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Therapeutic hands in the intervention for adult with Fragile X syndrome

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Fragile X syndrome (FXS) is a genetic disorder. Person who suffer from this disorder often have mild to moderate intellectual disability. They have physical features like long narrow face with large ears; their fingers are flexible and they also have large testicles. Genetics (Home Reference 2012)They have similar features of autism like difficulties in social interactions and delayed speech. They also seem to be hyperactive. FXS is caused by a trinucleotide repeat within the fragile X mental retardation 1 (FMR1) gene at the end of the long arm of the X chromosome. (McLennan, Y 2011) It is recognized that the genes on this chromosome mutation takes place and cause learning disabilities. Thus fragile X is commonest cause renowned to be X-linked learning disabilities. In this disorder where there is an involvement of broad spectrum fragile X cause learning and emotional problems than mental retardation. Though intellectual difficulties present in this population it is only mild to moderate this is resulted by correlation with the molecular measures this may be also some extent contributed by developmental delay, difficulties in social interaction, emotional dysregulation and learning disabilities. The FMR1 mutation leads to a fragile site causing commonest developmental disorder in this population is learning disabilities. The special education is needed for this population to be identified until they become significant. FMR1 mutation causes mild emotional problems and learning difficulties.

Developmental problems are universal differ with degree especially transition to adolescence and from adolescence to adulthood. It becomes more difficult for individuals with FXS because of their cognitive deficits and emotional difficulties.For individuals withFXS the gradual transition into dependent more independent living can help. May be providing separate individual units to live near to place where the family resides this kind of approach can extend some degree of independence people with FXS, thus allowing for limited supervision from the family. *(*Rueda JR, 2009) Here family supervises them to participate in adult programmes to boost their interactive skill. Other alternative adult with FXS is to arrange group homes for them. (Hagerman, R.J. 1996b). Most adult males with FXS require some degree of supervision is needed for this population in managing their living. Families can supervise them with limited support visiting them daily and going-over happenings with them. Regularly scheduled group activities are one of the key issues for enhancing their social interaction.

Periodic outbursts either with verbal or physical aggression is common in adults with FXS. These behaviour problems are often associated with anxiety and mood instability these behavioral symptoms responds well with treatment using SSRI s are useful in decreasing anxiety and obsessive–compulsive behaviour and also helps to manage affective symptoms such as moodiness and aggression. Other medication such as mood stabilizer, atypical antipsychotics (risperidone or quetiapine) also used. (Garber, KB2008) The combinations of SSRI with atypical antipsychotics are used to decrease aggression and decrease anxiety in adults with FXS. (Garber, KB2008)

In teaching calming down techniques to the adult with FXS occasionally sensory integrationtherapy can be used adjunctively with medication. Here the patients can be though self-management or the family members can be trained to help the patient. Professional help from psychologist may further beneficial. Here the professionals help these individuals to recognize his or her emotional state. They also teach them focusing, visualizing, counting, distracting from thought or off tracking from a situation causing behavioural outburst. Along with these self-calming techniques they counsel these individual and help them to manage their sexuality issues and interpersonal difficulties

It is important to start the intervention in early age. But the diagnosis of the FXS delays because of parental rigidity in accepting the fact. However once the individual is diagnosed with FXS next important step is genetic counselling also a part of the treatment programme. Further individuals at risk for being carriers or affected by FXS should be tested. This can be done through DNA studies; documentation for extent of the CGG repeat expansion is done.

Genetics counsellor and by the treatment team actively extend the counselling and psycho education regarding FXS which can aid the decisions regarding continuation of a pregnancy or termination of an affected fetus. Finally, this has been left for the personal decisions made by the family with an extended support of the counsellor.

The various supporting groups as a part of intervention to benefit the FXS population are: family groups, parental groups, national and international support groups. There are provisions in various setups where patients and the families are provided with psychoeducation in this aspect to be aware of the benefits. Psycho-educational programmes are set to advice about the syndrome to the families; the professionals are trained for extending these facilities to the families of FXS.

In addition to medical interventions various professionals are involved in the intervention programmes to enriching the lives and wellbeing in those affected population by FXS. In advancing intervention programmes also carry on with gene therapy or protein replacement therapy for individuals with FXS. (Hagerman RJ 2009)

In summary there is no cure for FXS, the management of these patients go with holistic approach where a number of pharmacological, behavioural and cognitive interventions are made use to improve their quality of life. Interventions included in the holistic approach are -: Speech therapy, special needs education behavioural therapy. (Hagerman RJ 2009) Genetic counselling and support of the parents and other family members are found to be helpful (Hogan, A 2012).

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