Dear Health Care Professional:

Today, people with Down syndrome are achieving more than ever thought possible. People born with Down syndrome, who only twenty years ago may have been institutionalized, are now living independently and semi-independently, attaining paid employment, graduating from high school and attending post-secondary schools. They are musicians, actors, artists and athletes. Many people with Down syndrome have friendships and relationships and some marry. While it is important to avoid placing unrealistic expectations on children with Down syndrome, they should be challenged to do their best and follow their dreams—just like any child.

As health care professionals, we are in a unique position to challenge ourselves to recognize and overcome our own challenges and the medical community must also change. We must be prepared to see each child as an individual with unique strengths and challenges.

The world is changing for people with Down syndrome and the medical community must also change. We must challenge ourselves to recognize and overcome our stereotypes and to treat these children and their families with the highest level of care. We believe we are up to the task.

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PHYSICAL CHARACTERISTICS OF DOWN SYNDROME

While the only way to accurately diagnose Down syndrome is through a chromosomal karyotype, certain physical features may be present in these individuals:

- Muscle hypertonia
- Oblique palpebral fissures
- Flat facial profile
- Single crease across centre of palm
- Hypertelorism
- Dysplastic middle phalanx of 5th finger
- Hyperflexibility
- Excessive space between 1st and 2nd toes
- Dysplastic ear
- Enlarged tongue compared to mouth size

PRESENTING THE DIAGNOSIS

Keep the following suggestions in mind when giving a diagnosis, both prenatally and postnatally:

- If at all possible, give a prenatal diagnosis in person rather than over the telephone.
- Prenatally and postnatally, tell both parents together. Otherwise, the mother will have to tell the rest of the family by herself.
- A health care professional with a relationship to the mother should give the diagnosis.
- Postnatally, there may be clear indications of Down syndrome in the last stages of delivery. It is best to wait until the mother is in the recovery room to inform.
- Bring the baby into the room when discussing the physical indications of Down syndrome.
- Do not make a positive diagnosis until a karyotype has been done.
- Both pre- and postnatally, do not express an opinion about the parents’ situation. Ask what information they would find most helpful. Give resources: support groups, early intervention centres, literature.
- Make sure to discuss the diagnosis and field questions from the parents on more than one occasion. Parents may not be able to retain all the information in one sitting.
- Most importantly, don’t try to be predictive. It is impossible to foresee the future of any child. A broad picture should be painted which recognizes every child’s capacity to develop individually.

ABOUT DSRF

The DSRF is dedicated to improving the lives of people with Down Syndrome through research, education information dissemination, programs and services. Since 1995, we have assisted people with Down Syndrome, their families and the professionals they work with by developing educational and training programs, disseminating information and research results, collaborating with other resource centres and community service providers, and raising funds to support a broad spectrum of research projects.

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Neonatal (Birth–1 Month)

- Review parental concerns. Chromosomal karyotype; genetic counseling, if necessary.
- If vomiting or absence of stools, check for gastrointestinal tract blockage (duodenal web or atresia or Hirschsprung disease).
- Evaluation by a pediatric cardiologist including echocardiogram. Subacute bacterial endocarditis prophylaxis — SBE, in susceptible children with cardiac disease.
- Exams for plethora, thrombocytopenia.
- Review feeding history to ensure adequate caloric intake.
- Thyroid function test — check on results of province-reviewed baby screening for congenital hypothyroidism (at birth or 3 months).
- Ear, nose and throat exam (as needed), especially if suspicious of otitis media.
- Well-balanced, high-fibre diet.
- Monitor for obesity by plotting height for weight on the growth charts for typical children.
- Screen for celiac disease IgA antigliadin antibodies and total IgA (between 2 and 3 years).
- Question about obstructive sleep apnea; ear, nose and throat exam (ENT) (as needed).
- Dental exam (2 years; follow up exams every 6 months after). Twice-daily teeth brushing.
- Reinforce need for subacute bacterial endocarditis prophylaxis (SBE) for cardiac problems (as indicated).
- Well child care: immunizations; pneumococcal vaccine (2 years).
- Evaluation by a speech and language pathologist to maximize language development and oral communication.
- Review parental concerns; current level of functioning; monitor for behavior problems.
- Continue speech therapy and physical therapy (as needed).

Infant (1–12 Months)

- General neurological, neuromotor and musculoskeletal examination.
- TSH and T4 — Thyroid Function Test (at 6 & 12 months).
- Evaluation by a pediatric cardiologist including echocardiogram. Consider progressive pulmonary hypertension in patients with a VSD or atrioventricular septal defect who have new constitutional symptoms of heart failure.
- Subacute bacterial endocarditis prophylaxis — SBE (as indicated).
- Well child care — immunizations.
- Feeding consult, especially if constipated. Consider Hirschsprung disease.
- Subacute bacterial endocarditis prophylaxis — SBE (as indicated).
- Review parental concerns. Chromosomal karyotype; genetic counseling, if necessary.
- Referral to DSRE.
- Pediatric ophthalmological evaluation (by 6 months) for screening purposes.

Childhood (1–12 Years)

- TSH and T4 — Thyroid Function Test (annually).
- Echocardiogram by a pediatric cardiologist if not done previously.
- Behavioral Auditory Testing (every 6 months to 3 years, then annually).
- Lateral cervical spine x-rays (neutral view, flexion, extension) to rule out atlanto-axial immobility. Radiologist to measure atlanto-dens distance and neutral canal width (at 3-5 years, then as needed).
- General pediatric and neurological exam, including evaluation for signs of spinal cord compression: deep tendon reflexes, gait, Babinski sign.
- Use Down syndrome growth charts and head circumference charts, as well as growth charts for typically developing children.
- Eye examination (annually, or more often as indicated).
- Reinforce need for subacute bacterial endocarditis prophylaxis (SBE) in susceptible adolescents.
- Adolescent medicine consult for puberty/sexuality issues; health, abuse prevention and sexuality education.
- Pelvic exam (only if sexually active).
- Low calorie, high fibre diet. Regular exercise. Monitor for obesity.
- Clinical evaluation of functional abilities (consider accelerated aging); monitor loss of independent living skills.

Adolescence (12–18 Years)

- TSH and T4 — Thyroid Function Test (annually).
- Psychoeducational evaluations (every 2 years) as part of Individualized Educational Plan (IEP).
- Begin functional transition planning (age 16 years).
- Monitor independent functioning.
- Pap smear and pelvic exam (every 1-3 years after first intercourse). If not sexually active, single-finger bimanual examination with finger-directed colposcopy. If unable to perform, screen pelvic ultrasound (every 2-3 years). Breast exam (annually).
- Monitor for behavioral or emotional changes and/or mental health problems. Psychiatric Referral (as needed).
- Continue speech and language therapy, as indicated.

Adulthood (Additional Tests Required in Adulthood)

- TSH and T4 — Thyroid Function Test (annually).
- Auditory testing (every 2 years).
- Ophthalmologic examination, looking especially for keratoconus and cataracts (every 2 years).
- Reinforce the need for subacute bacterial endocarditis prophylaxis (SBE) in susceptible adults with cardiac disease.
- Baseline mammography at 40 years. Follow up every other year until 50 years, then annually.
- Clinical evaluation for sleep apnea.
- Low calorie, high fibre diet. Regular exercise. Monitor for obesity.
- Clinical evaluation of functional abilities (consider accelerated aging); monitor loss of independent living skills.
- Neurological referral for early symptoms of dementia (decline in function, memory loss, ataxia, seizures and incontinence of urine and/or stool).