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Aicardi Syndrome Presenting with Neurogenic Bladder and Recurrent Pyelonephritis

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Description

Aicardi syndrome is a rare neurodevelopmental disorder characterized by a classic triad of agenesis of the corpus callosum, chorioretinal lacunae, and infantile spasms. Since its first description by Jean Aicardi in 1965, fewer than a thousand cases have been documented worldwide. The condition occurs almost exclusively in females and is thought to result from a de novo mutation on the X chromosome that is lethal in males. Clinically, Aicardi syndrome manifests with severe developmental delay, seizures, distinctive ocular lesions, and various systemic abnormalities involving the musculoskeletal, gastrointestinal, and genitourinary systems. Among its lesser-known complications is neurogenic bladder dysfunction, which may predispose patients to urinary retention, recurrent urinary tract infections, and ultimately pyelonephritis. The coexistence of neurological impairment and structural urinary tract anomalies creates a complex clinical scenario requiring coordinated multidisciplinary management.

The pathogenesis of bladder dysfunction in Aicardi syndrome is linked to its widespread central nervous system abnormalities. Agenesis or dysgenesis of the corpus callosum and other midline structures can disrupt descending inhibitory control over the pontine micturition center and sacral spinal pathways, resulting in impaired detrusor-sphincter coordination. The type of neurogenic bladder observed in these patients varies from detrusor overactivity to detrusor areflexia, depending on the location and severity of the neural lesion. In practice, many children exhibit a mixed pattern with poor bladder compliance, incomplete emptying, and elevated residual urine volumes. The combination of urinary stasis and impaired sensation increases susceptibility to bacterial colonization and recurrent infections.

Recurrent pyelonephritis in Aicardi syndrome represents a serious complication with potential to cause progressive renal damage. The ascending route of infection is the most common, beginning with bacteriuria secondary to stasis and culminating in infection of the renal parenchyma. Patients with neurogenic bladder often fail to perceive the early symptoms of lower urinary tract infection, such as dysuria or frequency, resulting in delayed diagnosis. In addition, the frequent use of diapers and mobility limitations make sterile voiding and hygiene difficult to maintain. Consequently, bacterial proliferation within the bladder progresses unchecked until systemic manifestations-fever, flank pain, or irritability-appear. Over time, repeated episodes of pyelonephritis may lead to scarring, reduced renal function, and hypertension.

The diagnosis of neurogenic bladder in children with Aicardi syndrome requires high clinical suspicion and systematic evaluation. Urodynamic testing remains the gold standard for characterizing detrusor behavior, bladder compliance, and outlet function. Typical findings include elevated detrusor pressure

during the filling phase, reduced bladder capacity, or incomplete emptying during voiding. Ultrasonography of the kidneys and bladder can reveal structural consequences such as hydronephrosis, thickened bladder wall, or residual urine post-void. Voiding cystourethrography may identify vesicoureteral reflux, a common accompaniment that further amplifies the risk of upper tract infection. Because many patients are nonverbal or severely developmentally delayed, clinical assessment must rely on objective measures and caregiver observation of urinary patterns, incontinence, or unexplained fevers.

Management of neurogenic bladder in Aicardi syndrome is primarily directed toward maintaining low intravesical pressure, ensuring complete bladder emptying, and preventing infections. Clean intermittent catheterization is the cornerstone of therapy, promoting regular bladder evacuation and minimizing stasis. Caregivers require extensive education and support to perform catheterization aseptically and on schedule, as adherence is often challenging in children with severe neurological disability. Anticholinergic agents such as oxybutynin or tolterodine may be prescribed to reduce detrusor overactivity and improve bladder compliance. For patients with high leak-point pressures or refractory incontinence, intravesical botulinum toxin injection or bladder augmentation may be considered, though such interventions are rarely performed in the profoundly disabled population affected by Aicardi syndrome.

Antibiotic prophylaxis is a contentious but sometimes necessary measure in patients with recurrent pyelonephritis. Continuous low-dose prophylaxis with agents such as trimethoprim-sulfamethoxazole or nitrofurantoin can reduce infection frequency but risks fostering antibiotic resistance. Prophylaxis should therefore be individualized, guided by urine culture results and infection recurrence patterns. Equally important is meticulous hygiene, adequate hydration, and routine bladder irrigation in patients prone to mucus accumulation or catheter-related encrustation. Regular follow-up with renal ultrasonography and serum creatinine measurement is recommended to monitor kidney function and detect early evidence of upper tract deterioration.

From a pathophysiological standpoint, the occurrence of neurogenic bladder in Aicardi syndrome exemplifies how congenital cerebral malformations can exert far-reaching effects beyond the nervous system. The central integration of micturition involves cortical, pontine, and spinal pathways; any disruption of this complex circuitry can produce profound bladder dysfunction. Agenesis of the corpus callosum impairs communication between hemispheres and descending control, while associated midbrain anomalies may interfere with pontine micturition coordination. The result is an unmodulated sacral reflex arc that fails to achieve synchronized contraction and relaxation, predisposing to urinary retention, overflow incontinence, and infections.

Conclusion

Aicardi syndrome presenting with neurogenic bladder and recurrent pyelonephritis illustrates the intricate interplay between neurological and urological pathology in rare congenital disorders. The neurogenic bladder arises from central nervous system dysgenesis and predisposes to urinary stasis and ascending infections. Early diagnosis through urodynamic evaluation, vigilant monitoring, and consistent bladder management are critical to preserving renal function. A comprehensive approach that integrates medical, surgical, and supportive care ensures the best possible outcomes for affected children. As awareness of this rare association grows, timely recognition and multidisciplinary collaboration can significantly mitigate morbidity and improve quality of life for patients living with Aicardi syndrome.