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OPKO Health's GeneDx Adds Repeat Expansion Analysis Genetic Tests for Diagnosis of Spinocerebellar Ataxia (SCA), Friedreich Ataxia, and Other Common Forms of Hereditary Ataxia

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GAITHERSBURG, Md., January 26, 2021 — GeneDx, Inc., a wholly owned subsidiary of BioReference Laboratories, Inc., an OPKO Health company (NASDAQ:OPK), today launched several new genetic tests, including repeat expansion analysis for spinocerebellar ataxia (SCA), Friedreich ataxia, and other common forms of hereditary ataxias. With these additions, GeneDx has created a comprehensive and affordable offering that covers the vast majority of genes involved with pediatric-onset and adult-onset ataxias.

Ataxia refers to clumsiness or a loss of balance and coordination that is not due to muscle weakness. While there are a number of factors that can cause ataxia, approximately 60-70% of ataxia cases have an underlying genetic cause.[1,2] Genetic testing for adult-onset ataxia is currently limited to a small number of diagnostic laboratories, most of which only offer a portion of the relevant testing, have long turnaround times, and high out of pocket costs to patients.

Most types of adult onset hereditary ataxia are caused by nucleotide repeat expansions within the deoxyribonucleic acid (DNA) and are usually identified by specialized testing. The remaining types of hereditary ataxia may be caused by single nucleotide variants (SNVs) and copy number variants (CNVs) that can be identified by sequencing and deletion/duplication testing.

"Individuals with ataxia need more diagnostic testing options for genetic forms of the disease," said Amanda Lindy, Ph.D., FACMG, Director of Neurogenetics for GeneDx. "Historically, genetic testing for ataxia has been limited, creating a barrier for some individuals to obtain testing. GeneDx's expanded test offerings provide the flexibility of

ordering single or multi-gene repeat expansion analyses, concurrently or reflexively, with a phenotypically driven Xpanded panel or an exome. Thus covering the broadest possible differential, delivering more answers to patients and their families, and enabling precision medical management."

"GeneDx has a deep clinical knowledge of the ataxias and related movement disorders, gained from our long history and industry-leading development of neurogenetic testing," said Sean Hofherr, Ph.D., FACMG, Executive Vice President and CLIA Laboratory Director of GeneDx. "Expanding our menu to include adult-onset ataxias, in addition to the existing portfolio for childhood-onset ataxias, underscores GeneDx's commitment to rare disease identification as well as filling an unmet need for patients and providers, alike."

About GeneDx, Inc.
GeneDx, Inc. is a global leader in genomics, providing testing to patients and their families from more than 55 countries. Led by its world-renowned clinical genomics program, GeneDx has an acknowledged expertise in rare and ultra-rare genetic disorders, as well as one of the broadest menus of sequencing services available among commercial laboratories. GeneDx offers a suite of additional genetic testing services, including diagnostic testing for hereditary cancers, cardiac, mitochondrial, neurological disorders, prenatal diagnostics and targeted variant testing. GeneDx is a subsidiary of BioReference Laboratories, Inc., a wholly owned subsidiary of OPKO Health, Inc. To learn more, please visit www.genedx.com.

About OPKO Health
OPKO is a multinational biopharmaceutical and diagnostics company that seeks to establish industry-leading positions in large, rapidly growing markets by leveraging its discovery, development, and commercialization expertise and novel and proprietary technologies. For more information, visit www.opko.com.

Cautionary Statement Regarding Forward-Looking Statements
This press release contains "forward-looking statements," as that term is defined under the Private Securities Litigation Reform Act of 1995 (PSLRA), which statements may be identified by words such as "expects," "plans," "projects," "will," "may," "anticipates," "believes," "should," "intends," "estimates," and other words of similar meaning, including statements regarding GeneDx's test offerings and the effectiveness and utility of its ataxias tests, as well as other non-historical statements about our expectations, beliefs or intentions regarding our business, technologies and products, financial condition, strategies or prospects. Many factors could cause our actual activities or results to differ materially from the activities and results anticipated in forward-looking statements. These factors include those described in the OPKO Health, Inc. Annual Reports on Form 10-K filed and to be filed with the Securities and Exchange Commission and in its other filings with the Securities and Exchange Commission. In addition, forward-looking statements may also be adversely affected by equipment and reagent shortages, general market factors, competitive product

development, product availability, federal and state regulations and legislation, the regulatory process for new products and indications, manufacturing issues that may arise, patent positions and litigation, among other factors. The forward-looking statements contained in this press release speak only as of the date the statements were made, and we do not undertake any obligation to update forward-looking statements. We intend that all forward-looking statements be subject to the safe-harbor provisions of the PSLRA.

References:

1. Fogel, Brent L et al. “Exome sequencing in the clinical diagnosis of sporadic or familial cerebellar ataxia.” JAMA neurology vol. 71,10 (2014): 1237-46. doi:10.1001/jamaneurol.2014.1944
2. Németh, Andrea H et al. “Next generation sequencing for molecular diagnosis of neurological disorders using ataxias as a model.” Brain: a journal of neurology vol. 136,Pt 10 (2013): 3106-18. doi:10.1093/brain/awt236.

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