

## **M. Jawad Javed, MD**

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### **Why this genomic testing is so special**

The best way to think about it is when you're standing outside of a forest and you see a bunch of trees in front of you, the chromosomes are essentially each individual tree and you're able to identify each tree and see if there's one tree that's different than another tree or maybe they have two of the same tree standing together. What we're able to do now with the technology is not only look at the trees, but we're able to go deep into the forest and look at a single branch and what part of the branch - if it's closer to the leaf, if it's closer to the trunk - and where those defects lie within the forest themselves. :35

### **Searching for answers for very rare conditions**

These things are very rare, so not just a state or an institution or the world – globally - this may be an instance where it's one in several million that the child may have those same constellation of symptoms. When you're dealing with such a small numbers, it's very important to have an ability to be able to cohort these cases together and maybe give families hope to identify why something might be occurring and if there's potential to fix it and treat it. :34

### **Why these tests give families answers – looking at A through Z**

It really provides families that thought there was no hope – hey, there's something not right with my kid - and the traditional testing methodologies that we had doesn't explain everything because it maybe looks at A through X but it doesn't look at A through Z. Now we have the ability to look at A through Z. :19

### **Very exciting field**

This is a very exciting field because it involves a lot of the technology that's there, it involves institutions to collaborate with one another, it involves people to have dialogue and discussions with families. But it also provides a chance for new opportunities and new hope. :18