WILLIAMS SYNDROME

- **Williams syndrome is a genetically determined disorder that occurs in approximately 1 in 25,000 live births.**
- **Williams Syndrome is caused by a microdeletion on chromosome 7.**
- **The condition typically presents in infancy, with difficulties in feeding, profuse vomiting, irritability, constipation and failure to thrive, and distinctive facial features. They also often experience colic for an average length of 7.4 months.**
- **The diagnosis of WMS is made initially by clinical evaluation, usually during midchildhood when the characteristic facial features, cognitive profile, and cardiac findings become more apparent with age.**
- **Cardiovascular and renal abnormalities are common, as are other features such as joint laxity and growth retardation.**

Common Facial Features

- **Flattened midface with a broad forehead**
- **Upturned nose with a depressed nasal bridge and broad nasal tip**
- **Periorbital fullness and epicanthal folds**
- **Broad mouth with full lips**
- **Full and drooping cheeks**
- **Prominent ears**
- **The iris is usually described as radiating a white lacy or prominent starburst pattern that makes they eye "sparkle"**
- **Often termed "elfin-like"**
- **These typical facial anomalies become recognized during the first year of life and often as early as 4 months of age**

Physical Problems

- **Auditory problems - Chronic otitis media (ear infections)**
- **Visual Problems - Strabismus (it is very important for infants and children with WMS to have a detailed eye exam performed)**
- **Dental Abnormalities - overbite, excessive interdental spacing, microdontia, and small roots**
- **Feeding difficulties**
- **Genito-urinary problems - uncontrollable daytime and nighttime wetting**

- **Musculoskeleton difficulties - Hypotonia, scoliosis, and awkward gait**
- **Cognitive Abilities**
- **95% of cases have mild to severe intellectual disabilities**
- **Average IQ 55 (range 40-90)**
- **Performance scores are significantly lower than verbal IQ**
- **General knowledge and numeracy skills tend to be poor, and relative deficits are also found on tasks involving hand-eye coordination, performance speed and visual sequencing**

Language

- **Early language may be delayed, but later expressive skills often appear complex and well developed**
- **Various semantic and pragmatic deficits have been identified**
- **There is an excessive, and frequently inappropriate, use of clichés and stereotyped phrases, and problems in reciprocity, such as poor turn-taking and topic maintenance.**
- **Adolescents with WMS tend to be articulate, and are talkative to the point of being loquacious.**
- **The key language characteristic of WMS is called "Cocktail Party syndrome" - the affected children are quite loquacious, over friendly, and posses an outgoing personality.**
- **They often develop a hoarse or brassy voice, which is one of the more unique characteristics of this disorder.**

Academic Attainments

- **They do relatively well on test of reading or spelling**
- **They have difficulty in mathematics and its application to everyday life - making change, balancing a checkbook, and cooking from recipes**
- **Poor nonverbal skills - number manipulation, spatial cognition, motor planning, and problem solving**

Social and Behavioral Functioning

- **Distractible**
- **Poor concentration**
- **Attention seeking behavior**

- **Excessive anxiety**
- **Preoccupations and obsessions**
- **Eating and sleeping difficulties**
- **Poor peer relationships**
- **Hyperactive**
- **Hyperacusis - oversensitivity to sounds**

Treatment

- **Requires a multidisciplinary approach**
- **A physical therapist should evaluate and treat joint contractures, postural abnormalities, and spasticity**
- **Some children with WS will require speech therapy focused on all aspects of language and even the highest functioning children will benefit from therapy focused on pragmatics**
- **The occupational therapist should work on the visual-motor integration**
    - **Children with WS benefit from being taught to talk their way through fine motor tasks and to use verbal cues to help remember spatial information**