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# What's Next After the Search for a Diagnosis? Hearing families' experiences

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# Hearing families' experiences

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- Receiving results
- What happens next?
- Personal utility
- Implications for health services and RD community

# What to ask?

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*“We have never been in a situation like this. So we didn’t even know what to expect, what to ask. And where to focus.”*

(CZ 1)

# “Overloaded”

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*“We never really went into ‘what to expect’ because the day we went in and got the diagnosis, I think we got overloaded with what we got told, in the nicest possible way...”*

# “Like watching TV”

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*“...It was interesting listening what she told us. It was like if you’re watching something on TV.”*

(UK 4)

# What impact?

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*“I feel that the people are usually only sitting there and listening. They have a chance to ask questions but they are shocked, shocked and I get to see the impact only after a time. What the impact was and everything else...”*

*(Czech clinical geneticist)*

# Receiving results

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- About giving information and explaining what the result is
- Clinician-led
- Families focused on 'take it in'
- Difficult to come up with questions

# Hearing families' experiences

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# “No change”

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*“To be honest, not much has changed because of the results. Not much has changed because the network of different examinations and doctors and special schools and respite services had to be built up before. The fact that I got the label that my daughter has DDX3X did not change that too much. Not for me. We have the diagnosis. But everything else goes on, the problems to find a school, an assistant. The problem to somehow get any kind of help for my child, this continues.”*

(CZ3)

# What to do with it?

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*“It’s a bit like going to your doctor and the doctor saying: ‘you’ve got this condition, now go away.’”*

(UK2)

# What to do with it?

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*“I don’t think that finding a pathogenic variant is the same as having a diagnosis... There’s a gap to cure... Finding the cause is different than having a diagnosis.”*

*(UK clinical geneticist)*



# What to expect

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*“Through this we might get to talk to other people that have somebody that’s older than Claire, and know what to expect.”*

(UK 4)

# What to expect

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*“I was interested in what it’ll bring in the future. And what the symptoms are...But there is not a lot known about it yet, because there are really few children. And all of them are relatively young ... So there’s still very little information about what the future of the person looks like when they are say fifty or sixty.”*

(CZ 3)

**Parents of Jindřich (18 y/o)**

**Negative result**

**They are in the care of the genetic clinic  
of Motol University Hospital for 15 years**

**We have only one condition  
and we really insist on it permanently.**



# What happens next?

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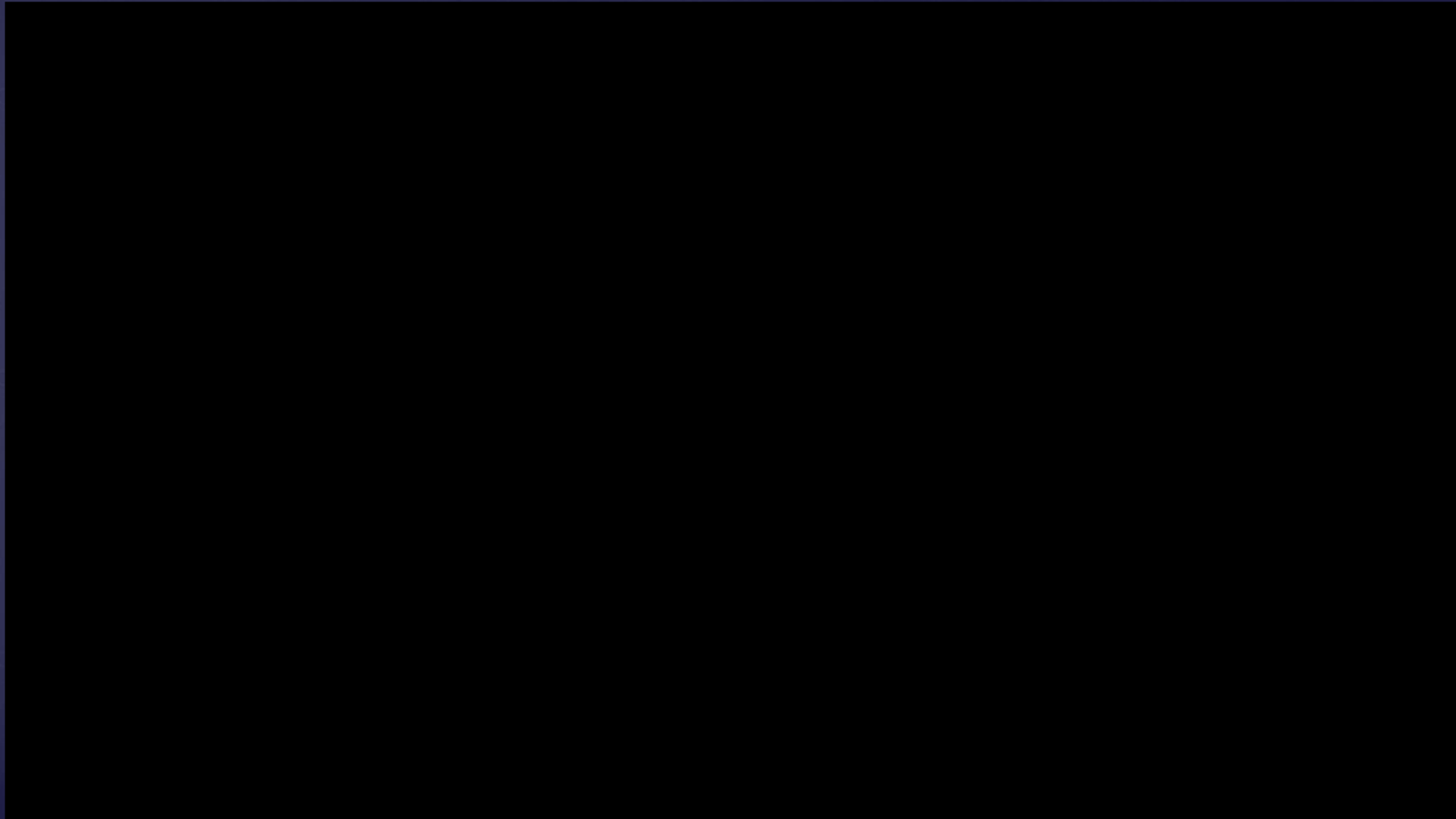
- Diagnosis provides answers, but also raises new questions
- Struggle to find and access right kind of support
- Poorly coordinated care
- Uncertainty about management and prognosis

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# “That was significant to us”

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- Personal utility broader than clinical utility
- Benefits for the whole family and for others
- Process and not just the outcome

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# Implications

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- Getting a diagnosis is not the end
- Unmet needs and unanswered questions
- Not just 'expectations' (*understanding*) but also enduring uncertainty (*lived experience*)

# Implications

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- Personal utility: how to think and talk about 'diagnosis'?
- Implications for services: follow up care and family-centred communication