Attached is a report from the Genome Diagnostics or Cytogenetics laboratory. For SickKids patients, a copy of the report will also be uploaded to the Epic patient chart. Please disregard the information below this line.

Environment Name: LIVE
Order Number: 190026564
Report Type: Test Specific Report for ARRAY
Tests: ARRAY
Delivery Reason: Attending Doctor, Requesting Doctor, Test Ordering Doctor RI_LEVINLE

SickKids®
The Hospital for Sick Children
555 University Avenue
TORONTO ON M5G 1X8

Division of Genome Diagnostics
Department of Paediatric Laboratory Medicine
Phone: (416) 813-7200 x1 Fax: (416) 813-7732
CLIA ID: 99D1014032
www.sickkids.ca/genome-diagnostics

Referring Physician: LEVIN, LEO
Children's Aid Society of Toronto (Isabella)
30 Isabella Street
Toronto ON M4Y 1N1
cc:

Patient Name: **WILLIAMS-MASSIAH, OMARI**
Order Number: **19-26564**
DOB (yyyy-mm-dd): 2008-08-25
Sex: M
MRN#: 5136451
External MRN:
HCN#: 7931682160 ON
Family Number:
Ethnicity:
Billing#: 19275216

Report Date: **2019/12/30 10:54 PM**
Specimen Type: Blood
Collection Date: 2019/12/05 6:15 PM
Registration Date: 2019/12/06 5:30 PM
Received Date: 2019/12/06 11:30 AM

MOLECULAR GENETICS REPORT

Fragile X syndrome and FMR1-related disorders

Test(s) Performed:
FMR1 gene CGG Repeat analysis

Reason for Referral:
Diagnosis

RESULTS:

Molecular results do not support a diagnosis of fragile X syndrome in this patient.

Genetic Analysis	***FMR1 (CGG)n repeat(s)***	***Expansion Range***
FMR1	25	Normal

INTERPRETATION:
*margin of error is +/- 2-3 repeats.

METHODS:
This patient was tested by PCR amplification using the Asuragen AmplideX® *FMR1* PCR kit to detect alleles in the normal, intermediate, premutation and full expansion (>200 CGG repeats) range.

LIMITATIONS AND OTHER TEST NOTES:
This analysis is based on current knowledge of the molecular genetics of *FMR1*-related disorders. Test results should only be used in conjunction with the patient's clinical history and any previous analysis of appropriate family members. Unless specifically stated it is assumed that family relationships are as indicated. It is recommended that these test results be communicated to the patient in a setting that includes appropriate counselling.

Report electronically signed by:

Natalie Boruvka, BSc, MLT

Dimitri J. Stavropoulos, PhD, FCCMG, Clinical Laboratory Director, Genome Diagnostics

Received in Health Services
Date: 31/DEC/2019
Action Taken:
Signature:

Note: For clinical use only. Results not generated in a forensically accredited lab.

HFH93903

Patient Name: **WILLIAMS-MASSIAH, OMARI**

Order Number: **19-26564**
DOB (yyyy-mm-dd): 2008-08-25
Sex: M
MRN#: 5136451
External MRN:
HCN#: 7931682160 ON

Family Number:

Ethnicity:

Billing#: 19275216

Report Date: **2019/12/30 10:54 PM**
Specimen Type: Blood
Collection Date: 2019/12/05 6:15 PM
Registration Date: 2019/12/06 5:30 PM
Received Date: 2019/12/06 11:30 AM

FRAGILE X SYNDROME

Fragile X syndrome (FRAXA), the leading cause of inherited intellectual disability (ID), affects approximately 1 in 4,000 males and 1 in 8,000 females. FRAXA causes a range of symptoms. Early signs include delayed speech and language. Intellectual abilities vary from mild learning disabilities to severe intellectual disability. Behavioural characteristics can include autism, hyperactivity and poor eye contact. Physical features, such as a long face and large or prominent ears, are usually more noticeable in adults than children.

GENETICS

FRAXA is an X - linked disease caused by loss of expression of the *FMR1* gene. FRAXA is caused mainly by the expansion of the trinucleotide sequence CGG located in the 5' UTR of the *FMR1* gene. The abnormal expansion of this triplet leads to hypermethylation and consequent silencing of the *FMR1* gene. The trinucleotide repeat is inherited in an unstable fashion in fragile X families and displays intergenerational expansions.

WHO SHOULD BE TESTED

- Individuals of either sex with ID, developmental delay, and autism, especially if they have (a) any physical or behavioural characteristics of FRAXA, (b) a family history of FRAXA, or (c) male or female relatives with undiagnosed intellectual disability.
- Individuals seeking reproductive counselling who have (a) a family history of FRAXA, or (b) a family history of undiagnosed intellectual disability.
- Fetuses of known carrier females.

TEST SENSITIVITY

All cases of FRAXA caused by CGG expansion will be detected by this assay (~99% of patients with FRAXA). Rare cases (~1% of patients with FRAXA) are caused by pathogenic variants within the *FMR1* gene and will not be detected by this assay.

RISK OF POF & FXTAS

About 20% of women with a premutation have premature ovarian failure (POF), in which menstrual periods stop by age 40. About 30% of men, and some women, with a premutation have an increased risk of developing a disorder known as fragile X - associated tremor/ataxia syndrome (FXTAS). This disorder is characterized by progressive problems with movement (ataxia), tremor, memory loss, loss of sensation in the lower extremities (peripheral neuropathy), and mental and behavioural changes.

POTENTIAL OUTCOMES & INTERPRETATION OF TEST RESULTS

Repeat size	# of CGG repeats	Clinical Phenotype	Transmission
Normal	~5 to 44	Normal	Stably transmitted
Intermediate	~45 to 54	Normal	May increase in size in subsequent generations
Premutation	~ 55 to 199	Risk of POF & FXTAS	Risk of expansion to full mutation
Full mutation	Over 200	Symptoms of FRAXA	

DISCLAIMER

1. Current molecular testing may not detect all possible genetic variants for this disease. A negative test does not rule out the possibility of Fragile X.
2. The clinical course or severity of symptoms cannot be predicted by molecular analysis.
3. Test results should be interpreted in the context of clinical findings, family history and other laboratory data.
4. This test was developed and its performance characteristics validated by the Molecular Genetics Laboratory at the Hospital for Sick Children. It has not been cleared or approved by the U.S. Food and Drug Administration. The FDA has determined that such clearance or approval is not necessary. This test is used for clinical purposes.

Note: For clinical use only. Results not generated in a forensically accredited lab.

Patient Name: **WILLIAMS-MASSIAH, OMARI**

Order Number: **19-26564**
DOB (yyyy-mm-dd): 2008-08-25
Sex: M
MRN#: 5136451
External MRN:
HCN#: 7931682160 ON

Family Number:
Ethnicity:
Billing#: 19275216

Report Date: **2019/12/30 10:54 PM**
Specimen Type: Blood
Collection Date: 2019/12/05 6:15 PM
Registration Date: 2019/12/06 5:30 PM
Received Date: 2019/12/06 11:30 AM

FOR MORE INFORMATION

- The National Fragile X Foundation www.fragilex.org
- Online Mendelian Inheritance in Man http://www.ncbi.nlm.nih. gov/omim/ Item # 300624
- GeneTests online clinical information resource http://ww w.genetests.org/profiles/fragilex
- To locate a genetics center near you, please visit the Canadian Association of Genetic Counsellors website at www.cagc-accg.ca or the National Society of Genetic Counsellors website at www.nsgc.org
- SickKids Genomic Diagnostics Laboratory:www.sickkids.ca/genome-diagnostics

OMG1620LI/03

Note: For clinical use only. Results not generated in a forensically accredited lab.

Attached is a report from the Genome Diagnostics or Cytogenetics laboratory. For SickKids patients, a copy of the report will also be uploaded to the Epic patient chart. Please disregard the information below this line.

Environment Name: LIVE
Order Number: 190026564
Report Type: Test Specific Report for 021000P
Tests: 021000P
Delivery Reason: Attending Doctor, Requesting Doctor, Test Ordering Doctor RI_LEVINLE

CHILD AND ADOLESCENT NEUROPSYCHIATRIC ASSESSMENT

Mitesh Patel, BSc, MD, FRCPC; Forensic Psychiatrist,
Child & Adolescent Psychiatry
Mitesh.Patel@camh.ca

Confidential and for Medical Purposes Only

This report constitutes personal medical information and contains sensitive and confidential information. It is to be shared with the recipient to whom it is addressed and utilized for medical purposes only. As this report is protected under the Personal Health and information Privacy act, any sharing of information from this report must be approved by either the youth, where capable of making decisions regarding personal health information sharing, or the appropriate decision maker which may be the care provider or CAS worker. Should there be requests to share information from this report, we ask that this request be facilitated through the pediatrician to whom this letter is addressed.

Date: January 28, 2020
Dr. Leo Levin
Children's Aid Society
Toronto, Ontario

Received in Health Services
Date 05/FEB/2020
Action Taken Complete Rec
Signature

Re: WILLIAMS-MASSIAH, Omari
Date of Birth: August 25th, 2008
CPIN #: 30875413

Dear Dr. Levin,

Thank you for your referral of this pleasant 11-year-old boy who was seen here today in the company of his youngest sister, Nevaeh Williams-Massiah, as well as his foster mother, Pametta Edwards, whom he refers to as "Auntie Pam", as well as his CAS worker, Ms. Kalpa Patel.

I had the opportunity to explain the nature of the assessment to all parties present. Standard reporting criteria were reviewed. Consent was obtained to proceed.

Identifying Information:

Omari is now 11 years old and is residing in a foster care home located in Brampton, Ontario. He is 11 years old and is enrolled in the Royal Orchard Middle School in grade six. He has an IEP put in place.

In the home are his youngest sister, Nevaeh, as well as two other maternal sisters named Szodina and Mulekwa, age 5 and 8 respectively. He is the eldest in a sibline of four and is the only brother. Szodina and Mulekwa share a biological father and the other siblings do not. Currently, all siblings are placed in interim society care.

Background and Personal History:

As you are aware, there is a complex history of neglect and trauma. Two of the youngest siblings were reportedly discovered in the community at night and following this, all children were brought into care. There were noted to be significant concerns regarding alcohol abuse and possible difficulties with gambling. There may also have been exposure to drugs in the home. Mother was noted to have a history of being in care herself and there is a history of intergenerational trauma. The grandmother also provided care and there were concerns apparent regarding level of care provided.

There had been a history of CAS involvement prior to the most recent apprehension, which occurred in August of 2019. This is the children's first time being placed in foster care and they are due to return to court in March, at which time the two eldest may enter a kinship care arrangement with an aunt named Zeticia Gill and the youngest two children may move to Nevaeh's biological father. Further plans are not fully confirmed.

The biological mother reported alcohol intake over the course of all pregnancies to Ms. Kalpa Patel. All four children were thus exposed to alcohol prenatally. She informed that she did not learn of each of the pregnancies until well into the gestation prior. Omari was her first child and she reportedly was not aware of the pregnancy until four to five months in. She reportedly drank throughout much of the pregnancy leading up to her discovery that she was pregnant. There were concerns expressed regarding a chronic history of alcoholism. The specific quantities and duration of use were not described fully.

Past Psychiatric History:

There have been no admissions to hospitals nor have there been other psychiatrists involved. There have been no further psychiatric diagnoses provided. The only physician involved thus far has been yourself and Dr. Cohen.

Past Medical History:

There is a history of a "coffee burn" in which Omari indicated, "When I was with my mom, I burned myself and had to go to hospital... it was in 2016". He revealed a keloid scar on his chest which resulted as a consequence of this burn.

Medications:

He is not prescribed any medications at this time. There is no history of psychotropic medications.

Allergies:

There are no known drug allergies.

Social History:

He enjoyed playing basketball and running. He also liked "being in heat" and specifically wanted to attend tropical climates. He identified his three wishes as "I could be like a basketball player Kobe Bryant; I want to verse [compete against] Lebron James; I want to go to Jamaica". He noted that he had not ever been to Jamaica previously. He also stated that he was of part Jamaican descent and that he had a grandparent who was Jamaican. As per your noted, there are no further details regarding his father or a history therein.

Omari has a CYW who takes him out every Tuesday and also plays basketball with him at those times. Omari is not enrolled on a sports team at this time. Omari is the only youth in the home who has a CYW established for his care.

Functional History:

Omari was noted to do quite well in the home. He was described as listening well and he stated that he would "help out and do everything around the house". He was described as "always smiling" and was also noted to be quite clever. He was also noted to care a lot for his sisters and spoke about needing to protect them and there was some role reversal apparent.

He was described as being highly oppositional at the outset of his stay in care. It was noted by his foster mother, "He didn't like to obey the rules and he is getting better from when he came in... much better from the last several weeks". He was noted to have had a temper before but there have been ongoing improvements. Omari informed his foster mother that "He told me he doesn't like it if I shout so I change my techniques of talking to him and that has changed him a lot".

He was noted to have some difficulties expressing certain feelings. He was noted to have presented with significant amounts of information which related to his ongoing stay in care and potential transfer out of care.

His mood was described as remaining otherwise quite good. At times he displayed significant mood changes and he was noted to become quite angry quite quickly. Foster mother stated, "After five minutes, it's like he's a different person".

His appetite was described as being "too good" and he was eating well. He had engaged in some fights at school and also indicated that was being picked on by others. At school, he had also been "walking up and down and pacing" and had refused to enter the classroom on occasion. The school has become increasingly concerned about such behaviours. The genesis of these behaviours remains unclear but is likely related to increased anxiety and tension regarding possible transfer out of care. He was noted to be immensely inattentive and hyperactive over the course of the day. He was also described as experiencing significant language delays and was noted to speak at a level that was much lower than anticipated for his age. He continued to have difficulties communicating his thoughts, particularly when these were more complicated or involved various subjects.

He was not noted to have any night terrors. He would sleep well throughout the night and was described as being a "security guard" who would watch over the family at night and would not go to bed until all others were upstairs sleeping. There was no history of overt nightmares or flashbacks.

Mental Status Examination:

Omari presented as a well-groomed boy who looked his stated age. As per your examination completed November 26th, 2019, he is noted to be dysmorphic. His intercanthal distance was 4 cm and he was described as having a transverse crease of his ears. He has a flat philtrum and the score provided is 2 – 3. Overall, his faces was in keeping with that of sentinel facial features suggestive of FASD. His speech was of normal rate and tone. There were minor articulation difficulties. He did not speak at a level anticipated for his age in terms of complexity of work usage. His affect was bright. There were no perceptual abnormalities noted. He followed conversation well; however, he appeared somewhat more confused when more complex themes were discussed. Insight and judgment were age appropriate. He did not have a history, nor did he voice any thoughts of wanting to harm himself or harm others.

Impressions and recommendations follow below; these have been separated so that a copy can be detached from this report and provided to other members in the Circle of Care:

CHILD AND ADOLESCENT PSYCHIATRIC ASSESSMENT

Mitesh Patel, BSc, MD, FRCPC; Forensic Psychiatrist,
Child & Adolescent Psychiatry
Mitesh.Patel@camh.ca

Confidential and Personal

Date: January 28, 2020

The following is taken from a psychiatric report prepared by Dr. Mitesh Patel in relation to the client identified above. Consent should be obtained to share these impressions and recommendations. Please consult with the referring pediatrician or family doctor at CAST to review these and other findings from the assessment.

Psychiatric Impressions:

1. **Neuropsychiatric Assessment**

   As indicated above, Omari has a positive history of maternal alcohol intake as confirmed by the CAS worker. The specific amount and period of ingestion is relatively unknown; however, biological mother was unaware of the pregnancy until well into the gestation period (between four and five months) and had consumed alcohol throughout. Furthermore, he has facial features that are suggestive of alcohol exposure in utero.

   He is determined to meet criteria for impairment across various neurodevelopmental delays including cognition, language, academic achievement, attention, executive function, hyperactivity, affect regulation and social communication. Further psychological testing in these areas will potentially confirm deficits; however, given the number of deficits and the facial features as well as the biological history, it is my impression that he meets criteria for **fetal alcohol spectrum disorder (FASD) with sentinel facial features.**

2. **Other Psychiatric Diagnoses**

   Omari is at high risk of experiencing ongoing inattentiveness and hyperactivity that are in keeping with attention deficit hyperactivity disorder. He should continue to be monitored for further symptoms in this regard. Furthermore, he has a history of oppositionality though has responded well to further behavioural changes and communication techniques used in the home. He does not meet criteria for an externalizing disorder at this time.

   He will likely meet criteria for underlying learning disabilities and further psychological testing in this regard will provide further details in this regard.

Recommendations:

1. **Psychopharmaceutical Interventions:**

   I do not recommend any psychopharmaceutical interventions at this time.

2. **Psychotherapeutic Interventions:**

Omari has done well with some limited behavioural interventions provided in the home environment. He is reportedly experiencing some difficulties at school and further implementation of improved communication techniques as well as other behavioural interventions would be beneficial in this environment. The school may have behavioural therapy available to further inform Individualized Education Plans that could be utilized in this setting. Behavioural interventions should be applied across the home and school environments in order to improve effectiveness therein. His level of oppositionality does not remain heightened at this time; however, he remains at risk of this moving forward.

3. **Social Interventions:**

Omari has a significant interest in playing basketball. It would be beneficial for him to be enrolled in an organized sports team. It is helpful for him to continue with his CYW and to engage in sporting activities with the CYW on a one to one basis. It would also be helpful for him to engage in with other youth and to develop further mentorship relations with other prosocial individuals.

Furthermore, he expresses a significant interest in Jamaican culture and would benefit from further education and exposure to cultural events in this regard. His foster mother is of Jamaican descent and has been involved in exposing him to his culture in this way. Further engagement with a mentor of Caribbean descent, specifically Jamaican descent would be beneficial.

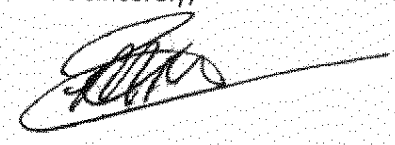
4. **Complex Case Review**

I understand Omari's case will now be reviewed by the Complex Case Review team and it will be beneficial for further recommendations and interventions to be provided moving forward. It is also anticipated that he will be seen on at least a yearly basis to provide further updates and treatment recommendations in keeping with a changing psychiatric profile, which is in keeping with his underlying neurodevelopmental condition.

I trust that the above is helpful to you. Should you have any questions, please feel free to contact Meaghan Coulter at 416-924-4640 ext. 2044 who will then assist with further appointments and contacts.

Thank you for involving me in the care of this patient.

Yours sincerely,



Mitesh Patel, MD, FRCPC
Forensic & Child and Adolescent Psychiatry
Assistant Professor, Faculty of Medicine, University of Toronto
Forensic Staff Psychiatrist, Law and Mental Health Forensic Unit,
Centre for Addiction and Mental Health
Outreach Adolescent Psychiatrist, Turning Point Youth Shelter, Inner City Health Associates

Child Psychiatric Consultant, Children's Aid Society of Toronto