DETECTION OF MICRODELETION IN CHROMOSOME 7 q11.23 BY FLUORESCENT IN SITU HYBRIDIZATION TECHNIQUE AMONG AN EGYPTIAN COHORT SUSPECTED TO HAVE WILLIAMS SYNDROMRE

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INTRODUCTION

Williams syndrome (WS) is a unique type of microdeletion syndromes that causes a wide spectrum of multisystemic clinical manifestations. Its incidence is somewhere between 1 in 7500-20000 livebirths. It results from a deletion of 1.5-1.8 megabases (Mb) in chromosome 7q11.23 that can be detected by fluorescent in situ hybridization (FISH) technique which is the gold standard for its diagnosis. WS is characterized by dysmorphic facies, cardiovascular disease, most commonly.
supravalvular aortic stenosis (SVAS), intellectual disability, a characteristic cognitive profile, and idiopathic hypercalcemia. This study was aiming to evaluate the clinical, cardiac, and biochemical aspects in a cohort of children with WS in Egypt and confirm the microdeletion by FISH technique.

AIM OF THE WORK

1. Clinical assessment of children suspected to have WS attending the genetics clinic at Smouha Hospital University, Faculty of medicine.
2. Establishing molecular diagnosis for confirmation of the suspected cases by FISH technique through detection of chromosome 7q11.23 microdeletion.

PATIENTS AND METHODS

The study involved twelve WS patients who came to Smouha university hospital to seek medical advice. Full history taking especially developmental, medical and surgical history, clinical examination, cardiovascular evaluation including echocardiography, IQ testing and laboratory investigations including plasma calcium level, thyroid function tests, complete blood picture and blood glucose level, were performed. FISH testing was done for all patients except one patient who died before testing due to severe SVAS.

RESULTS

Table : Demographic data and anthropometric measurements

Age		
Min - Max	**0.33-9.25 years**	
Mean ± SD	**4.86 ± 3.13 years**	
	No of patients (n=12)	**% of patients Total=100%**
Sex		
Male	7	64.29 %
Female	5	35.71 %
Anthropometric measurements		
Height:		
Short	3	25%
Normal	9	75%
Tall	0	0%
Weight:		
Underweight	4	33.33%
Normal	8	66.67%
Overweight	0	0%
Head Circumference:		
Microcephalic	3	25%
Normal	9	75%
Macrocephalic	0	0%
Facial dysmorphic features		
Common WS features	12	100%
Less common WS features	10	83.33%
Congenital Cardiovacular disorder		
SVAS	9	75%
PAS	6	50%
Biochemical disorders		
Hypercalcemia	1	8.33%
Hypothyroidism	2	16.67%
Hyperglycemia	0	0%
IQ test (n= 10)		
Mentally retarded	9	90%
Mentally normal	1	10%
Clinical scoring[31]		
Mean: 9.5		
Range: 6-13		
Results		
Score: < 3	0	0%
Score: > 3 and ≤ 9	5	41.67%
Score: > 9	7	58.33%
Deletion by FISH	10 out of 11	90.9%

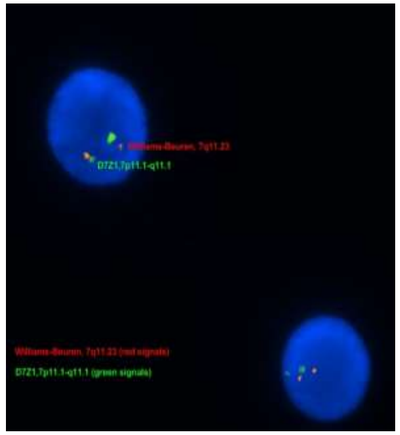

Figure 1:
FISH metaphase spread showing no deletion of WBSCR.

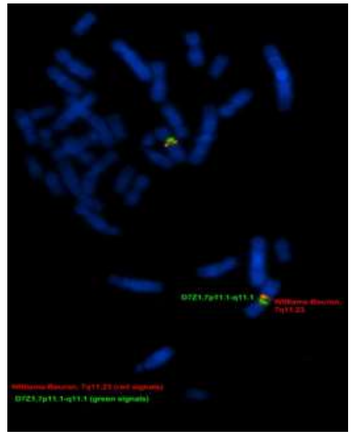

Figure 2:
FISH metaphase spread showing positive deletion of WBSCR.

CONCLUSION

From Our current study, we can conclude that:

- FISH is the gold standard technique for diagnosing WS but this must follow multidisciplinary clinical evaluation.
- Management of WS patients depends on understanding the nature of this condition, familiarity of its medical problems, and routine clinical, laboratory and imaging evaluations and undertake necessary interventions needed for them.