Nemours. Children's Health System

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11/3/2015

Amer Haider
Growing Stronger
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RE: Grant, *Survey of C-Type Natriuretic Peptide Levels in People with Skeletal Dysplasia*

Dear Mr. Haider:

We are pleased to submit this report on the above named project, supported by a grant from Growing Stronger.

C-type natriuretic peptide (CNP) is a hormone that is an important regulator of skeletal growth. We have found that people with achondroplasia have high levels of CNP in their blood, suggesting their bones are resistant to this hormone. This resistance may explain some of their skeletal problems. A modified version of CNP is being tested as a drug to treat the skeletal growth problems of achondroplasia. For this study, we are measuring CNP levels in the blood from people with other types of skeletal dysplasia, to see if CNP resistance or possibly CNP deficiency might be present. Identifying the skeletal dysplasia types with altered CNP levels will help us to better understand these conditions and will suggest those in which CNP treatment may be beneficial.

Our work on CNP and achondroplasia was published in February of this year (Olney RC, Prickett TC, Espiner EA, Mackenzie WG, Duker AL, Ditro C, Zabel B, Hasegawa T, Kitoh H, Aylsworth AS, Bober MB. C-type natriuretic peptide plasma levels are elevated in subjects with achondroplasia, hypochondroplasia, and thanatophoric dysplasia. *J Clin Endocrinol Metab.* 2015 Feb;100(2):E355-9).

To date, the current project has received approval from the Nemours Institutional Review Board (for protection of human research participants) and the administrative structure has been established. Recruitment began this summer at both the study sites of Nemours/A.I. duPont Hospital for Children (Wilmington, DE) and Nemours Children's Hospital (Orlando, FL). Twenty subjects (11 boys and 9 girls) have been enrolled, ranging in ages from 4 months to 17 years. The diagnoses of the subjects can be found below:

Disorder	Subjects
osteogenesis imperfecta	5
metatropic dysplasia	3
Majewski osteodysplastic primordial dwarfism type II (MOPD II)	3
acampomelic campomelic dysplasia	1
cleidocranial dysplasia	1
dyspondyloenchondromatosis	1
Jeune syndrome	1
Kneist dysplasia	1
Morquio syndrome	1
pseudoachondroplasia	1
rhizomelic chondrodysplasia punctata (RCDP)	1
spondyloepiphyseal dysplasia congenita	1

The table shows we are sampling a wide range of syndromes. The samples from the subjects are being stored and will be assayed in two batches; one in the Spring and the last in the Fall of 2016. No results are currently available to be reported.

In summary, the project has been initiated and is progressing as planned. We anticipate no difficulties reaching our goal of 100 subjects in the final year of the study. We would like to express our appreciation to Growing Stronger for supporting this important research.

Sincerely,

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