# GENETIC SYNDROMES, MENTAL RETARDATION AND EPILEPSY

| GENETIC SYNDROME | MENTAL RETARDATION | EPILEPSY |
|---|---|---|
| | | |
| Amino Acid Disorders: Tay-Sachs, Sandhoff's | YES | YES |
| Angelman | YES | 86-96% |
| Cornelia de Lange (CdLs) | YES | 23% |
| Down | YES | 6-13% |
| Fragile-X | YES | occasional |
| Homocystinuria | YES (58%) | YES, onset from 6 months to 5 years of age, with abnormal EEG pattern |
| maternal PKU | YES(>80%) | >80% |
| Mucopolysaccharide Disorder : Sanfilippo | YES | YES |
| Neurofibromatosis | YES (5-50%) | 20% |
| Prader-Willi (PWS) | YES | occasional |
| Rett's | YES | YES |
| Rubinstein-Taybi | YES | 23% |
| Smith Magenis | YES | occasional |
| Sturge-Weber | YES (61%) | 56% |
| Tuberous sclerosis | YES (62%) | 93% |
| Velo-cardio-facial (VCFS) ( 22q- ) | YES (40%) | frequent |

## PREVENTION STRATEGIES

| Possible Precipitating Factors ("Triggers") | Prevention |
|---|---|
| Fatigue, lack of sleep | Follow a schedule/routine to maintain adequate amounts of sleep |
| Exercise | Monitor amount of exercise, stay cool, take breaks |
| Hypoglycemia (low blood sugar) | Maintain regular meal & snack times |
| Emotional stress | Monitor closely, avoid if possible |
| Infection, febrile illness | Monitor for signs & symptoms of illness: changes in behaviour, increase in seizures. See MD for proper treatment-antibx. |
| Alcohol | Limit intake |
| Hyperventilation | Monitor closely, try to avoid |
| Hormonal changes (menstruation, ovulation, menopause) | Monitor closely, Rx by MD for HRT (hormone replacement therapy), AED adjustments |
| Particular odours, loud noises or music, being startled | Monitor & keep to a minimum |
| Flashing lights (TV, video & computer games, disco lights, sun flickering through the trees when driving or bright sunlight in general) | Keep person 2m away from TV and 30 cm away from computer screen.<br>Keep room well lit.<br>Decrease exposure to fluorescent bulbs.<br>Wear sunglasses or tinted glasses to decrease effect of flickering lights.<br>Cover one eye, if exposure not preventable, to decrease photosensitive effect. |

**Best Prevention: Take AED's regularly to maintain adequate blood levels

| Challenging Behavior | Intervention |
|---|---|
| *Can include any or several of the following behaviors:*<br><br>Aggression against objects : hitting the table, the wall, the chair, throwing personal possessions<br><br>Aggression against persons – specify if targeting staff, peers or himself, specify exact type of aggression<br><br>'Refusals' - specifically by sitting or falling down on the ground, or by performing 'odd behaviors' – wringing hands, picking lint off clothes, chewing movements, lip-smacking or by<br>'Ignoring' – seeming distracted, as if daydreaming, staring into space & not responding<br><br>Removing/pulling at clothing at inappropriate times<br><br>Yelling, crying, laughing, moaning at inappropriate times<br><br>Running away, wandering<br><br>**If the person is having a seizure, he will need to return to his 'usual self' before resuming any scheduled activities. | **INTERVENE PHYSICALLY <u>ONLY</u> IF THERE IS RISK OF DANGER TO THE PERSON OR OTHERS, OTHERWISE:**<br><br>***<u>LOOK :</u>***<br>• Observe position of the person, is there any posturing?<br>• Are the eyes or head deviated to one side? Are the pupils dilated?<br>• Does he seem to be aware of his surroundings: is he systematically targeting certain persons or objects or just those that are most easily accessible to him?<br>• Is he pale/flushed, or sweating?<br>• Are there any asymmetric movements of the body or facial grimacing?<br>• Is the movement purposeful & is there a connection to what is presently going on?<br>• Is or was there a potential trigger present – sunlight, heat, exercise, stress?<br>***<u>LISTEN :</u>***<br>• What sounds is the person making?<br>• Is he repeating words or phrases, if so, what is he saying?<br>• Did he stop speaking in mid-sentence?<br>• Is he hyperventilating, snorting or breathing loudly or differently than usual? Describe this.<br>***<u>TEST :</u>***<br>• Does the person respond or acknowledge you when you speak to him, or when you call his name?<br>• Does he know where he is? Can he identify familiar objects?<br>• Does he remember what was said to him during the episode?<br>• How does he react when you approach? (*ALWAYS APPROACH SLOWLY)<br>***AND DOCUMENT ALL OF THE ABOVE INFORMATION.*** |