



WHAT TESTS SHOULD BE DONE ONCE MY CHILD IS DIAGNOSED WITH TRISOMY 13?

By Christine Nelson MD, Pediatric Hospitalist

Embracing Life - One Moment at a Time™

LivingWithTrisomy13.org

IF THE DIAGNOSIS IS NOT KNOWN PRENATALLY you should expect what would amount to a survey of all systems, and within a day or two, depending on how sick the baby is at birth. (Some things go first, like cardiac evaluation, of course, and might influence the course of testing after that.)

- Chest x-ray
- Echocardiogram
- Renal (kidney) ultrasound—sometimes included in the abdominal ultrasound
- Abdominal ultrasound (the rest of the belly)
- Brain (head) ultrasound
- Chromosome testing
- Hearing testing—mandatory in nearly all states now, but a priority with suspected chromosome or other congenital problems
- State screening (mandatory in all states), usually within 48 hours of birth

CONSULTATIONS would be expected from any or all of the following, depending on what hospital the baby is in (community or referral hospital) and what problems are found in the first few days or weeks. Some could potentially be done by phone, and not necessarily in the first few days, but certainly arranged by the time of discharge, if a baby is stable and expected to be discharged:

- Neonatologist (within a day)
- Pediatric cardiology (sooner)
- Geneticist (if available nearby or on hospital staff; later if in less served area)
- Plastic surgeon if there is a cleft and/or extra digits
- Feeding specialist (this would be an occupational therapist, physical therapist or speech pathologist with special expertise in feeding special needs kids)
- Social services specialist for support, community liaison, advocacy, community referrals
- Pediatric ophthalmologist
- Pediatric gastroenterologist (but often not needed for 1-3 months, since many MDs know well how to treat reflux medically)
- Pediatric neurologist
- Pastoral support if parents wish (they should be asked specifically—virtually all hospitals have clergy available, some more trained and adept in hospital pastoral care than others)

REFERRALS TO BE EXPECTED BY THE TIME OF DISCHARGE:

- Craniofacial (cleft palate) team consultation should be arranged by time of discharge (usually includes a pediatrician, plastic surgeon, ENT, pediatric dentist, orthodontist, specialized nurse, social worker)
- Community support services for developmental therapy, occupational therapy, physical therapy
- Sometimes arrangements for respite care or even hospice care
- A pediatrician (a “medical home”) who understands Trisomy 13, special needs kids, and community resources, and with whom parents feel comfortable

IF DIAGNOSIS KNOWN PRENATALLY:

- Hopefully there would have been discussions with the neonatal team **AHEAD OF TIME**, including a face-to-face meeting, to discuss parents’ wishes and hospital/unit policies and procedures
- An echocardiogram should be expected, **REGARDLESS** of whether there was one ahead of time. This is especially true if parents have decided a DNR status for their infant and he or she is breathing and does not appear to need vigorous resuscitation
- Parents should know that they can request specific things regarding resuscitation, at least in most places; they have to be specific though. For example, “I would like my baby to have oxygen and even bag and mask resuscitation, but if it looks as if a respirator is needed, I would like you not to put in a tube and put the baby on a ventilator.” Or, “NO intubation, no heart medicines, no chest compressions.” And so on.
- I would expect that any and all of the previously listed testing would and should be done unless an infant clearly has features of T-13 **AND** is expected to die within a day or so. Prenatal testing (chromosome testing, that is) is usually accurate, but not perfect.
- If parents express a desire for no resuscitation and their baby clearly has T-13 features, they should expect that their wishes be honored in terms of private time, comfort care and hospice.

IF YOU ARE HAVING TROUBLE GETTING THE NEEDED TESTING YOU MAY CONSIDER THESE OBSTICALS

- Arranging or withholding referrals, assessments and services has little to do with insurance or resources, and more to do with attitudes of the pediatric and referral team—
- Although there may be hassles in actually getting the care arranged with some insurances (HMOs, and Medicaid and other public/government can take more time, although, getting a good case manager and staying on top of the referrals can help many of these to move through quickly.) Most if not all, MDs are going to do what they can to get services for their patients

if they are committed to doing the best for the kids that can be done—even if it means a lot more work.

- Attitudes of the various caregivers are the toughest to crack—whether they are based on lack of knowledge, financial issues, past experiences, both personal and professional, social and cultural background and beliefs, or just on a misguided paternalistic approach. And when these clash with the values and wishes of parents, or with each other, all heck breaks loose and parents get confused, hurt and angry—and rightfully so.
- Hopefully you have found this site before your child's birth, or right after, and you feel empowered. By getting facts, making decisions, and making your wishes known in a direct, positive way will enhance communication with your healthcare team. This way, parents are more likely to get an alliance, rather than an adversarial situation, especially if your wishes are in conflict with the beliefs and recommendations of the healthcare team or some of its members.