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Attitude of pediatricians toward establishing a “Down Syndrome Clinic”

Ahmed Juma Nasib Al Farsi

Abstract

Background: Down syndrome (DS) is the commonest chromosomal abnormality in human, and one of the commonest genetic causes of intellectual disability. ⁽¹⁾ The syndrome cost the families and health care system a lot. The patient of DS needs multi-disciplinary team approach. In 2015, Oman Ministry of Health (MOH) published informative DS guideline booklet. ⁽⁷⁾ However, the implementation of that guideline in the current clinical practice is still suboptimal, hence our study aims to evaluate the attitude of the physicians in the child health department particularly in Ash Sharqiyah South Medical Governate (SSMG) toward establishing a new clinic operated specifically for DS patient.

Methods: In this cross-sectional study, questionnaires distributed to evaluate the current service, and to explore the attitude of the physicians toward establishing a new clinic for DS patients. Data processed SPSS version 29. Sample size included all pediatricians SSMG hospitals.

Results: 42 doctors working in Child Health Department (CHD) included in the study. 40 (95%) agreed that there is no current DS clinic in South Sharqiyah governorate. All (100%) agreed that a new well-structured clinic is needed. About (19%) of the physicians are not aware about the MOH guidelines for DS and (12%) only who follow exactly the MOH DS booklet recommendation.

Most of the physicians (61%) agreed that Sur polyclinic is the most suitable place for such clinic.

Conclusion: Our study showed that the current down syndrome follow-up system is suboptimal, and establishment of a new, well-structured clinic designed and operated specifically for DS patients in Sur polyclinic, following the national and international guidelines is highly recommended.

Keywords: Down syndrome, follow up clinic, genetic counselor, Oman

Introduction

In 1866, Down syndrome (DS), formally called trisomy21, was first identified and named after John Langdon Down who described its clinical features. ⁽¹⁾ Down syndrome has a clinical and psychosocial impact on the baby and the family, and hence it needs unique care and multidisciplinary approach. It is considered as the most frequently occurring chromosomal abnormality in humans ⁽¹⁾ and well known of causing intellectual disabilities.

Genotype: Down syndrome occurs due to an extra copy of the whole or part of chromosome 21 in all or some cells of the body. Trisomy 21 karyotype is 47, XX, + 21 for females and 47, XY, + 21 for males ⁽²⁾. There are 3 types of DS; first and most common type, almost (95%) of the cases, is non-disjunction, which is caused by failure of maternal chromosome 21 to separate more frequently during first meiotic division. Second type is caused by chromosomal translocations which make up nearly (3-4%). In this type, a segment of chromosome 21 is transferred to another chromosome, usually chromosome 14,15 or 21. The last type is called mosaicism and it's the rarest type, less than (2%) of the cases. In this uncommon type, the third copy of chromosome 21 is present in some but not all cells. This type of DS is caused by abnormal cell division after fertilization. ⁽¹⁾ In mosaicism, one set of cells may have normal chromosomes and another type may have trisomy 21. ⁽¹⁾

Phenotype: DS is a multisystem disorder that affects the individual physically, medically and psychologically. The physical; head, face and neck features ^(3,4,5,6) include brachycephaly, round face, short neck, low-set, small ears, flat nasal bridge, microgenia, macroglossia due to small oral cavity, almond shape of the eyes caused by an epicanthic fold of the eyelid and

oblique palpebral fissures. Other features may include shorter limbs, a single transverse palmar crease, lax ligaments, excessive space between large toe and second toe called sandal sige, dry skin, muscle hypotonia, brachydactyly and short fifth finger with clinodactyly. Ocular and visual features include high refractive errors, amblyopia and strabismus, accommodative and vergence anomalies, ptosis, blepharitis, nasolacrimal duct obstruction nystagmus, keratoconus, speckling of the iris; Brush field's spots, cataracts, glaucoma and retinovascular anomalies. Mental impairment which many be mild; IQ 50-70, moderate; IQ 35-50 or severe; IQ 25- 35. ⁽⁷⁾

Medical conditions associated with Down Syndrome

Cardiac anomalies: occurs in (40-60%) of DS patients.⁽⁷⁾ Of these (30-40%) are complete Atrio Ventricular Septal Defects (AVSD) which requires surgical correction usually around 4-6 months before it cause pulmonary vascular disease (PVD) which is more common in child with DS and AVSD.⁽⁷⁾ Other cardiac defects include atrial septal defect (ASD) and ventricular septum defect (VSD).⁽³⁾ They also might develop asymptomatic mitral valve prolapse (MVP) especially during adolescent period which might progress into mitral valve regurgitation (MVR).⁽⁷⁾

Hearing loss: more than (50%) of DS children have hearing impairment. ⁽⁷⁾ Most common cause being otitis media with effusion (OME) causing conductive hearing loss. other major problem is obstructive sleep apnea. It has been reported that about (33%) of DS patients will require ENT surgery to address tonsils and adenoids. ⁽⁷⁾ Furthermore, DS children have a higher incidence of sensorineural deafness. ⁽⁷⁾ They have also narrow ear canal which predisposes to accumulation of wax.

Vision: Approximately (60%) of individuals with DS have visual disorders. Congenital cataract is ten times higher among them compared to the general population. ⁽⁷⁾ Refractive errors can start from early childhood; keratoconus may develop in teenage years or later.

Thyroid: hypothyroidism is very common in DS children; those children have 28 times more frequent thyroid disease compared to the general population ⁽⁷⁾. More than (12%) of adults with Down syndrome have thyroid disease. ⁽⁷⁾ Other endocrinopathies are hyperthyroidism, obesity, diabetes and impaired stimulation of growth hormone. ⁽³⁾

Leukemia: There is (1%) chance of occurrence of Leukemia in individuals with Down syndrome. The most common type among children is acute lymphoblastic leukemia (ALL) and a subtype of acute myeloid leukemia (AML). ⁽⁷⁾

Gastro Intestinal Disorders: There is a (12%) chance for individuals with Down syndrome to suffer from gastrointestinal atresia. Around (2%) of these children may suffer from Hirschsprung disease. ⁽⁷⁾ other common finding is gastro-esophageal reflex ⁽³⁾

Cervical spine disorders: About (10-20%) of individuals with DS may develop cervical spine disorders radiologically and around one fifth of those with the radiological findings may have symptoms (e.g., atlantoaxial subluxation). ⁽⁷⁾

Hip dislocation: About (6%) of DS children are at risk of acquired hip dislocation. ⁽⁷⁾

Cognitive: Mainly intellectual disability which affects learning, memory, and language that leads to a range from mild to profound impairment in intellectual functioning. ⁽³⁾

Neurology: Developmental delay, early onset dementia and Alzheimer's disease. ⁽³⁾ **Neuropsychiatric/ behavior:** Epilepsies, autistic spectrum disorder, depressive illness, attention deficit hyperactivity disorder, conduct oppositional disorder, or aggressive behavior reported in about (5.4%). ⁽³⁾

Immunological: Immune dysfunction, autoimmune disease such as arthropathy, vitiligo, alopecia and increased susceptibility to infections. ⁽³⁾

Dermatological: Dry skin and folliculitis ⁽³⁾

Reproduction: Impaired fertility both genders; males are usually unable to father children, while females have fertility and birth problems including miscarriages, premature births and difficult labor. ⁽³⁾

Dental: caries and malocclusion.

inheritance of DS: Most cases of DS are not inherited ⁽⁸⁾, the chromosomal abnormality occurs as a random event during the formation of reproductive cells in a parent. The incidence of life births with DS increases with the age of the mother ⁽⁸⁾. However, due to higher fertility rates in younger women, the probability of having a child with DS increases with the age of the mother and more than (80%) of children with DS are born to women under 35 years of age ⁽¹⁾. People with translocation Down syndrome can inherit the condition from an unaffected parent. ⁽⁸⁾ The parent carries a rearrangement of genetic material between chromosome 21 and another chromosome. This rearrangement is called a balanced translocation. No genetic material is gained or lost in a balanced translocation, so these chromosomal changes usually do not cause any health problems. However, as this translocation is passed to the next generation, it can become unbalanced. People who inherit an unbalanced translocation involving chromosome 21 may have extra genetic material from chromosome 21, which causes Down syndrome. The chance of passing the translocation depends on the sex of the parent who carries the rearranged chromosome 21. If the father is the carrier, the risk is around 3 percent, while with the mother as the carrier, the risk is about 12 percent. ⁽¹⁾ Like trisomy 21, mosaic Down syndrome is not inherited. It occurs as a random event during cell division early in fetal development and as a result, some of the body's cells have the usual two copies of chromosome 21, and other cells have three copies of this chromosome. ⁽⁸⁾

Why is this study important? Because of the multi-system and multi-organ involvement nature of DS, and because of the Physical, Medical, Phycological, and social impact of this syndrome on the patients and their families, a good mature well organized and well-structured follow up system is needed to deal with all of the disease aspects, to involve actively a multidisciplinary team, and to provide

up-to-date standard of care management plans. My quality project will scope on the current practice, in a particular Medical Governorate region of Oman (hence it can be considered as pilot study which can be expanded in the future) and will emphasis the need for a better follow up system actively and effectively implemented to the clinical practice which definitely will help to improve the quality of care of DS patients

Literature Review

Internationally, the birth prevalence of Down syndrome is approximately 1 in 700 to 1 in 1000 ⁽⁹⁾ depending on maternal age, and prenatal screening schedules. In Oman, Prevalence of down syndrome is 1:350 live births. ⁽⁷⁾ It is 2.3 down syndrome case per 1000 population in the year 2010, which leads to about 120 affected births per year ⁽⁷⁾. The numbers vary according to the regions of Oman, for example in 2012 the DS cases all over Oman were 141 cases ⁽⁷⁾, distributed as follow; 46 cases from Muscat, 18 cases from Ash Sharqiyah South, 15 cases from North Al Batinah North, 14 cases from Al Batinah South, 14 cases from Dhofar, 13 cases from Ad Dakhiliyah, 10 cases from

Ad Dhahirah, 8 cases from Ash Sharqiyah North, and 3 cases in Al Buraimi. ⁽⁷⁾ No cases were reported from Musandam, possibly due to suboptimal reporting system at that time or possibly people in that area used to go to United Arab Emirates for treatment as its nearby country and geographically easier for them. The third possible explanation is that the population number is little in Musandam compared to other regions of Oman. ^(Figure1) In South Sharqiyah, where we conducted our study, there were 15 cases diagnosed with DS in 2012⁽⁷⁾, and the prevalence reported was 1: 353 ^(Table1). After that, according to the latest annual report in 2019 released by South Sharqiyah Medical and Health Governorate child and mother health section, 22 new cases diagnosed with DS in 2019, and the number of all cases registered by 2019, including the new cases, was 82 patient, ^(Table2) among these, 26 patients had cardiac anomaly by echocardiography, 3 of them had ophthalmic disorders, 2 of them developed hypothyroidism, 1 was underweight, 1 had developmental delay, 1 had renal pathology and 1 had vascular complication. ^(Table1)

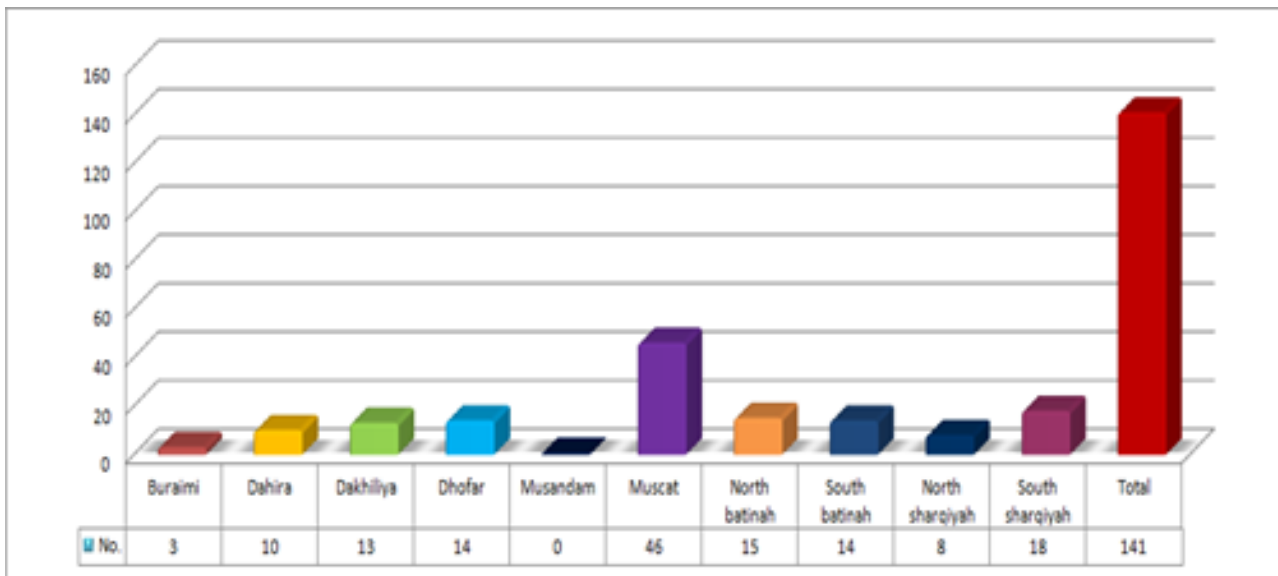


Fig. 1: Geographic Distribution of Down Syndrome cases among Oman regions, Year 2012.

Musandam	1:514
North batinah	1:625
South batinah	1:253
Muscat	1:294
North sharqiyah	1:358
South sharqiyah	1:600
Wousta	1:355
Dhofar	1:460
Dhahira	1:351
Dakhliyah	1:283

Table 1: Prevalence of the Down Syndrome among Oman Governorates, Year 2010.
Annual Report of Children with Down syndrome (MCH Coordinator)

Governorate: South Sharqiyah. Year2019. MCH Coordinator: [redacted]

Institution Name	Focal point		Frequency of clinic per month	Chromosomal analysis		Number of newly diagnosed cases	Total # of children registered in the clinic (including newly diagnosed cases)	Total # of registered cases					*Age-appropriate Health Assessment	
	Doctor	Nurse		Done	Not done			Age group			Gender		completed	Not completed
								0-5	6-12	13-18	Male	Female		
Sur hospital / down's syndrome clinic (genetic clinic)	4	2	All days	82	0	22	82	47	10	25	35	47	82	0
Total	4	2	All days	82	0	22	82	47	10	25	35	47	82	0

Total number of cases with comorbidities (per governorate)	Cardiac Based on ECHO	Ear & Hearing	Ophthalmic	Thyroid problem		Diabetes	Hematological problems	Growth			Developmental delay		Gastrointestinal problems	Skeletal	Others
				Hypothyroidism	Hyperthyroidism			Under weight	Normal weight	Overweight/obese	Yes	No			
Total	26	0	3	2	0	0	0	1			1				1 renal 1vascular

* Age-appropriate Health Assessment e.g.; hearing, vision, cardiac, endocrine.... etc, according to the follow up/screening sheet

Table 2: Annual report of DS cases in South Sharqiyah Medical Governorate, Year2019.

Clinical gab

Despite the availability of a written clinical guideline published by the MOH since 2015 (Figure2)(7), which is applied to all health care institutes in Oman, and despite the presence of the newborn card which contain informative time-line table (Table3) for clinical examinations and schedule of different investigations in different times for DS patients, scoping the clinical field showing that the current practice in this regards is still suboptimal; some hospitals in the studied region has no trained doctors to conduct the care of this particular group, other institutes don't have a regular clinic or follow up system. This practice gap has not measured properly by research before the date this study is running as far as I know. No doubt that DS patient needs a particular laboratory and clinical evaluation in a timely manner, starting from antenatal until the adult life,

they need specific follow up in deferent specialties and subspecialities. Lacking of such follow up system will definitely compromise the care and hence the quality of life for this population. Furthermore, while some Medical Governorates in Oman have already established well-structured clinics for DS, there are 4 clinics in fact, up to the time this thesis written, one is in Sohar Polyclinic covering Al Batinah North region, the second is in Nizwa Polyclinic covering part of Ad Dakhiliyah region, the third is in Bahla Polyclinic covering other part of Ad Dakhiliyah region, and the last one is located in Bousher Polyclinic covering Muscat DS patients, in the other regions and governorate this service is not yet established, including Ash Sharqiyah South Medical Governate where we conduct our project. In fact, there might be an allocated doctor and possibly nurse to look after DS patients, however, there is

no specific clinic operated only for DS patients and there is no regular follow-up system and most of the cases are seen as an acute presentation or in a random follow up appointment.

Sur Hospital is a secondary level hospital, the main health institute in the whole Ash Sharqiyah South Medical Governate, cases being referred to Sur Hospital from 3 other main hospitals in the governorate, Jalan BB Ali Hospital, Jalan BB Hasan Hospital, and Masira Island Hospital. They refer to Sur Hospital as they lack the basic

teams required for DS patients' assessments and follow up like cardiology team, ophthalmologist, endocrinologist and other multidisciplinary team members.

The hypothesis of this project is that Down syndrome Clinic is needed in South Sharqiyah governorate to help in proving standard of care medical practice DS patients. The results and recommendation will be provided to the decision makers. This might help establishing DS clinic not only in Sur Hospital, but to the other regions of Oman lacking such service.

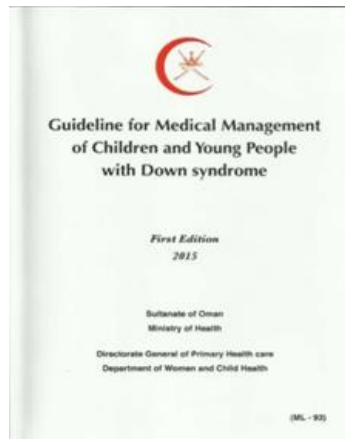


Fig. 2: The Ministry of Health Guide line of treating DS patients, 2015.

Follow up table

Health Assessment	At birth	3/12	6/12	One yr	3 yrs	5 yrs	7 yrs	9 yrs	12 yrs	16 yrs	18 yrs
Chromosomal analysis	√	√ If not done									
Cardiac examination (according to protocol)	√	√	√	√	√	√	√	√	√	√	√
Ears & Hearing	√ echo hearing test	√	√	√	√	√	Complete audio logical assessment	√	√	√	√
Ophthalmic evaluation by history	√	√	√	√	√	√	√	√	√	√	√
TFT should done annually	√			√	√	√	√	√	√	√	√
CBC	√			√	√	√	√	√	√	√	√
RBS should done annually	√			√	√	√	√	√	√	√	√
Counseling and parental support	√	√	√	√	√	√	√	√	√	√	√
Anthropometrics measurements	√	√	√	√	√	√	√	√	√	√	√

Table 3: Newborn Card for Down Syndrome patient Follow up plan Ministry of Health, Oman, 2013.

Objectives

- To investigate the current Down Syndrome, follow up practice in Ash Sharqiyah South Medical Governate hospitals, looking for areas of improvement.
- To evaluate the attitude of pediatricians toward establishing Down Syndrome Clinic.
- To study the practical details of establishing the suggested Down Syndrome Clinic
- To explore the challenges of establishment Down Syndrome Clinic.
- To open the door for any future projects providing holistic approach for DS patient and their families meeting the stander of care worldwide.

Methodology

After getting the ethical approval for the research proposal from the development and research committee in Ash Sharqiyah South Medical Governate, researcher started to enroll the participant of the study, inclusion criteria were defined clearly that the participant should be a doctor who is working in Child Health Department in the time of the study, October 2019, excluding those who are not physicians or haven't work in pediatrics clinic in the time of the study (Figure3) The study duration was planned to be three to six months from the idea until the manuscript ready for publication.

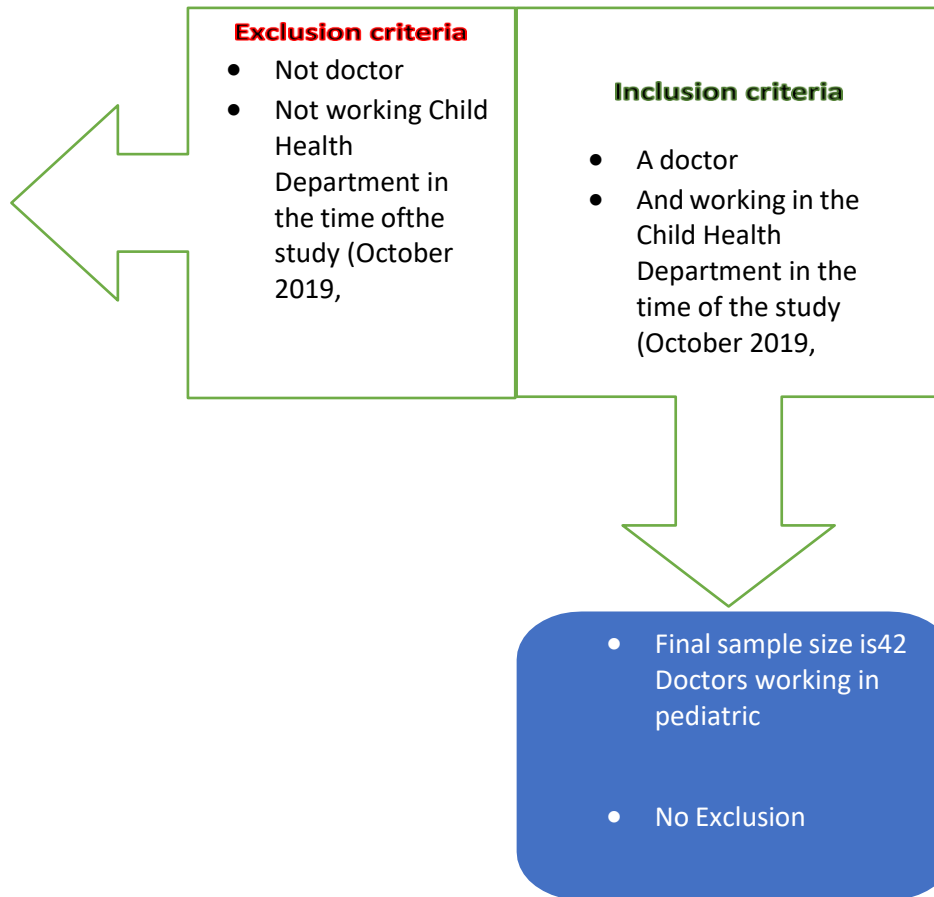


Fig. 3: recruitment of the participants.

The study designed to be a cross-sectional descriptive quantitative study, its a quality improvement project which aim at the end to improve the current practice. Informative Consent form (appendix1) is optioned to enroll in the study. Confidentiality assured and participant information is securely. Participants were aware that they have the right to refuse to participate or to withdraw from the study at any stage. Sample size included all pediatricians in Four hospitals; Sur Hospital, Jalan BB Ali Hospital, Jalan BB Hasan Hospital, and Masirah Island Hospital. A questionnaire was distributed, there was no specific randomization method in this particular study as the investigator distributed the questionnaire to the whole number of the doctors in all pediatric department in the 4 hospitals so everyone was included. The questionnaire has three parts (Appendix2) the first part is to evaluate the current available service, and the second part is to explore the

doctor’s attitude toward establishing a new clinic for DS patients, and third part is to collect suggestion and ideas regarding the details of that clinic. Finally, the answers, information and data were collected and transferred from the paper sheets to an Excel sheet (Microsoft 2019) for processing and analyzing the data, at this stage data treatment was done to ensure information accuracy and to avoid any human error, SPSS.29 version was used for data analysis and obtaining the figures and charts.

Results

42 doctors (n=42) working in pediatric departments in 4 different hospitals were enrolled at the beginning of the study. 23 doctors from Sur Hospital (55%), 11 doctors from Jalan BB Ali Hospital (26%), 4 doctors from Jalan BB Hassan Hospital (9.5%), and 4 doctors from Masirah Island Hospital (9.5%). (Figure5)

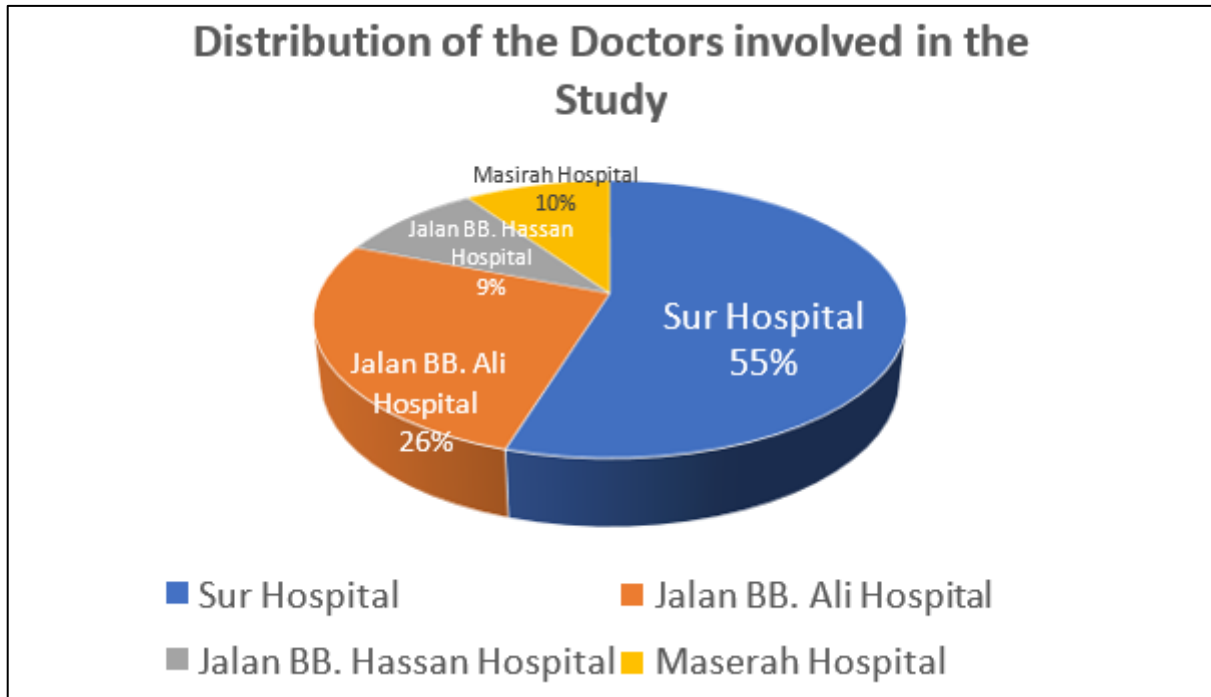


Fig. 5: Distribution of the Doctors involved in the study.

Regarding the relevant characteristic of the participants, all (100%) are doctors, and all (100%) are working in Pediatric Departments, 2 (5%) are consultants, 12 (28%) are

pediatric specialists, and 28 (67%) are medical officers working in Child Health Department. (Figure6)

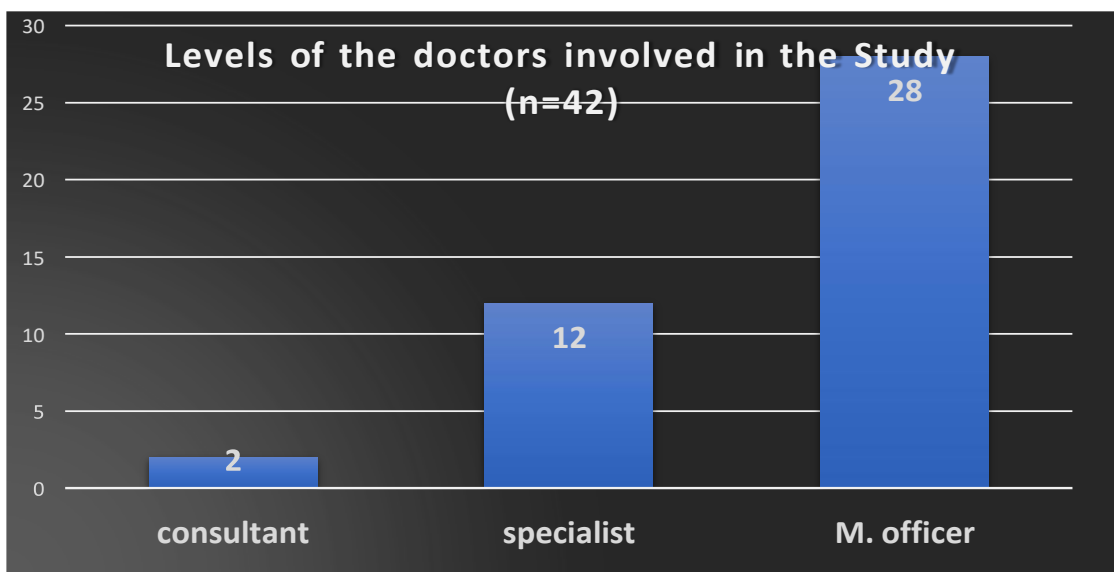


Fig. 6: Levels of the doctors involved in the Study (n=42).

Regarding the years of experiences, about (24%) of the participants worked in Ministry of health in Oman Child Health Departments for more than 10 years, while (40%) worked for 5 to 10 years period, and (36%) have less than 5 years' experience. All of the participants (100%) have been involved in treating DS patients. Evaluating the load of DS patients on the health service, about (14%) of the participants doctors see more than 5 DS patient per month, while (55%) of them they see 1 to 5 patients per month, and (31%) of the participants see less than one DS patient per month. Most of the DS patients, according to (71%) of the participants end up to the pediatric clinic when they are sick because of acute illness not related to regular follow up

system, however about (27%) of the participants answered that DS patient end up to their clinic as a follow up appointment, and (2%) as a referral from other clinics or other hospitals. About (97%) of the participants believes that currently at the time of this project there is no specific clinic for DS in their institute or in another institute in South Sharqiyah Medical Governate catchment region, while only 2 doctors believed that there is currently ongoing clinic (3%). Results showed that about (83%) of the participants were aware of the presence of a guide line and protocols booklet (Figure2) provided by Ministry of Health in Oman, while (17%) of the participant were not aware of the presence of such guide line, (Figure7)

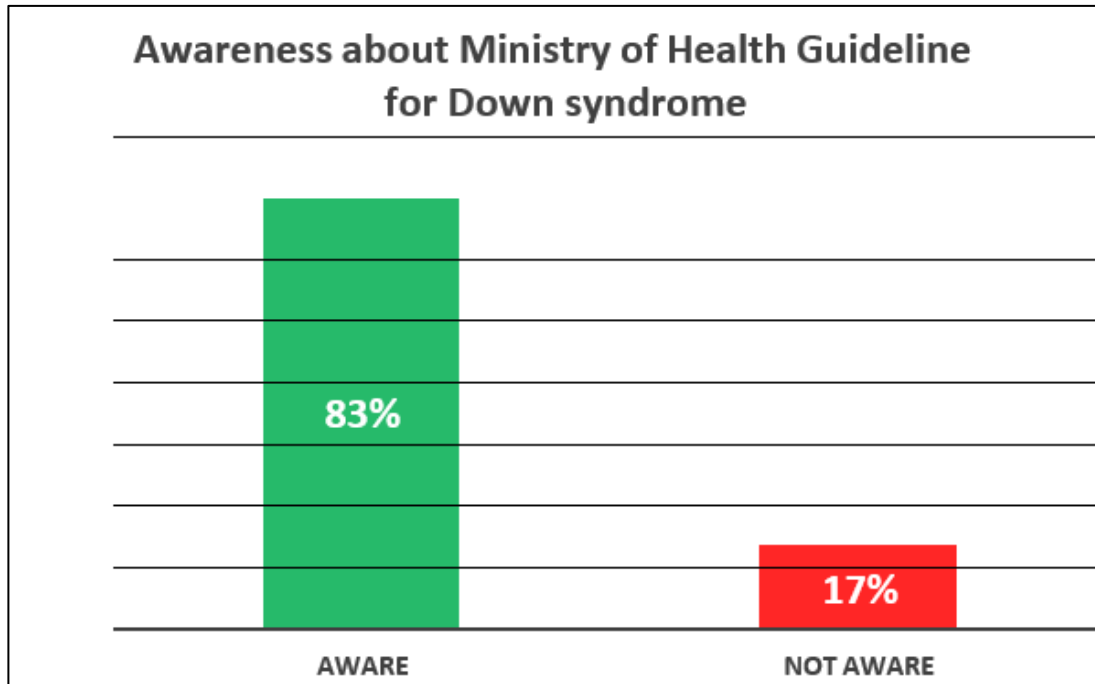


Fig.7: Awareness about Ministry of Health Guideline for Down syndrome.

Evaluating the real practice by our questionnaire revealed that only (19%) of the participant are following exactly and strictly the Ministry of Health Guideline when they treat DS patients in planning the follow ups and referral to subspecialty teams. (24%) of the participants follow the Guideline most of the time, (21%) they following it (50%) of the time, (24%) often not following the Guideline, and (12%) of the participant are not following the Guide line at

all. (Figure8) Similarly, only (17%) of the participant are following exactly and strictly the Newborn Card schedule of treating Down Syndrome. About (31%) of the participants follow the Guideline most of the time, about (19%) are following it 50% of the time, and about (24%) often not following the Guideline, while (9%) of the participant are not following the Guide line at all. (Figure8)

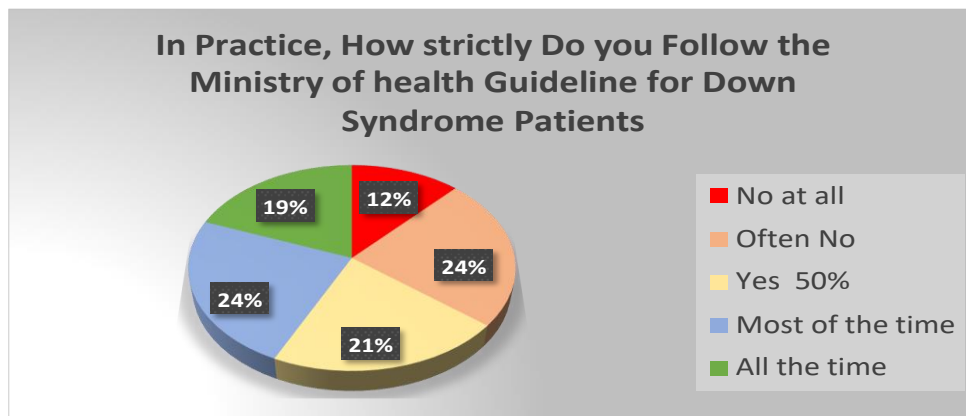


Fig. 8: In Practice, how strictly Do you Follow the Ministry of Health Guideline for Down Syndrome Patients.

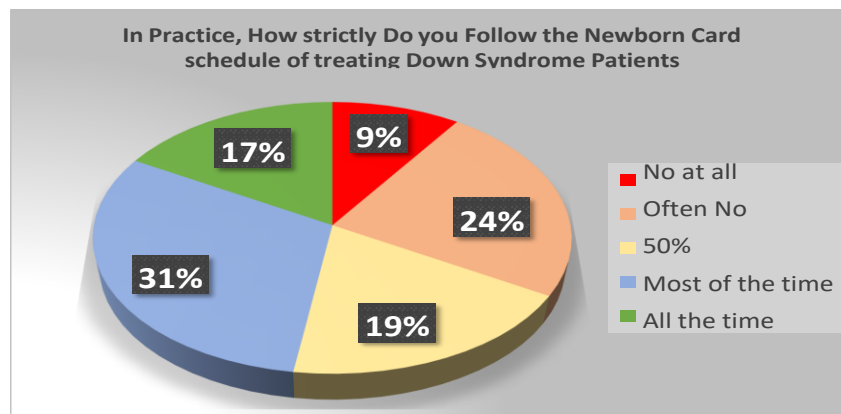


Fig. 9: In Practice, how strictly Do you Follow the Newborn Card schedule of treating Down Syndrome Patients.

Seconds part of the questionnaire designed to explore the attitude toward establishing new clinic only for DS patients. Among the participants, 31 doctors (71%) agreed that DS is really needed in the governorate, while 12

doctors (29%) felt that establishing new clinic for DS in not necessary and they are satisfying with the current service. (Figure10)

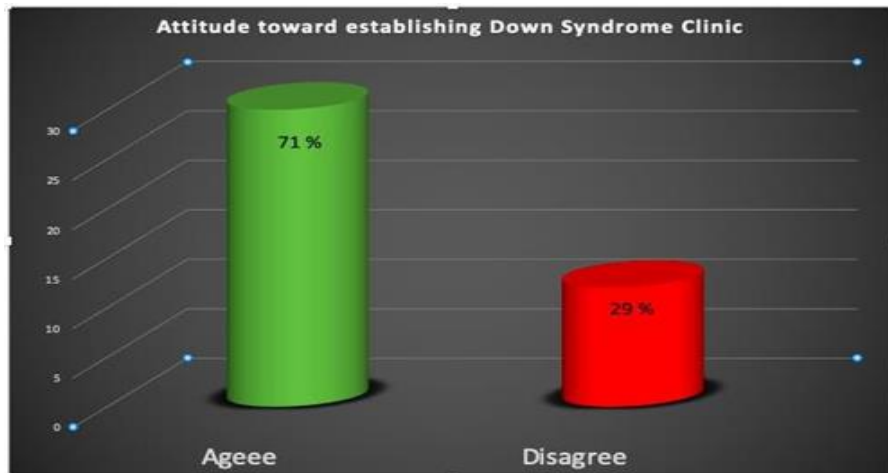


Fig. 10: Attitude toward establishing Down Syndrome Clinic.

Regarding the suggested place of the clinic, most of the participants (61%) agreed that Sur Hospital is the best place of such clinic as Sur is the center of the Governorate and has subspecialty teams needed for DS care which might not be available in the other Hospitals. However (37%) of the participant have different idea, they agreed that the clinic should available in the place that most of the patients coming from, and that means we need another quantitative

assessment of the patient’s distribution and we might need more than on clinic which might be non-feasible at this time. Anyhow that is beyond the scope of this Project. 2% of the participants agreed that the clinic should be in Jalan BB Ali. (Figure11). All participants (100%) agreed that the clinic follow up should follow the MOH guideline and newborn screening card schedule.

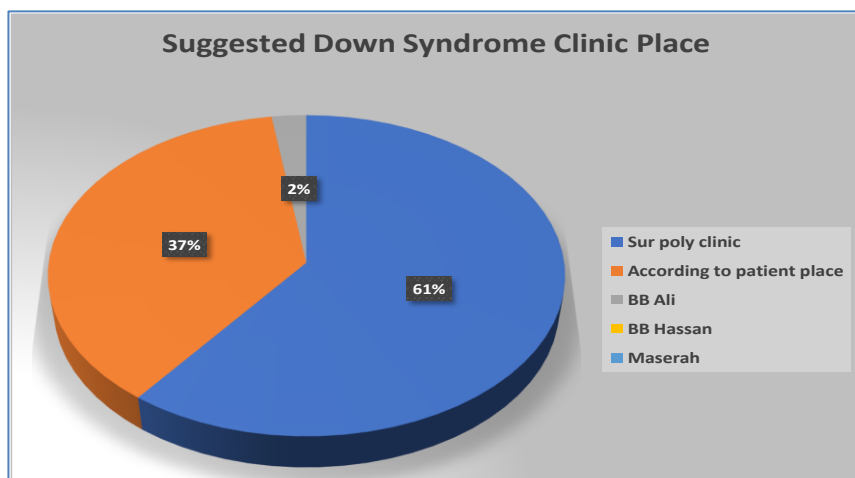


Fig. 11: Suggested Down Syndrome Clinic Place.

About (90%) of the participants agreed that the clinic should be operated by a pediatrician with a special training how to deal with DS patient in particular, by special training we mean that he or she received a training from National genetic center at Royal Hospital, or from Ministry of Health directly. In the other hand, (10%) agreed that the

clinic can be run by any pediatrician, and special training is not necessarily needed. (Figure12) However, in another question, 100% of the participants agreed about that DS training workshops is needed for Ash Sharqiyah South Medical Governate doctors prior to establish this clinic.

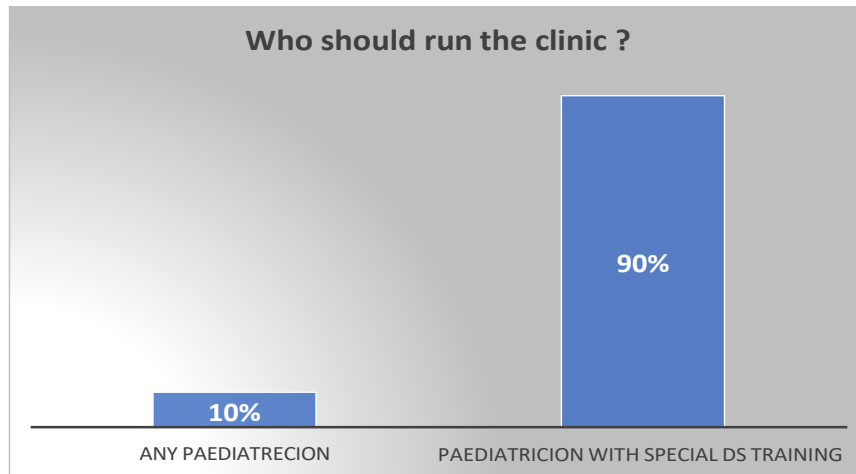


Fig. 12: Who should run the clinic?

At the end of the Questionnaire, there was an open question regarding the difficulties and challenges facing the establishment of such clinic, most of the answers were around shortage of staff and human resources, and the high load and busy nature of the current daily clinics so difficult to free specific doctors for this clinic. The second challenge was social, the family's awareness in Ash Sharqiyah South region regarding down syndrome need to be advocated by health education and public awareness campaigns before starting this clinic, to prepare people to be ready and really understand the importance of follow up in DS, because even if follow up process initiated you don't want to end up with families skipping the appointments. The last Challenge is the clinic place, as all the pediatric clinics are busy treating the high load of the acute pediatric presentations.

Discussion

Down syndrome is It is the most prevalent genetic disease worldwide and the common genetic cause of intellectual disabilities, ⁽¹⁾ it causes a huge medical and social cost. Down syndrome patient and his family suffer not only from medical complications associated with syndrome, but also from the psychosocial impact of the disease and the need of subspecialty like special education, rehabilitation, physiotherapy and other services. A multidisciplinary team approach is the ideal way to treat DS patients. ⁽¹⁾ A proper follow up system is needed by every hospital treating DS patients to provide them with UpToDate standard of care management and follow up. In our quality project, we scoped the current practice and then the attitude of the pediatrics doctors treating DS patients in Ash Sharqiyah South Medical Governate four hospitals. Findings supported that the current practice needs to be optimized. Currently, there is no specific DS clinic in all of these 4 hospitals, and patient being engorged with other pediatric clinics. Our data showed that all of the doctors are involved in treating DS, even those who didn't receive any special training apart from pediatric basic background and most of the participants are medical officers (67%) working in Child Health Departments but not pediatric specialists. Data also revealed that there is a load of DS patients clinic visits, about (55%) stated that they are seeing 1-5 DS patients per month. Unfortunately, most of these OPD visits of DS patients are due to acute pediatric illness, and (71%) of the answers agreed with that DS patient end up in the clinic only when they are acutely sick, which is another

indication of deficiency in the current follow up system. Almost all agreed that there is currently no clinic available to deal with DS patient specifically. However, two doctors had different idea and they answered yes there is a clinic for DS patients, and possibly these two doctors confused between the "clinic" and the "service", possibly they mean the service is available and DS, patient are seen and given appointment. But in fact, there is no specific clinic for them, they are just being seen like any other pediatric patient without specific team and specific appointment plans following specific guide lines. Surprisingly, there is recognizable percentage of the doctors (17%) who are not aware about the MOH guideline booklet published in 2015 and distributed to all health institutes in the governorate. Likewise, there is (12%) of the doctors who don't follow the MOH guidelines at all, and other (24%) who are not following the Guideline most of the time, in investigator opinion, lack of awareness in the first point, and then lack of supervision might be the main reason of this unfavorable finding. Only (19%) who are following the MOH guideline strictly, this percentage should approach (100%) in such county with developed health system. In Investigator opening, regular evaluation of the current service might help to reach that aim. Doctors in Ash Sharqiyah South agreed that Down Syndrome Clinic is needed to make the follow up system easy and more organized, they agreed on following the MOH DS booklet and newborn card schedule and time tables in term of referrals and follow up OPD appointments and multidisciplinary team involvements. All agreed that the clinic should be operated by a pediatrician who received training at a central level for example in national genetic center in Royal Hospital and has been trained how to deal with the different complications associated with DS patient and what are the different laboratory and genetic tests to send and when to refer to other teams. Most of the Doctors agreed also that the clinic need to be in the center of the governorate, in Sur polyclinic, and that's because of some factors available in Sur City, firstly, it contains the largest number of populations, secondly, it has the secondary level hospital Sur Hospital with some, but not all, of the teams required for treating DS patient like; ophthalmology, adult cardiology (There is no pediatric cardiologist in the time when this thesis written) and ENT team. However, other important teams like pediatric endocrinology and pediatric haemato-oncology and developmental team all are lacked in Sur Hospital. Challenges which might face such clinic,

and which might obstruct the way toward establishing the clinic are; shortage of the staff, both doctors and nurses, lack of doctors with special training for DS cases, lack of awareness with the DS families to insure that they will follow the provided follow up plans once established, the other challenges is that Masira island patients are somehow separated geographically from Sur and evacuating a patient from an island needs special arrangements with all costs associated with that.

Conclusion

Down syndrome has a major health burden on the patient, family, and health care system. In Ash Sharqiyah South, there is no specific DS clinic, and the current practice is suboptimal and needs to be enhanced, our study showed that doctors in Ash Sharqiyah South are aware about the importance of a mature well organized follow up system for DS, and agreed that this follows up system will need specific well-structured regular clinic in the center of the governorate, Sur Hospital. the clinic to be operated by a trained pediatricians with trained nursing team following the national and international UpToDate guidelines, providing heights standard of care level for DS patients. There are several societies and communities for DS patients and their family in Oman like; Oman Down Syndrome Association, and Early Intervention Association, these associations providing much of social support. We still need to optimize the medical support and we can do that by many ways, establishing a specialized clinic for Down Syndrome patients is one of them.

Recommendations

- Establishment of a new well-structured clinic, used specifically for DS patient, supported by organized UpToDate follow up system connected with proper multidisciplinary team.
- The clinic to be operated by a team of pediatrics doctors and nurses who received proper practical training on how to treat DS patients.
- Enhance following the Guidelines of DS provided ministry of health and international protocols.
- Sur polyclinic is the suggested place to start the clinic.
- Community health education and advocacy especially for DS is needed to emphasizing the importance of the follow up.
- Ongoing training in term of Workshops and lectures for the doctors and health care workers who take care of DS patients.

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References

1. Kazemi, Mohammad, et al. "Down Syndrome: Current Status, Challenges and Future Perspectives." *Indian Journal of Community Medicine*, vol. 5, no. 3,

- 10 Oct. 2016, 10.22088/acadpub.BUMS.5.3.125. Accessed 13 Feb. 2020.
2. Asim, Ambreen, et al. "'Down Syndrome: An Insight of the Disease.'" *Journal of Biomedical Science*, vol. 22, no. 1, 11 June 2015, 10.1186/s12929-015-0138-y. Accessed 13 Feb. 2020.
3. Sam, Wajuihian. "Down Syndrome: An Overview." *African Vision and Eye Health*, vol. 75, no. 1.346, 6 June 2016, 10.4102/aveh. v75i1.346. Accessed 11 Feb. 2020.
4. Maino D. Overview of special populations. In: Scheiman M, Rouse M, editors. *Optometric management of learning-related vision problems*, 1st ed. St. Louis, MO: Mosby, 2006; p. 85–106.
5. Baum RA, Nash PL, Jessica EA, Foster JE, Spader M. Primary care of children and adolescents with Down syndrome: An update. *Curr Probl Pediatr Adolesc Health Care*. 2008; 38:241–261.
6. Tracy J. Australians with Down Syndrome-Health matters. *Aust Fam Physician*. 2011; 40:202–208.
7. Ministry of Health Oman. *Guideline for Medical Management of Children and Young People with Down Syndrome*. Ministry of Health Oman, 2015
8. Genetics Home Reference. "Down Syndrome." *Genetics Home Reference*, 2004, ghr.nlm.nih.gov/condition/down-syndrome#inheritance. Accessed 15 Feb. 2020.
9. Al Harasi, Salma. *Down Syndrome in Oman: Etiology, Prevalence and Potential Risk Factors. A Cytogenetic, Molecular Genetic and Epidemiological Study*. 16 June 2010.