



**Guideline for Medical Management of Children  
And Young People with Down syndrome**

**First Edition**

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**Sultanate of Oman**

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## Table of contents:

Title		Page No
Abbreviations		4
Preface		6
Section one:	Policy Guidelines	8
Section two:	Understanding Down Syndrome	11
	2.1. Definition and Diagnosis	12
	2.2. Types	12
	2.3. Clinical features	13
	2.4. Complications associated with Down Syndrome	14
Section three:	Tasks of Down syndrome health care	19
Section four:	Clinical Screening guidelines	25
	4.1. Clinical guidelines at birth	26
	4.2. Clinical guidelines up to 5 years of age	30
	4.3. Clinical Guidelines from 6 to 12 years	35
	4.4. Clinical guidelines from 13 to 18 years of age	39
Annexures		42
Annex 1	Overview of the National Down Syndrome follow up pathway.	43
Annex 2	Screening for congenital heart disease in Downs Syndrome	44
Annex 3	Down syndrome Neonatal Discharge Form	45
Annex 4	Counseling sample using the 5A construct	46
Annex 5	Follow up table	48
Annex 6	Information of existing services and links	49
References		50

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## Abbreviations:

AR	Aortic Regurgitation
AVSD	Atrioventricular Septal Defect
CBC	Complete Blood Count
CH	Congenital Hypothyroidism
CHD	Congenital Heart Disease
CHR	Child Health Record
DGHA	Directorate General of Health Affairs
DS	Down Syndrome
DSNDF	Down Syndrome Neonatal Discharge Form
IQ	Intelligence Quotient
IMCI	Integrated Management of Childhood Illnesses
GOR	Gastro Oesophageal Reflux
MOH	Ministry of Health
MR2	Medical Record
MVP	Mitral Valve Prolapse
MVR	Mitral Valve Regurgitation

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OME	Otitis Media with Effusion
PVD	Pulmonary Vascular Disease
RBS	Random Blood Sugar
TBG	Thyroxin Binding Globulin
TFT	Thyroid Function Test

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*Preface:*

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Down syndrome is one of the commonest chromosomal abnormalities known today. Internationally, the birth prevalence is estimated to be 1 in 600 to 1 in 800. In Oman, it is 2.4 in 1000 which leads to about 120 affected births / year. (MOH, [2011]). Individuals born with Down syndrome are at risk for a number of associated medical conditions such as congenital heart disease, impaired hearing, gastrointestinal tract anomalies, thyroid disorders and cataracts. Health care providers need to be aware of these conditions as some warrant immediate intervention and medical treatment.

The objectives of these guidelines are:

1. Provision of a care pathway for individuals affected with Down syndrome and their families.
2. Description of the appropriate management for children with Down syndrome within the existing health care set up in Oman.

Hence, in order to build and strengthen the capacity of providers in giving standardized comprehensive health care to children and young

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people with Down syndrome the Department of Family and Community Health of Ministry of Health – Oman developed practical clinical guidelines at different service levels. The guidelines are divided into four sections, these are:

1. Section one: Policy Guidelines
2. Section two: Understanding Down Syndrome
3. Section three: Tasks of Down syndrome health care
4. Section four: Clinical Screening guidelines

These guidelines are not meant to replace text books. Hence, whenever detailed information is required, textbooks and other latest references should be used for gaining in-depth knowledge and understanding of the subject. The guidelines are based on the work done by the Down Syndrome Medical Interest Group (UK and Ireland). Where doubt was there in regards to applicability to local setting, evidence was drawn from other resources and consensuses of local experts.



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**Section One**  
**Policy Guidelines**

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## **Section One: Policy Guidelines**

Down syndrome is one of the major causes of intellectual disability and development delay. Therefore, the Ministry of Health is initializing a set of standardized services for children born with Down syndrome with the following goals:

- To optimize health services provided to Down syndrome patients
- To reduce mortality and morbidity associated with Down syndrome

Hence, services for evaluation and management of Down syndrome patients will be provided as an essential and integral component of maternal and child health services through the Ministry of Health network of health centers and hospitals of all levels in all regions.

### **The service will target**

1. All children and young people with Down syndrome

### **These services will include**

1. Clinical services: clinical evaluation, investigation and management.  
The provision of these services will vary from the primary to referral health care level
2. Counseling: will be done at all levels by the providers trained in counseling skills
3. Health education: through sessions and provision of educational material

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## **Who will provide these services**

All health personnel who are trained on the management of children and young people with Down syndrome

## **Services at different levels**

- **At Primary Health Care Level**

The staff at this health care level will conduct initial evaluation, screening and preventive measures as well as first line management and referral of cases to secondary health care level whenever needed.

- **At Secondary and Tertiary Health Care level**

The staff will further evaluate and manage referred cases and might collaborate with other specialties in the facility if needed.

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## **Section Two**

### **Understanding Down syndrome**

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## **Definition:**

Down syndrome is a genetic condition caused by the presence of all or part of a third copy of chromosome 21 which is why it is also known as trisomy 21. It is one of the major causes of intellectual disability and development delay. It is typically associated with physical growth delays, characteristic facial features and some level of intellectual disability but because no two people are alike these features will vary from one person to another.

## **2.1 Diagnosis:**

- Clinical features (table 1).
- Chromosomal analysis by routine karyotyping.

## **2.2 Types of Down syndrome:**

**Full trisomy 21:** this is the most common type, as it is detected in about 95% of individuals with Down syndrome. It indicates that all the cells have an extra chromosome 21. The most common error is maternal nondisjunction in the first meiotic division, with meiosis I errors occurring 3 times as frequently as meiosis II errors. The remaining causes are paternal in origin, and meiosis II errors predominate.

**Mosaicism of trisomy 21:** affecting 1-2% of individuals with Down syndrome, by which individuals have 2 cell lines; one is normal and one is trisomy 21.

**Translocation:** affecting about 3-4% of individuals with Down syndrome. The extra genetic material is the result of an unbalanced translocation between

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chromosome 21 and another chromosome, especially (14; 21) of which around 50% are familial.

\*The incidence of trisomy 21 conceptions increases with maternal age.

### 2.3 Clinical features of Down syndrome:

It is important for health care providers to familiarize themselves with the clinical features and complications of the syndrome. Making an early diagnosis can reduce mortality and morbidity.

#### Clinical features of Down Syndrome (AAP Guidelines, 2001)

- Hypotonia.
- Brachycephalic.
- Epicanthic folds.
- Flat nasal bridge.
- Upward slanting palpebral fissures.
- Brush field spots.
- Small mouth
- Small ears
- Excessive skin at the nape of the neck
- Single transverse



Figure.1: Frontal appearance of Omani Down Syndrome child with

<p>palmer crease</p> <ul style="list-style-type: none"> <li>• Short fifth finger with clinodactyly</li> <li>• A wide space (sandal gap) between the first and second toe.</li> <li>• Mental impairment which many be mild (IQ 50-70), moderate (IQ 35-50) and severe (IQ 25-35).</li> <li>• Developmental delay</li> </ul>	<p>Upward slanting palpebral fissures, epicanthic fold, flat nasal bridge and short neck</p>
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## 2.4 Complications associated with Down syndrome:

Children with Down syndrome are at risk of having certain congenital anomalies and associated conditions. Hence it is important for health care providers to be familiar with these conditions to ensure proper screening and early diagnosis.

### 1. Cardiac anomalies:

- There is 40-60% chance for heart defects to occur in individuals with Down syndrome. Of these 30-40% are complete Atrio Ventricular Septal Defects (AVSD). Most AVSD can be successfully treated if detected and operated early before pulmonary vascular disease (PVD) which is more likely to

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develop in child with Down syndrome and AVSD. Ideally surgery should be done by 6 months and there is some evidence indicating that surgery before 4 months provides optimum results.

- Doctors should also keep in mind high index of suspicion of other congenital heart diseases for all new born with Down syndrome. However other congenital heart defects can be approached with less surgical cardiac urgency (DSMIG,2007)
- There is a risk of developing asymptomatic mitral valve prolapse (MVP), and aortic regurgitation from adolescence period. The MVP may progress into mitral valve regurgitation (MVR).

## 2. Hearing loss:

- Hearing impairment can present in more than half of the children with Down syndrome. Main cause to hearing impairment is fluid accumulation leading to otitis media with effusion (OME) causing conductive loss of hearing.
- Children with Down syndrome may also have narrow ear canal which predisposes to accumulation of wax.
- Children born with Down syndrome also have a higher incidence of sensorineural deafness.
- As children grow, obstructive sleep apnea becomes more common than the general population. ENT surgery to address tonsils and adenoids is effective in at least third of the patients, and should be sought for those most likely to benefit (AAP, feedback 2012).
- In order to identify and prevent secondary disability from hearing loss in Down syndrome, it is important to focus prevention efforts on screening for hearing through physical examination of the ears using an otoscope and audiological assessment. Full audiological assessment should include:



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- inspection, tympanometry, hearing testing and speech testing if at appropriate developmental level.
  - Management of these problems should be started in the early infancy. If undetected, hearing impairment is likely to be a significant cause of a preventable secondary handicap.

### **3. Vision:**

- Approximately 60% of individuals with Down syndrome have visual disorders. These include refractive errors which can start from an early age.
- Special attention must be paid to screening for congenital cataract as they are ten times higher among them than in the general population.
- Cataracts and keratoconus may develop in teenage years or later.
- Screening and management of these problems needs to be started very early in infancy to prevent secondary handicap.

### **4. Thyroid:**

- Clinical forms of hypothyroidism found in individuals with Down syndrome include transient and primary-hypothyroidism, pituitary hypothalamic hypothyroidism, thyroxin-binding globulin (TBG) deficiency and chronic lymphocytic thyroiditis. Hyperthyroidism also occurs occasionally. The frequency of thyroid disease is elevated in patients with Down syndrome, starting in the newborn population where it is 0.7% (or 28 times more frequent than in the general population). Twelve percent or more of adults with Down syndrome have thyroid disease. Thyroid disease is

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difficult to diagnose clinically in individuals with Down syndrome because of an overlap of symptoms. This makes thyroid blood screening a particularly

important part of the screening of each person with Down syndrome. Risk is also higher at increased age.

- Newborn screening at birth is practiced in Oman for all children. However screening by obtaining venous sampling for TFT needs to be repeated in children with Down syndrome.
- Doctors should also maintain a high index of suspicion if a child presented with symptoms suggestive of hypothyroidism.

#### **4. 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).

#### **5. 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
- Doctors should maintain a high index of suspicion.

#### **6. Cervical spine disorders:**

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- 10-20% of individuals with Down syndrome may develop cervical spine disorders radiologically and around one fifth of those with the radiological findings may have symptoms (e.g. atlantoaxial subluxation).
  - Not enough evidence was found to support cervical spine X ray in childhood as a screening tool for predicting cervical spine disorders in future.
  - Doctors should keep a high index of suspicion if symptoms are suggestive and request X ray on clinical grounds.
  - Routine annual checks for symptoms of cervical spine problems should be carried out. Check for pain behind ear, neck pain, numbness or tingling in the lower limbs, or new onset bladder control problems.
  - Advice for transport of children in cars should be routinely given to parents and advice to professionals in case the child requires anaesthesia.

## **7. Hip dislocation:**

- 6% of Down syndrome children are at risk of acquired hip dislocation.

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## **Section Three**

### **Tasks of Down syndrome Health Care**

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### **Section Three: Tasks of Down syndrome Health Care**

In order to achieve above mentioned goals the following tasks must be carried out:

- **At primary health care level**

The staff at primary health care level will perform the following tasks:

**At diagnosis:**

Parents should be counseled by an appropriately trained person in their local health care system.

Chromosome test to confirm diagnosis to be marked URGENT in the system.

If the diagnosis is suspected, the child should be referred to the genetic clinic for consultation about risks in future pregnancies.

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**Follow up care:**

- Assess the child's growth and development as per the protocol.
- Screen for any other medical conditions.
- Carry out standardized lab and radiological investigations.
- Treat any medical conditions if possible and refer as necessary.
- Plan a follow up care pathway for the child.
- Counsel the parents and guide them to available services.
- Refer parents to health educator and community support person
- If child is going to school, manage a follow up plan with the school health nurse.

**Components of follow up visit:**

A schedule of 10 structured visits has been developed for follow up of children up till 18 years of age. Refer to (Annex 2, routine visits schedules and tasks) for the schedule including tasks to be performed at each visit.

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Each visit will carry multiple tasks and screening for a number of conditions

focusing on the most common ones:

Cardiovascular system: AVSD & other cardiac anomalies.

Ears: otitis media, sleep apnea and hearing loss.

Eye: cataracts and refractive errors.

Thyroid: hypothyroidism.

Skeletal: hip dislocation & cervical spine disorders, possibility of arthritis.

Growth & development: obesity and developmental skills.

Sleep issue: sleep apnea, and other breathing disorders.

This will be in addition to managing medical conditions and counseling.

Therefore longer time slots should be allocated, probably (30 -40) minutes as

most of the time families have a lot of questions and concerns, particularly in

the first few months after the diagnosis. Parents should also be encouraged

to take appointments.

**Documentation of care**

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Information should be documented in the child health record

▪ **At secondary health care level**

The pediatrician at secondary health care level should perform the following:

**At diagnosis:**

**Counselling for parents**

- Urgent Confirmation of the diagnosis by Karyotype.
- Notify the case using the congenital anomalies notification form to parent institution.
- Screen for any other medical conditions.
- Carry out standardized lab and radiological investigations.
- Counsel the parents and guide them to available services.
- Plan a follow up care pathway for the child.

**Follow up care:**

- Assess the child's growth and development.
- Screen as per the protocol.
- Treat any medical conditions if possible and refer as necessary.
- Refer parents to health educator and community support person.

**Components of follow up visit:**

Refer to (Annex 2, routine visits schedules and tasks) for the schedule including tasks to be performed at each visit.



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Therefore longer time slots should be allocated, probably (30-40) minutes as most of the time families have a lot of questions and concerns, particularly in the first few months after the diagnosis. Parents should also encouraged to take appointments.

### **Documentation of care**

Information should be documented in the child health record. A Down syndrome neonatal Diagnosis Form (DSNDF) should be issued from the birthing facility and sent to the parent's institution along with the congenital anomaly form. A copy

should be kept with the family. This standardized form with the agreed set of parameters will aid health care providers in providing and following up recommended care to children.

- **Tertiary health care level**

The pediatrician at secondary health care level should perform the following:

#### **At diagnosis:**

- Confirm the diagnosis by Karyotype.
- Notify the case using the congenital anomalies notification form to parent institution.
- Screen for any other medical conditions
- Carry out standardized lab and radiological investigations.
- Counsel the parents and guide them to available services.
- Plan a follow up care pathway for the child.

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**Follow up care:**

- Assess the child's growth and development.
- Screen as per the protocol.
- Treat any medical conditions if possible and refer as necessary.
- Refer parents to health educator and community support person

**Components of follow up visit:**

Refer to (Annex 2, routine visits schedules and tasks) for the schedule including tasks to be performed at each visit.

Therefore longer time slots should be allocated, probably (30 -40) minutes as most of the time families have a lot of questions and concerns, particularly in the first few months after the diagnosis. Parents should also be encouraged to take appointments.

**Documentation of care**

Information should be documented in the child health record. A Down syndrome neonatal Discharge Form (DSNDF) should be issued from the birthing facility and sent to the parent's institution along with the congenital anomaly form. A copy should be kept with the family. This standardized form with the agreed set of parameters will aid health care providers in providing and following up recommended care to children.

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**Families should be kept informed about the finding of clinical evaluation, investigation and management planned**

## **Section Four**

# **Clinical Screening Guidelines**

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## SECTION THREE: CLINICAL SCREENING GUIDELINES.

### 4.1. Clinical guidelines at birth (implemented at the delivery hospital)

#### 1. Diagnosis:

- Diagnosis should be suspected on **clinical findings** (features of Down syndrome in table 1). Parents should be informed of the suspected diagnosis as soon as possible and have a follow up meeting with their health care professional within 48 hours.
- Chromosomal analysis should be requested **urgently**.
- Disclosure of the diagnosis **should not be delayed** to wait for the results, and preferably done by a senior pediatrician.

#### 2. Vaccinations:

- Check vaccination status of the child. All vaccinations should follow standard MoH schedule in addition to vaccinations available to high risk groups.<sup>2</sup>

#### 3. Routine clinical examination:

- Assess child's weight, height and head circumference and plot them on the growth charts.
- Assess the vital signs including 4 limbs Bp assessment.
- A full routine clinical examination of the child with particular attention to common complications such as cardiac anomalies, bowel atresia, Hirschsprungs, and cataracts as detailed below.

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**a. Congenital heart disease:**

**Clinical examination:**

- Look for cyanosis at birth.
- Listen for a murmur.<sup>3</sup>

**Investigations:**

- ECG.
- Chest X-ray.
- Echocardiogram if available.
- Pre / postductal O<sub>2</sub>.

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<sup>2</sup> Currently Influenza vaccine is available for children 6 to 35 months: 0.25 ml (two doses one month apart).

<sup>3</sup> AVSD usually presents with no murmur at birth

**Management:**

- Those with abnormal clinical signs or ECG abnormality (in particular a superior QRS axis) are potentially at high risk for pulmonary vascular disease and should be referred urgently to a pediatric cardiologist for expert clinical assessment and echocardiogram.
- Treat increased pulmonary vascular resistance at birth if baby is cyanosed.
- Treat if baby is in failure with diuretics, captopril and added calories.
- Those with no abnormal clinical signs or ECG abnormality on initial examination give them an appointment for an expert appointment before 6 week, for further clinical assessment and echocardiogram. (**Annex 3: cardiac Guidelines**).

**b. Ears:**

- Check results of the echo hearing screening test.
- If the test is not done or results are inconclusive or abnormal give the child an appointment before discharge.
- If results were abnormal refer the child to ENT for full audiological assessment.

**C.Vision :**

- Congenital cataracts: look for cloudiness of the lens (like a white spot in an otherwise dark pupil).
- Look for nystagmus: rapid eye movements either vertical or horizontal.

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- If an abnormality is noted refer to ophthalmologist.

**d. Congenital Hypothyroidism :** review results of TFT, if results are suggestive of CH, start the child with an initial dose 10-15mg/kg. Adjust the thyroxin dose according to the thyroid function test every 2-4 weeks initially, then as required. Refer to congenital hypothyroidism guidelines 2010 for further management.

**e. Congenital hip dysplasia:** Hips can also be assessed by looking at the symmetry of the height of the knees when the infant is supine. A positive Galeazzi sign (unequal knee heights) suggests a unilaterally dislocated hip. It is important to remember that bilateral dislocations will likely appear symmetric, so this assessment should only be used in combination with the Ortolani and Barlow maneuvers.

**f. Gastrointestinal malformations:** (check for Hirschsprung's disease, intestinal obstruction and imperforate anus).

- Ask mother / nurse about passage of stools.
- Ask about vomiting
- Look for abdominal distention.
- Perform a thorough abdominal examination including looking for imperforated anus.

#### **4. Laboratory investigations:**

- Completed blood count (higher risk of hematological abnormalities such as neutrophilia, thrombocytopenia and polycythemia, than in general population) (normal ranges)
- Chromosomal studies by routine karyotyping to confirm the diagnosis and identify the genetic form of Down syndrome.
- Parental chromosomal studies by routine karyotyping if the child is having a translocation abnormality.
- Children born with Down syndrome are more prone to congenital hypothyroidism (primary and secondary), hence its preferred to do both (FT4 & TSH) at day 7 of life. If it is difficult, then serum FT4 & TSH should be performed at the time of discharge.

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#### 4. Counseling:

A successful counseling session should always have defined goals and objectives, the following points should be included in your session. Please note that most parents may need more than few sessions of counseling.

- Family's preparedness for the diagnosis, did they know antenatally?
- Features, risks and potentials of child with Downs Syndrome.
- Test of confirming the diagnosis, recurrence risks.
- Extra tests to be done.
- F
  
- Feeding with emphasis on benefits of breast feeding a Down syndrome baby (improving mother –tongue coordination, hence may improve speech problems in the future. Protection from infections (Respiratory and Bowel). Bonding and parenting skills.
- Increased susceptibility to respiratory tract infections and ear infections.
- Constipation and possibility of an underlying medical condition.
- Care of the baby with special attention to hypotonia.
- Importance of promoting the inclusion of the child in the society.
- Review the family's support system, such as family friends, and extended help. Discuss efficacy of early intervention and availability of early intervention services and therapies in the community. Initiate referral as appropriate (Early Intervention Association and Down Syndrome Society).

When counseling the families try to ensure:

- Confidentiality and privacy
- Parents' readiness: you can set a time when both parents are available.
- Using a neutral tone of voice when counseling.
- Give the parents some time to accept the news.
- Use open ended questions and ask them if they would like to ask any more questions.
- Define main issues and concerns for the family and outline available options.
- Professional must be well informed.

#### 6. Documentation:

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- Fill the congenital anomalies form.
  - Fill details in the Child Health Record (CHR).
  - Fill in the necessary details in the DSNDP.
  - Send a copy of the discharge summary to the child parent's institution with a clear discharge plan.

## **4.2. Clinical guidelines Up to 5 years of age (At 2 months, 6 months, one year, 3 & 5 years) (Implemented at Pediatrics OPD)**

### **1-History:**

- This should include general health status of the child, current and past medical illnesses.
- Feeding and nutrition, history of GOR.
- Bowel habits.
- Play and engagement in social activity, behavioral problems
- Developmental milestones.
- Any medical concerns.
- Details of follow up at a higher level of care.
- Check vaccination status of the child. All vaccinations should follow standard MoH schedule in addition to vaccinations available to high risk groups.

### **2-Physical examination**

#### **Vital Signs:**

Take a record of the temperature, heart rate and respiratory rate.

#### **Growth parameters:**

Weight, height and head circumference.



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- Children with DOWN SYNDROME may not gain weight in the first month of life; this is due to feeding difficulties. However after the first month onwards, weight should increase parallel to centiles.
  - Those with measurements below the 2<sup>nd</sup> centile may have an underlying pathology or failure to thrive and hence must be evaluated further. Such children will need referral to a dietitian or a higher level of care.
  - If child is having problems with feeding then you need to refer him to a speech therapist. (Trained staff nurse with appropriate skills might be better resource, particularly in relation to feeding problems)

### **Systemic examination:**

Look at the general hygiene status of the child.

Examine for jaundice, pallor, lymph nodes, swellings, bruises, skin and hair condition and dental.

#### **a. The cardiovascular system:**

##### **History and clinical examination:**

- Ask about history of cyanosis, sweating and tachypnea.
- Look for signs of heart failure (may present by 1-2 months in children with AVSD).
- Auscultate the pericardium for a murmur.

##### **Cardiac Investigations:**

If not known to have any cardiac anomalies at 2 month visit:

- ECG: look for superior QRS complex and bi-ventricular hypertrophy.
- EXR: increased pulmonary vascular markings and cardiomegaly after 1 month.

##### **Management:**

- If investigations are normal and no clinical signs were found, a routine appointment to a pediatric cardiologist should be sent for further assessment.
- Those with abnormal clinical signs on examination or investigation, an urgent referral should be sent.
- Indicate in the referral if an echocardiogram was done previously and what were the findings.

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- Please note that children with heart lesions are at higher risk of infective endocarditis, therefore antibiotics prophylaxis prior to and after various procedures should be prescribed.

Older children who have never had an echocardiogram should be dealt with as follows:

- (a) Those with no symptoms or clinical signs and normal ECG should be referred routinely for further evaluation by pediatric cardiologist.
- (b) Those who are symptomatic and/or have abnormal clinical signs or ECG should be referred urgently.

#### **b. Ears:**

##### **History:**

- Check the results of the hearing assessment done at birth and make sure referrals are organized and child has received standard care.
- Ask about history of symptoms suggestive of otitis media (acute and chronic): Poor feeding, Pulling of the ear, irritability, restlessness at night, unresponsiveness to quiet sounds repeated upper respiratory tract infection.
- Ask about sleep and whether there is a disturbance. Refer to ENT if needed.

##### **Clinical Examination:**

Examine the ears using an otoscope and look for:

- Fluid leaking into the outer ear.
- Try and visualize the ear drum, it is normally pink in color. If there is Otitis media it may be red or yellow in color.
- Check if the ear drum is bulging.
- Check if the ear drum is perforated.
- Examine the throat and to look for lymph nodes

Send a request to ENT specialist (Secondary level) for a full audiological assessment including auditory threshold, impedance testing and autoscopy. This should be done between 6 to 10 months and repeated again in the second year (DSMIG, 2004).

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**Management:**

- If the child has AOM, treat according to IMCI guidelines.
- Children with hearing impairment should be referred to ENT for hearing support services.

**c. Vision:**

- Look for squint, cataract, and any redness in the eye.
- Check if the infant can follow objectives. A 6 weeks old child should be able to fix and follow an object held at arm's length through 90 degrees in the horizontal plane. A child at 12 weeks should be able to fix and follow an object 180 degrees in the horizontal and vertical planes.
- All children with Down syndrome should have a full check by ophthalmologist and orthoptist to check visual acuity before their second birthday.

**d. Thyroid:**

- Review previous results of newborn screening for congenital hypothyroidism/TFT, if results are suggestive of congenital hypothyroidism (CH), start the child with an initial dose of 4-5mg/kg daily. Adjust the thyroxin dose according to the thyroid function test every 2-4wks initially, then as required. Refer to congenital hypothyroidism guidelines 2010 for further management.
- If child is suspected to have congenital hypothyroidism<sup>4</sup>, (key pointers: history of lethargy, changes in affect, cognition, growth or weight or weight) repeat TFT by venous sampling
- Check weight gain of the child. Hypothyroidism should always be thought of if there is an accelerated weight gain. If child is suspected to have congenital hypothyroidism, repeat TFT by venous sampling.
- TFT should be done annually after the first year of life.

**e. Cervical spine disorders:**

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- Ask and look for change in gait or use of arms or hands, change in bowel or bladder function, neck pain, stiff neck, head tilt, torticollis, and how the child positions his or her head.
  - Children may also complain of pain behind the ears or elsewhere in the neck.
  - Refer to a specialist orthopedic surgeon urgently if above complaints were present.

Advise parents about the risk of playing certain sports... Discuss symptoms of sleep disordered breathing, including heavy breathing, snoring, restless sleep, frequent night awakening, daytime sleepiness, apneic pauses, and behavior problems.

### **3. Laboratory investigations:**

- Complete blood count (higher risk of hematological abnormalities such as neutrophilia, thrombocytopenia and polycythemia, than in general population) at 1 year and 5 years of age.
- TFT Routine screening for asymptomatic children should be carried annually accompany with thyroid antibodies .If antibodies positive then TFT should be done every 6 months.
- Fasting blood sugar (FBS) should be done once a year accompanied with glycohemoglobin (HbA1C) or as indicated. If FBS >5.6 mmol & HbA1C >6.2% then child should referred to endocrinologist for oral glucose tolerance test (OGTT).

### **4. Education:**

- Explain the importance of early intervention, including physical therapy, speech therapy and enrollment in a preschool program.

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<sup>4</sup> (Refer to newborn screening for congenital hypothyroidism guidelines 2010)

- Encourage the parents to establish an optimal dietary and physical activity pattern that will prevent obesity.
- Review injury and abuse prevention with special consideration of developmental skills.
- Provide advice about management of child's behavior and refer to a psychologist if needed.
- Refer the family to the community support person/nurse (if possible).
- Guide the family to available services for children.

### **4.3. Clinical guidelines from 6 years to 12 years: (At 7, 9 and 12 years)**

#### **(Implemented at Pediatrics OPD)**

##### **1. History:**

- This should include general health status of the child, current and past medical illnesses.
- Diet and physical activity.
- Developmental milestones and adaptive functioning.
- Pre and pubertal changes.
- Any medical concerns.
- Details of follow up at a higher level of care.
- Check vaccination status of the child. They should follow the standard MoH schedule

##### **2- Physical examination**

##### **Assessing the Body Mass Index (BMI):**

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Childs weight and height should be measured at each visit, and the BMI calculated  $(\text{weight [kg]} / (\text{height [m]})^2)$

Most children with Downs Syndrome are shorter than average. They are prone for obesity, particularly from late childhood

### **Systemic examination:**

Look at the general hygiene status of the child.

Examine for jaundice, pallor, abnormal lymph nodes, swellings, bruises, skin and hair condition and dental review.

Observe for unusual reactions to physical examination. Children with Down syndrome are at higher risk for child abuse.

Look for appearance of secondary sexual characteristics.

### **The cardiovascular system:**

#### **History and Clinical examination:**

- Ask about history of tiredness, difficulty of breathing and sweating.
- Look for signs of heart failure.
- Auscultate the pericardium.
- Carry out an ECG.
- 

#### **Management:**

- Older children who have **never** had an echocardiogram should be dealt with as follow:
  - (a) Those with no symptoms or clinical signs and normal ECG should be referred routinely for further evaluation by pediatric cardiologist.
  - (b) Those who are symptomatic and / or have abnormal clinical signs or ECG should be referred urgently.
- Please note that children with heart lesions are at higher risk of infective endocarditic, hence antibiotics prophylaxis prior to and after various procedures should be prescribed.

- 
- Ask about symptoms related to sleep disordered breathing: Snoring, restless sleep and sleep position. Refer any child with signs or symptoms of sleep disordered breathing to ENT
  - Monitor for neurologic dysfunction, including seizures

**b. Ears:**

**History:**

- Ask about supportive nasal and ear conditions.

**Clinical Examination:**

- Review hearing test result. Annual retest required to examine ears.
- Examine the ears using an otoscope and look for:
  - Fluid leaking into the outer ear.
  - Try and visualize the ear drum, it is normally pink in color. If there is otitis media it may be red or yellow in color.
  - Check if the ear drum is bulging.
- Check if the ear drum is perforated.
- Examine the throat and to look for abnormal lymph nodes.

**Management:**

- Review results of regular audiological evaluation.
- Request for a full audiological assessment including auditory threshold, impedance testing and autoscopy. This should be done between 9 to 10 years. Speech perception tests may also be useful.
- Children with hearing impairment should be referred to hearing support services. Remember use of hearing aids can be very successful.

**c. Vision:**

**History:**

- Ask if any abnormalities (squint, cataracts and nystagmus) have been noticed.
- Do a refraction error test, if any abnormality refer for a further follow up

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**Clinical examination:**

- Congenital cataract; look for cloudiness of the lens (like a white spot in an otherwise dark pupil)
- Look for nystagmus: rapid eye movements either vertical or horizontal.

**Management:**

- Refer to ophthalmologist if any abnormality found.

**d. Thyroid:**

- Review previous results of TFT.
- If the child is suspected to have congenital hypothyroidism<sup>5</sup>, (key pointers: history of lethargy, changes in affect, cognition, growth or weight) repeat TFT by venous sampling.
- Review results within 2 weeks, if you thought the child has CH, send blood for thyroid antibodies and start treatment with an initial dose of 3-4 mg/kg
- Daily for children aged 6-10 years. If the child is more than 11 years then start with a dose of 2-3 mg/kg/day.
- Repeat TFT every 2-4 weeks initially then as required and adjust thyroxin dose accordingly.

**f. Cervical spine disorders:**

- Look for abnormal head posture, torticollis, and abnormal gait, any new onset of bladder problems.
- Children may also complaint of pain behind the ears or elsewhere in the neck
- Refer to an orthopedic surgeon for further follow up, if any signs were noted, send an urgent referral.

**2. Laboratory investigations:**

- Complete blood count (higher risk of hematological abnormalities such as neutrophilia, thrombocytopenia and polycythemia, than in general population) at 9 year.
- TFT should be done annually. Fasting blood sugar (FBS) should be done once a year accompanied with glycohemoglobin (HbA1C) or as indicated. If FBS >5.6 mmol & HbA1C >6.2% then child should referred to endocrinologist for oral glucose tolerance test (OGTT).



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#### 4. Education:

- Review the child's development and available educational opportunities. Offer the parents referral of the child to a clinical psychologist for further developmental assessment
- Discuss behavioral problems and management of problematic behaviors, potential of poor sleep as an underlying cause.
- Consider parent skills training when available
- Offer support to careers.
- Discuss the development of age appropriate social skills, self-help skills and the sense of responsibility.
- Discuss psychosexual development, physical and sexual development, menstrual hygiene and management and issues related to puberty and fertility.
- Educate parents on abuse prevention.

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<sup>5</sup>(refer to newborn screening for congenital hypothyroidism guidelines 2010, for clinical features)

#### **4.4. Clinical guidelines from 13 years to 18 years (At 16 and 18 years) (Implemented at Pediatrics OPD)**

#### 3. History:

- This should include general health status of the child, current and past medical illnesses.
- Diet and physical activity.
- Development skills and adaptive functioning.
- Pre and pubertal changes.
- Any medical concerns.
- Details of follow up at a higher level of care.
- Check vaccination status of the child. All vaccinations should follow standard MOH schedule.

#### **Routine physical examination:**

#### **Assessing the Body Mass index (BMI):**

- 
- Child's weight and height should be measured at each visit, and the BMI calculated (weight [kg] / height [m x x]).
  - Most children with Down Syndrome are shorter than average. They are prone for obesity, particularly from late childhood.
  - Examine for secondary sexual characteristics and ask about Menarche. Individuals with Down syndrome follow similar age of onset and completion of puberty as normal individuals.
  - Check developmental milestones.
  - Assess for symptoms related to celiac disease and refer to gastroenterologist if needed
  - Ask for history of seizures.

### **1. Specific examination :**

#### **The cardiovascular system:**

##### **Clinical examination:**

Auscultate the pericardium for a murmur. There is a risk of developing asymptomatic mitral valve prolapse (MVP), and aortic regurgitation from adolescence period.

- Consider prophylaxis of infective endocarditis before dental procedures.
- Consider prophylaxis of infective endocarditis before dental procedures.
- MVP may progress into mitral valve regurgitation (MVR).

##### **Investigation:**

- ECG
- Chest x-ray

##### **Management:**

- Older children who have never had an echocardiogram should be dealt with as follows:
  - (a) Those with no symptoms or clinical signs and normal ECG should be referred routinely for further evaluation by pediatric cardiologist.
  - (b) Those who are symptomatic and / or have abnormal clinical signs or ECG should be referred urgently.

- 
- 
- (c) An echocardiogram should be obtained if there is a history of increasing fatigue, shortness of breath, or exertional dyspnea or abnormal physical examination findings, such as a new murmur or gallop.

**Vision:**

Check for onset of cataracts, refractive errors, and keratoconus, which can cause blurred vision, corneal thinning, or corneal haze and is typically diagnosed after puberty

**Ears:**

- Discuss symptoms related to sleep disturbed breathing, including snoring, restless sleep and sleep position. Refer to specialist if needed.

**2. Laboratory investigations:**

- CBC at 16 years.
- TFT should be done annually for asymptomatic children should be carried annually accompany with thyroid antibodies .If antibodies positive then TFT should be done every 6 months.
- Fasting blood sugar (FBS) should be done once a year accompanied with glycohemoglobin (HbA1C) or as indicated. If FBS >5.6 mmol & HbA1C >6.2% then child should referred to endocrinologist for oral glucose tolerance test (OGTT).

**3. Education:**

- Discuss psychosexual development, menstrual hygiene and issues related to puberty and fertility.
- Consider use of hormone preparations for control of menstrual cycle and foe excessive bleeding.
- Young males should be encouraged to have regular testicular checks because of the increased incidence of testicular tumors and poor compliance to self-examination.
- Discuss the option of hysterectomy with the family.
- Educate parents on abuse prevention.

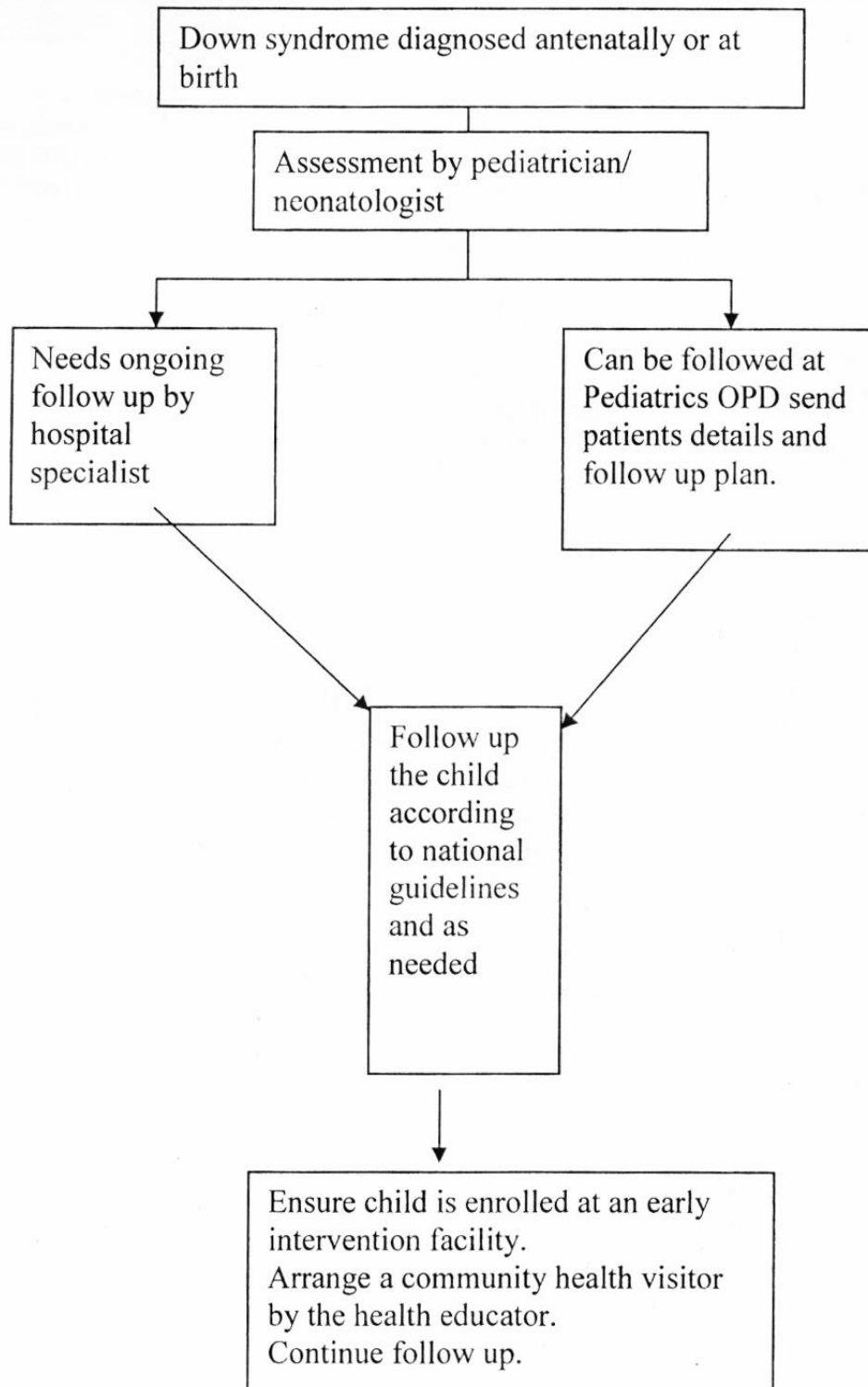
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- Discuss behavioral problems and management of problematic behaviors
  - Discuss opportunities for employment.
  - Counsel regarding healthy diet and a structured exercise program.

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# ANNEXURE

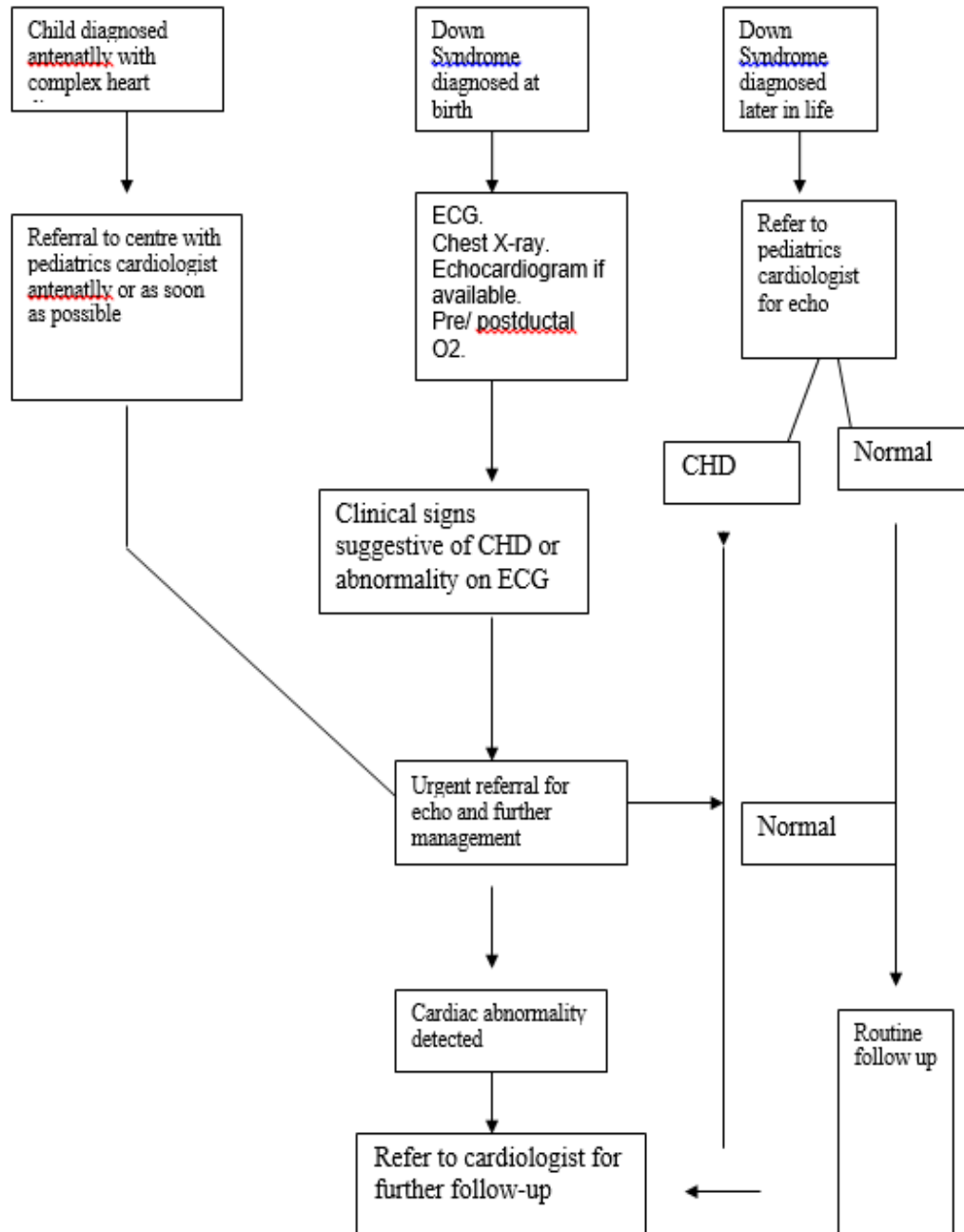
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## Annex 1: Overview of the National Down Syndrome follow-up:



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## Annex 2: Screening for congenital heart disease in Downs's Syndrome



**Annex 3: Down syndrome Neonatal Discharge Form:**

Childs name: Sex: Date of birth: Delivery hospital: Hospital number: (or place a sticker)	Childs parent institution: Region: Willayat: Name:			
Clinical findings:  APGAR score: Did the child require resuscitation:  Meconium- time passed:	Chromosomal test:	Thyroid screening: TSH: T3: T4:	Follow up plan:	
Sharing of news with parents	When done:	By whom:		
Cardiac evaluation:	Investigations done: <input type="checkbox"/> ECG <input type="checkbox"/> C Xray <input type="checkbox"/> Pre & post O2	Results:	Follow up plan:	
Echocardiogram:				
Hearing screening:	<input type="checkbox"/> Done <input type="checkbox"/> Not done	Results:	Follow up plan:	
Parents main concerns				
Follow up of the child:	<input type="checkbox"/> Delivery hospital <input type="checkbox"/> Secondary hospital <input type="checkbox"/> Primary health care	Notes:		
<b>Doctors name:</b>		<b>Signature:</b>		



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**Annex 4: Sample of Counseling for Down syndrome using the  
(5A CONSTRUCT FOR COUNSELLING)**

**ASSESS:**

- ❖ Assess details of karyotyping.
- ❖ Review results of other investigations (ECG, Chest X-ray, Echocardiogram, TFT, and CBC) to evaluate risks of other associated complications.

Assess the availability of personal support to the family.

**ADVICE:**

- ❖ Give results of karyotype.
- ❖ Care of the baby and feeding.
- ❖ Developmental Skills
- ❖ Educational and rehabilitative options.
- ❖ Support groups.
- ❖ Injury and abuse prevention.
- ❖ Birth spacing options and risks of having another child with DOWN SYNDROME.
- ❖ Explain the condition to the parents and assess their understanding and preparedness to deal with the child.

**AGREE:**

- ❖ Further management plan.

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ASSIST:

- ❖ Support groups contacts
- ❖ Available rehabilitation option

ARRANGE:

- ❖ Give appointment for net visit if needed.
- ❖ Give appointment for further investigations
- ❖ Clear discharge plan with parent's institution.

ASK THE PARENTS FOR ANY QUERIES AND REASSURE ACCORDINGLY.

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## Annex 5: Follow up table

	At birth	2/12	6/12	One yr	3 yrs	5 yrs	7 yrs	9 yrs	12 yrs	16 yrs	18 yrs
Health Assessment											
Chromosomal analysis	✓	✓ If not done									
Cardiac examination (according to protocol)	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓
Ears & Hearing	✓ echo hearing test				Complete audio logical assessment						
Ophthalmic evaluation by history	✓	✓	✓	Ophthalmological review and acuity check	✓	✓	✓	✓	✓	✓	✓
TSH	✓										
TFT + thyroid antibodies should done annually	At day 7 of life				✓	✓	✓	✓	✓	✓	✓
CBC	✓			✓		✓				✓	
FBS + HbA1C should done annually				✓	✓	✓	✓	✓	✓	✓	✓
Counseling and parental support	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓
Anthropometrics measurements	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓

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**ANNEX 6: Available Non-Governmental Organizations for children with Down Syndrome in Oman**

(1)Oman Down syndrome association, Muscat

Tel: 00968 95621200, 00968 99345686

E.m [http://www.omandsa.org.com/photo\\_06.html](http://www.omandsa.org.com/photo_06.html)

Email address:zhouenli@gmail.com

(2)Association of Early Intervention for children with disabilities –

Athaiba – Muscat

Tel: 24496960

Email address: earlyint@omantel.net.om

Fax: 24492118

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