

New/Suspected Down Syndrome Diagnosis Protocol**

- 1) ***Inform both parents together of your suspicion*** after mother has had sufficient time to recover from the delivery, but ideally within the first 24 hours. Discuss in a private setting, without interruption, and in the parents' preferred language. Coordinate the message between the obstetric and pediatric staff, if possible. Have the infant present and referred to by name.
- 2) ***Congratulate parents on the birth of their newborn***; do NOT say "I'm sorry" or "I have bad news". Give a balanced approach about what Down syndrome means for that child and for that family. Choose your words with sensitivity, using "People First" language that emphasizes the child, not the disability. Avoid terms such as "Down's baby", etc. Be aware of the realities and possibilities for healthy, productive lives for people with Down syndrome today. Limit discussions to those medical conditions the newborn is suspected of having or that may occur in the first year. Allow time for questions and make plans for follow up.
- 3) ***Order chromosome karyotype as STAT or with FISH for 24-48 hour preliminary results***. The preliminary results should be available prior to hospital discharge, with the final karyotype results available within 1-2 weeks. Coordinate with parents whether they wish to receive the final results by phone or in person.
- 4) ***Request a Genetics consult with a Geneticist or Genetic Counselor***. Dr. Ed Spence, Dr. Susan Sparks and Dr. Laurie Demmer are available to see newborns at CMC-main or Presbyterian-main. Genetic counselors Amy Blevins and Cam Brasington are available during business hours to meet (if baby at CMC-main) or talk with families by phone (all others) prior to or after the genetics consult. For those newborns delivered at other hospitals, CMC Genetics will work the newborn in for an outpatient appointment within 1-2 weeks of discharge. *For consult or appointment, call 704-381-6810.*
- 5) ***Give parents information on DSAGC's First Call program*** that includes up-to-date information on Down syndrome, local resources, and parent contacts (www.dsa-gc.org). Offer to have another parent who has a child with Down syndrome contact the new family, initiated by *faxing the DSAGC First Call contact form to 704-705-2438.*
- 6) Heart defects are seen in ~50% of individuals with Down syndrome. ***Order an echocardiogram***, to be read by a pediatric cardiologist, preferably prior to hospital discharge. ***Obtain a CBC*** to rule out transient myeloproliferative disorder. Check for duodenal atresia, cataracts, reflux, and feeding difficulty. Follow up on newborn screening results for hypothyroidism and hearing loss.

Resources:

Down Syndrome Association of Greater Charlotte
First Call Program
704-916-9871 or tleyton@dsa-gc.org
www.dsa-gc.org

CMC/Levine Children's Hospital
Clinical Genetics
704-381-6810
www.levinechildrenshospital.org

National Down Syndrome Congress
www.ndscenter.org

National Down Syndrome Society
www.ndss.org

Basic Guidelines for Using People First Language (From the National Down Syndrome Congress)

- Put people first, not their disability
A "person with a disability", not a "disabled person"
A "child with autism", not an "autistic child"
- Use emotionally neutral expressions
A person "with" cerebral palsy, not "afflicted with" cerebral palsy
An individual who had a stroke, not a stroke "victim"
A person "has" Down syndrome, not "suffers from" Down syndrome
- Emphasize abilities, not limitations
A person "uses a wheelchair", not "wheelchair-bound"
A child "receives special education services", not "in special ed"
- Adopt family-preferred language
A "cognitive disability" or "intellectual disability" is preferred over
"mentally retarded"
"Typically developing" or "typical" is preferred over "normal"
"Accessible" parking space or hotel room is preferred over "handicapped"
- Never use the "R-word" as a joke
"That's so retarded!", "He's a retard"

*****References:***

Bull, MJ, and the Committee on Genetics. (2011). Health supervision for children with Down syndrome, *Pediatrics*, 128(2), 393-406.

Sheets, KB, et al. (2011). Practice guidelines for communicating a prenatal or postnatal diagnosis of Down syndrome: recommendations of the National Society of Genetic Counselors, *Journal of Genetic Counseling*, 20, 432-441.

Skotko, BG, et al. (2009). Postnatal diagnosis of Down syndrome: synthesis of the evidence on how best to deliver the news, *Pediatrics*, 124(4), e751-e758.