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## African Cleft GWAS Signal Replication in an Independent African Cohort

**Alexandria, VA, USA, June 17, 2022** – A study aiming to replicate GWAS signals and identify new signals by combining the replication and discovery using meta-analysis for SNPs will be presented by Mary Li of the University of Iowa, Iowa City during the Interactive Talk presentation, “African Cleft GWAS Signal Replication in an Independent African Cohort”, which will take place on **Friday, June 24<sup>th</sup>, 2022 at 8 a.m. China Standard Time (UTC+08:00)** during the “Clefts, Sutures, and Syndromes” session at the [100<sup>th</sup> General Session and Exhibition of the IADR](#), to be held in conjunction with the 5<sup>th</sup> Meeting of the IADR Asia Pacific Region.

Orofacial clefts (OFCs) are congenital birth defects where the independently derived facial primordia that form the orofacial complex fail to fuse properly during embryonic development. Recent African genome-wide association studies (GWAS) for OFCs identified 170 near significant SNPs ( $p=10^{-5}$ ).

170 GWAS derived SNPs were genotyped on 3000 replication samples from participants in Ethiopia, Ghana, and Nigeria. Genotyping was done using Fluidigm IFCs and results were recorded, integrated with previous GWAS data, and implemented in further meta-analyses. Association analyses were done using PLINK and meta-analyses using METAL. Of the 170 GWAS suggestive significant SNPs ( $p \leq 10^{-5}$ ), the study genotyped 96 SNPs and identified three significant SNPs in this new cohort after Bonferroni corrections. rs186309721 was significantly associated in all cleft types with a p-value of 0.0003234. Further subdivision of the cohort showed that rs2295012 was significantly associated with cleft lip with or without palate ( $p=0.0003424$ ). rs186309721 was found to be significantly associated with cleft palate only ( $p=0.0005241$ ). Meta-analysis between the prior GWAS and new cohort data showed that rs10257343 ( $p=3.30E-08$ ) was significantly associated with cleft palate only, with the same direction of effect in both GWAS and replication datasets.

Through this replication study and meta-analysis, the study identified additional genetic risk factors for OFCs that can be included in future clinical translational work for improved patient care.

[View this Interactive Talk in the IADR General Session Virtual Experience Platform.](#)

### About IADR

The International Association for Dental Research (IADR) is a nonprofit organization with over 10,000 individual members worldwide, with a Mission to drive dental, oral and craniofacial research to advance health and well-being worldwide. To learn more, visit [www.iadr.org](http://www.iadr.org).

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