

## Double Dominancy Pregnancy FAQs

### **Genetic outcomes when two parents have Achondroplasia:**

- Two individuals with achondroplasia have the below chances of conceiving:
  - 50% chance of a dwarf, heterozygous child (one parent passes the gene)
  - 25% chance of a child of average height (neither parent passes the gene)
  - 25% chance of a dwarf, homozygous child (both parents pass the gene) - fatal

### **Testing and Diagnosis:**

- **Amnio or CVS test:**
  - If the patient is going to an OB/GYN provider for the first time, or to a new provider who is not familiar with dwarfism/achondroplasia, it may be helpful to connect the provider with a MAB (Medical Advisory Board) specialist if they have questions. Also, if they are finding it difficult to perform a cervical exam, sometimes just explaining the angle of the pelvic tilt is helpful.
    - Members of the MAB describe a good way to discover the cervix is by having the provider squat or kneel almost underneath the exam table, and then look up in a 45 degree angle. Basically, to perform the exam more from underneath the patient instead of hovering over the patient.
  - Due to the early timing needed of these procedures, which are usually performed by a high risk OB specialist, it is really important to get into your OB team as early as possible after finding out you're pregnant. Many standard risk pregnancies do not get seen for the first time until closer to 10-12 weeks of pregnancy to confirm pregnancy, you would easily miss the CVS window if you wait to be seen for the first time at 10-12 weeks.
  - CVS procedure is usually done between 10 and 12 weeks of pregnancy
    - Is either done through the cervix (if have a posterior placenta) or through the abdomen (anterior placenta). Many pregnant people with achondroplasia have a cervix that is difficult to access due to the pelvic tilt and its usual more anterior location in comparison to AH pregnant people. This can make a CVS difficult to be performed via the cervix route; the providers could find it too difficult and not proceed via cervix.
  - Amniocentesis is usually done after week 16 of pregnancy
    - Done through the abdomen
  - Both of these specific time frames (for CVS and Amnio) are due to the risks to the fetus if done earlier
- **Growth monitoring:**
  - Starting at weeks between 18 - 20, if the long bones are shorter, and the head slightly larger, this is an indication that the baby may have both genes. With typical achondroplasia, the shortened bones usually aren't discovered until 26-28 weeks.
  - Growth measurements are not conclusive but have proven to be indicators of homozygous (DD) and heterozygous (one gene), depending on when the decline in growth is visible.

### What is exactly happening?

- The FGFR3 gene mutation for achondroplasia dwarfism is essentially “always on”, which slows down the process of turning cartilage into bone (i.e. like the overwatering of a plant)
- Achondroplasia dwarfism is a dominant genetic mutation: meaning only one copy of the mutated (changed) gene is needed for dwarfism to be caused/present.
- In homozygous, the presence of the two FGFR3 genetic mutations (both achondroplasia genes—one from each parent), causes even more delay in growth, a far more severe process of achondroplasia
  - Lungs are underdeveloped
  - Narrowing of the foramen magnum is tighter
  - Respiratory issues can either stem from the underdeveloped lungs (restrictive pulmonary disease), or lack of “signal” from the brain telling the child to breath, because of the extreme narrowing/tightness in the cervical spinal cord region.
  - The body is not creating red-blood cells (RBCs) fast enough to keep a reserve, to help the flow of oxygen, due to the smaller bones and therefore, bone marrow. The average baby is born with enough to sustain until they build up enough RBCs to maintain hemoglobin levels (approx 30 days). Because DD babies can't keep up, the lack of RBCs, even on an oxygen tank, fails to be enough. 5 weeks after our daughter was born, she went into distress. After a blood transfusion we learned of this complication. It was exactly a month later she passed (timeline of avg reserve creation)

### Planning for birth and after:

- Birth Plans aren't one size fits all
  - The medical team should support the parents' wishes on birth plans, provided the mother's health is taken into account - whether it's an earlier termination, an early vaginal delivery, or a full-term pregnancy with a cesarean section, or any plan in between.
    - Termination
    - Early Vaginal Delivery
    - Full term c-section
  - Parents rights and time spent with baby
  - Decisions after birth
  - There are levels of support to give to baby, and parents should know their options
    - No support - comfort care so baby is not in distress
    - Minimal oxygen
    - CPAP
    - Intubation
    - All can contribute to the length of life but just because you pick one, it does not automatically mean you get “x-time” with the baby. Each baby is different and their comfort should be top priority.
- Managing grief on your own timetable
  - LPA support groups

- Local groups/resources
- Maternal mental health therapists