

# State of North Carolina

## **ROY COOPER**

**GOVERNOR** 

#### PHELAN-MCDERMID SYNDROME AWARENESS DAY

2020

### BY THE GOVERNOR OF THE STATE OF NORTH CAROLINA

#### A PROCLAMATION

WHEREAS, Phelan-McDermid syndrome, also known as 22q13 deletion syndrome, is a rare genetic disorder caused by the deletion or structural change of part of chromosome 22, or a mutation of the SHANK3 gene; and

WHEREAS, although the range and severity of symptoms may vary, Phelan-McDermid syndrome is generally characterized by some degree of intellectual disability, delayed or absent speech, symptoms of autism spectrum disorder, low muscle tone, motor delays, and/or epilepsy; and

WHEREAS, is estimated that 1 percent of people with autism have Phelan-McDermid Syndrome; this genetic condition is often undetectable in standard genetic screening and requires a fluorescence in situ hybridization (FISH) test or whole exome sequencing (WES) to confirm the diagnosis; and

WHEREAS, individuals, parents, and advocacy groups can increase awareness of Phelan-McDermid syndrome and its symptoms through scientific education opportunities and informational exchanges; and

WHEREAS, the State of North Carolina joins in raising awareness of Phelan-McDermid syndrome and encourages people to promote resources, advocate research, and provide support for individuals and their families who are affected by this disease in our communities;

**NOW, THEREFORE,** I, ROY COOPER, Governor of the State of North Carolina, do hereby proclaim October 22, 2020, as "PHELAN-MCDERMID SYNDROME AWARENESS DAY" in North Carolina, and commend its observance to all citizens.



Roy Cooper

IN WITNESS WHEREOF, I have hereunto set my hand and affixed the Great Seal of the State of North Carolina at the Capitol in Raleigh this thirteenth day of October in the year of our Lord two thousand and twenty and of the Independence of the United States of America the two hundred and forty-fifth.