities. Some pre-mutated children may show signs of the autism spectrum disorder and females may develop FMR1-related premature ovarian insufficiency. An increased risk of a late onset fragile X tremor ataxia syndrome is identified in pre-mutated men (55 to 200 CGGR) and less in women.
Conclusions: The reduction or loss of FMRP leads to multisystem damage. Neuropsychiatric disorders such as mental retardation, speech and language delay, autism spectrum disorder, sensory hyperexcitation, social anxiety, abnormal eye contact, shyness and aggressive behaviour are common in individuals with the mutation. Affected women are often under-diagnosed because mental retardation is not constant, but minor disorders including a borderline IQ with learning difficulties and emotional disturbances have been reported. Conditions associated with fragile X premutation, a term proposed by the European Fragile X Network (FXPAC), seem to be characterized by many physical and psychological health symptoms. Anxiety, depression, sleep disorders and mood disorders are more common in permutated individuals. However, new reports suggested that FXS patients could be at unusually low risk of cancers, because FMRP is overexpressed in multiple cancer tissues.

Disclosure of Interest: None Declared

EPV0542

Dysmorphic physical appearance and psychosocial burdens in Klippel-Feil condition

N. Bouayed Abdelmoula*, R. Rannen and B. Abdelmoula

Genomics of Signalopathies at the service of Precision Medicine - LR23ES07, Medical University of Sfax, Sfax, Tunisia
*Corresponding author.
doi: 10.1192/j.eurpsy.2024.1222

Introduction: Klippel-Feil abnormality (KFA) is an association of bone defects characterized by a triad: fusion of the cervical vertebrae and consequent short neck, low hairline and a limited motion in the neck. KFA may be a feature of another disorder, such as MURCS association. Familial mutations in the GDF6 (KFS1 8q22), MEOX1 (KFS2 17q21), GDF3 (KFS3 12p13) and MYO18B (KFS4 22q11) genes cause inherited KFA.
Objectives: The aim of this study was to report dysmorphic features and psychological burdens in two sisters with Klippel-Feil condition.
Methods: Two sisters with amenorrhea and dysmorphic clinical features were examined at our genetic counselling. Assessment of dysmorphic and behavioral features and karyotyping using RHG banding were performed.
Results: Familial history revealed consanguineous parents and seven other healthy sisters. Physical examination shown typical triad of KFA. Karyotyping showed 46,XX formula in both patients. The first 22-year-old sister had body asymmetry with size difference between the two sides at the level of bones, pectus excavatum of the sternum, an ascent of the left scapula, scoliosis, dental position abnormalities and facial dysmorphism. The second 28-year-old sister had size difference between the two legs and scoliosis, vitiligo and facial dysmorphism. Anxious and depressed, the two sisters had normal learning abilities but shared many personal psychological concerns regarding their physical appearance and their amenorrhea. They were also exposed to significant discrimination and stigma making them feel excluded and ignored because of their visible difference.
Conclusions: Physical appearance has a profound impact on a person's life. To our knowledge, there is no reports that describe specific psychological burdens of KFA. Self-esteem, body image, and quality of life is negatively impacted in the case of dysmorphic physical appearance, always associated to social discrimination. Patients with KFA should be assessed not only for associated congenital defects but also for psychological distresses.

Disclosure of Interest: None Declared