

## AI in Pediatric Medical Imaging: Digital Dysmorphology for Diverse Populations with Genetic Syndromes

**Abstract:** Each year, over one million children are born with a genetic condition. These children experience a high incidence of serious medical complications and intellectual disability that require immediate treatment. Genetic conditions often present clinically with universally recognizable signs and symptoms, including recurring patterns of malformations on the face and body that are pathogenetically related. These patterns can be obvious or very subtle and manifest differently with age and ethnical background. However, quantitative facial measurements are not available to primary care physicians, pediatricians and neonatologist, nor are they performed in a standardized and reproducible fashion by geneticists. Delayed and erroneous diagnoses can cause significant suffering and irreversible injury.

Artificial intelligence (AI) is redefining the interpretation of large clinical data for informed and predictive healthcare. But AI faces challenges when data are limited, such as in pediatric health and rare diseases. The talk will present recent advancements in quantitative imaging and AI to detect facial dysmorphology. Developed at the Sheikh Zayed Institute for Pediatric Surgical Innovation at Children's National Health Center, mGene is a non-invasive, smart phone application that identifies a wide array of genetic syndromes. The technology detects specific, clinically-proven markers of these rare disorders using objective and quantitative facial analysis enhanced by machine learning. Working with the NIH National Human Genome Research Institute, we demonstrated widespread potential applicability of digital dysmorphology to diverse populations with distinctive dysmorphic features. mGene promises to make sophisticated genetic expertise accessible in areas without specialized genetic clinics, typically unavailable in community hospitals and the developing world.