

## **Handout #6**

### **Pediatric Genetics Vocabulary Exercises**

### **Conversion into a Non-English Language**

**Instructions:**     *Translate or sight translate the sentences below into your non-English language.*

1. That genetic material – some people call it the blueprints of life – it carries the instructions for how we're going to grow and develop, how our body's going to function, what we're going to look like.

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2. That genetic material is inside nearly every single cell in our body, packaged in these structures called chromosomes.

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3. These genes are actually the sets of instructions. One way you might think about would be like, say, a beaded necklace. So the necklace would be a chromosome, and each bead would be a gene.

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4. Some chromosomes can have an extra piece, or a missing piece, or pieces that have been sort of cut out and flipped end-over-end and stuck back into the chromosome.

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5. "Fragile X" – that's a funny name – but it describes a condition that typically affects boys, and it is a specific test that looks at the gene, that's like the bead, on the X chromosome.

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6. We'll also order what we call a "microarray." This is another type of test that will allow us to see if there are any places on your son's chromosomes where there are more or fewer genes from what we would expect. I

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7. If we do find an abnormality on one of these tests, then that probably gives us the answer as to what's causing your son's developmental delay.

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8. If we don't find anything clinically significant on the karyotype or on the microarray or in the biochemical testing, we can try some more sophisticated tests such as exome sequencing.

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9. Looking at your son's pedigree, it doesn't appear that there is any genetic predisposition for this delay. It doesn't seem to run in the family, so it may be due to just a random variant.

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10. When he's a bit older, we'll do a developmental assessment to evaluate his cognition, his motor abilities, his adaptive skills, and to see if there is any intellectual disability.

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