What was the research about?
Intellectual disability, or ID, and autism spectrum disorder, or ASD, are health problems that affect how people learn and relate to others. Many different genetic changes can cause ID or ASD. Knowing the genetic change can lead to better treatment and care for children with ID or ASD. Genetic tests can find the genetic change. But the results of these tests can be hard to understand.

In this study, the research team worked with parents of children with ID and ASD to design a report that helped parents learn and talk about genetic testing results. The report included

- A clear explanation of the test results and next steps for care
- Answers to common questions about the diagnosis
- A table describing what type of health issues the child might have and at what age
- Links to resources specific to the diagnosis
- A glossary of medical terms

The report was available on an online patient portal. A patient portal is a secure website that gives patients, or in this case parents, access to their children's health records.

The research team looked at how parents used the online report. They wanted to compare parents who accessed the reports with parents who didn't.

What were the results?
Of the 46 children who had reports on the patient portal, only nine parents opened the report. In interviews, parents said that the email message telling them the report was ready wasn't clear. Parents whose children didn't receive a genetic diagnosis said they didn't have a reason to open the report.

Two parents who looked at the report said they used it to talk with their children's teachers and doctors. The report helped them feel confident in talking about their children's problems.

Who was in the study?
This study included parents of 46 children with ID or ASD who received genetic testing results. All lived in rural areas of Pennsylvania. The research team interviewed 23 parents, and 52 parents filled out a survey. Of parents who filled out the survey, 96 percent were white, and 58 percent were women. In addition, 88 percent were married, and 63 percent had some college education.

What did the research team do?
First, the research team worked with parents of children with ID, ASD, or both to design the report. Doctors and other providers reviewed the report, and
the team made changes. Next, a separate team put the report on the patient portal.

Then, the research team invited parents to take part in the study by taking a survey. Half of those who took the survey received a link to the genetic testing report on the patient portal. The other half received the link three months later. All parents in the study received usual care. Usual care included an in-person clinic visit to explain the genetic testing results, followed by a summary letter. The letter described the diagnosis and care management.

To look at parents’ use of the online report, the research team interviewed parents about barriers to accessing the report and how they used the report.

The research team worked with a patient, parents of children with genetic disorders, and doctors in all parts of the study.

What were the limits of the study?
Too few parents opened the report for the research team to compare the usefulness of the online report with the summary letter as planned. Future research could look at other ways to share the report with parents.

How can people use the results?
Parents in this study didn’t open the report as planned. But similar reports may be helpful for parents of children who receive genetic testing results. Clinics that offer genetic testing may look at whether reports like this would be helpful if given to parents in other ways.

To learn more about this project, visit www.pcori.org/Williams132.