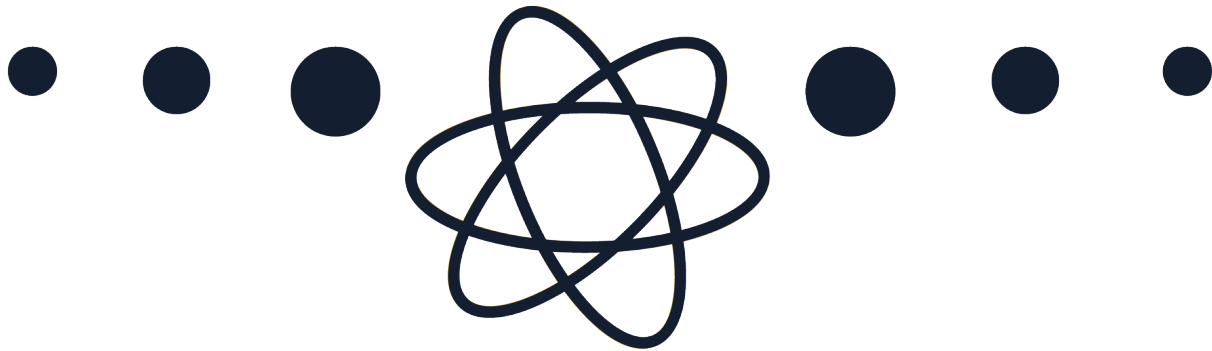

VOL 13 | MARCH 2026

THE PEDIATRIC JOURNEY

PROVIDER NEWSLETTER





CEO Message

El Paso Children's Hospital has evolved into a premiere pediatric hospital. The mission has remained the same and that is to provide the highest level of specialty healthcare for the children of our community.

At the beginning of a new year it is important to reflect on the journey and the work that still must be done. This past year we have continued to expand our resources throughout the region, allowing for more access to healthcare in some of the areas that need it most.

Last month we unveiled our new Pediatric Mobile Clinic. This mobile clinic will help El Paso Children's Hospital take specialty pediatric healthcare to the outlying rural areas of the community.

In addition to the Pediatric Mobile Clinic, we also unveiled the newest addition to our life-saving fleet called the "Jet" ambulance. This state-of-the-art vehicle will provide parents with the option to ride with their child as they are transported to our hospital. It also serves as a neonatal and pediatric mobile ICU.

These recent accolades accompany various others over the last several months, including the grand opening of another location for El Paso Children's Urgent Care in the Eastlake neighborhood and the continued growth of the El Paso Children's Multispecialty Center.

The future of El Paso Children's Hospital is being built every day through the hard work of our team. From physicians to frontline staff, each person plays a vital role in our continued progress. Your steadfast commitment to the health of our children is the foundation of our success.

Before El Paso Children's Hospital opened, more than 650 children each year had to leave El Paso to receive medical care. Fourteen years later, that number has been reduced by almost 85%. As the region's only pediatric specialty hospital, here is to many more years of growth, excellence, innovation, and—most importantly—**HEART**.

Dr Cindy Stout
DNP, RN, NEA, BC
President and CEO



Welcome NEW PHYSICIANS

El Paso Children's Hospital is proud to welcome a new group of exceptional healthcare professionals to its team. Their talent, dedication, and expertise represent more than an expansion of the hospital's medical staff—they are shaping the future of pediatric healthcare in the region.

Together with the existing staff, they will help advance El Paso Children's Hospital's commitment to keeping kids close to home for their healthcare—surrounded by family, culture, and the comfort of community—while delivering the excellence every child deserves.

El Paso Children's Hospital looks forward to the meaningful impact they will make.



Grace Nicksa, MD, MPH, FACS, FAAP
General Surgery



Kristopher Hooten MD, FANNS
Pediatric Neurosurgeon



Stephen Watty, MD
*Internal Medicine & Pediatrics/
Hospitalist*



in partnership with



Physician Spotlight

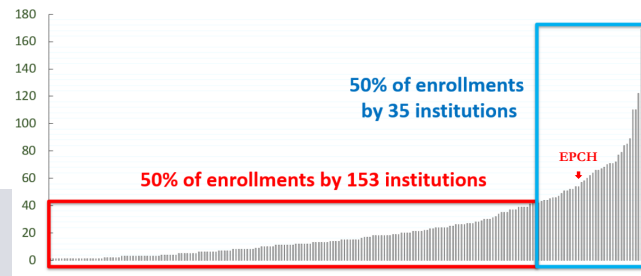
Dr. Benjamin Carcamo



COG 2025 FALL MEETING



COG Institutional Enrollment on MCI



Dr. Benjamin Carcamo and his research team were recently recognized by the National Cancer Institute (NCI) Community Oncology Research Program and the Children's Oncology Group. This recognition was to highlight their achievements in outstanding performance in patient enrollments at El Paso Children's Hospital. Dr. Carcamo was given a certificate of excellence along with the NCORP Mission Award.

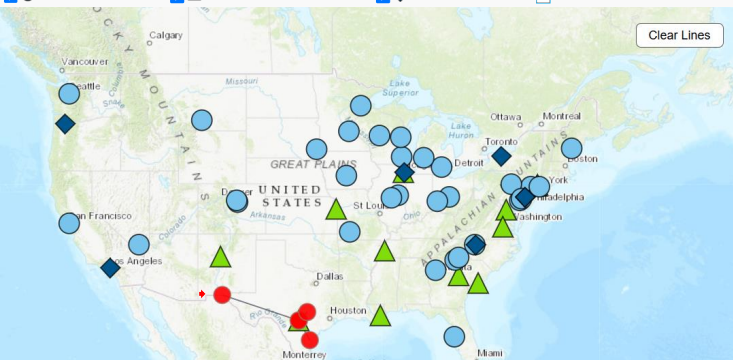
Texas Pediatric Minority Underserved NCORP

PI: Anne-Marie R. Langevin, M.D.
Site Address: San Antonio, Texas
Locations in: Texas

Hide affiliates and sub-affiliates

- Dell Children's Medical Center of Central Texas
- Driscoll Children's Hospital
- El Paso Children's Hospital
- Methodist Children's Hospital of South Texas
- University of Texas Health Science Center at San Antonio

Community Sites (31) Minority Community Sites (14) Research Bases (7) Affiliates / Sub-affiliates Sites (1024)



The PHO team at EPCH enrolled more patients than any of the other 4 institutions in the Texas Pediatric Minority Underserved NCORP group.



Texas Pediatric Minority Underserved NCORP

PI: Anne-Marie R. Langevin, M.D.
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Community and Minority/Underserved Sites



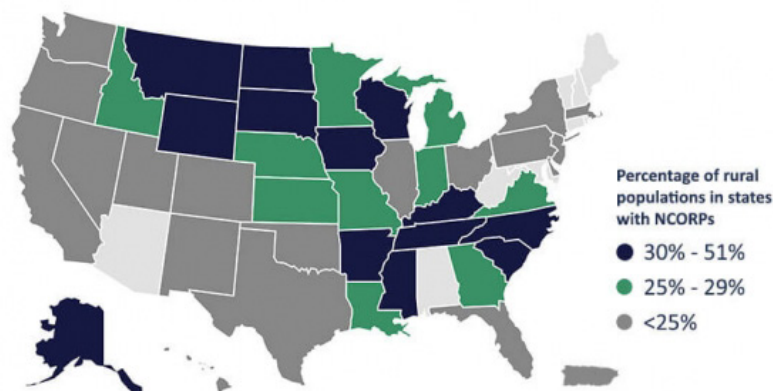
The PHO team at EPCH enrolled more patients than any of the other 53 institutions in the NIH NCORP grant for Community and Minority / Underserved sites across USA.



NATIONAL CANCER INSTITUTE

Clinical trials are available to people in communities across the U.S. through the NCI Community Oncology Research Program (NCORP), including 23 states with large rural populations.

23 RURAL STATES



For a full list of all the NCORP sites across the U.S.: <https://ncorp.cancer.gov/findasite/>

ALASKA ARKANSAS IOWA KENTUCKY MISSISSIPPI

MONTANA NORTH CAROLINA NORTH DAKOTA SOUTH CAROLINA

SOUTH DAKOTA TENNESSEE WISCONSIN WYOMING

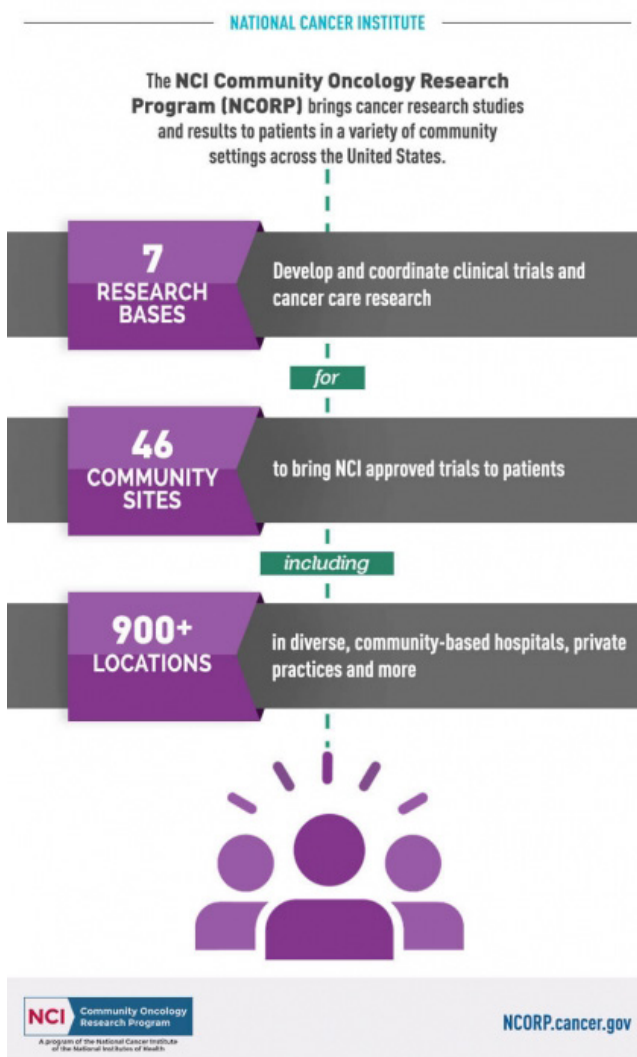
MISSOURI IDAHO INDIANA NEBRASKA LOUISIANA

MINNESOTA KANSAS MICHIGAN GEORGIA VIRGINIA

ncorp.cancer.gov

NCI Community Oncology Research Program
A program of the National Cancer Institute of the National Institutes of Health

Source: 2010 Census Data from the US Census Bureau



Physician Spotlight

Dr. Shauna Goldman

Congratulations to Dr. Shauna Goldman, as she was invited to speak at her alma mater, UT Southwestern Dermatology, where she completed medical school, Dermatology Residency, and her Pediatric Dermatology Fellowship. Dr. Goldman delivered a powerful presentation: “Building Bridges: Developing a Pediatric Dermatology Program on the U.S.-Mexico Border.” Her work demonstrates the importance of expanding access to specialized pediatric care and fostering cross-border collaboration. We are proud to celebrate her contributions and ongoing impact in the field of pediatric dermatology.





EPCH Physicians case report tracks new trend in VM management

Congratulations to our El Paso Children's Hospital physicians on their case report on TEK-related venous malformation of the orofacial region. This detailed report is meant to increase awareness among pathologists and clinicians of a promising new trend in VM management guided by molecular diagnosis. It also highlights the importance of multidisciplinary collaboration in providing optimal comprehensive care for these patients.

Quick glance at TEK-related venous malformation of the orofacial region: report of a case with multidisciplinary treatment guided by molecular diagnosis

Yi-Shing Lisa Cheng, DDS, MS, PhD, Daniel Bustamante, MD, David M. Yates, DMD, MD, FACS, Silvia Menendez, DDS, MS,d and Angela C. Chi, DMD

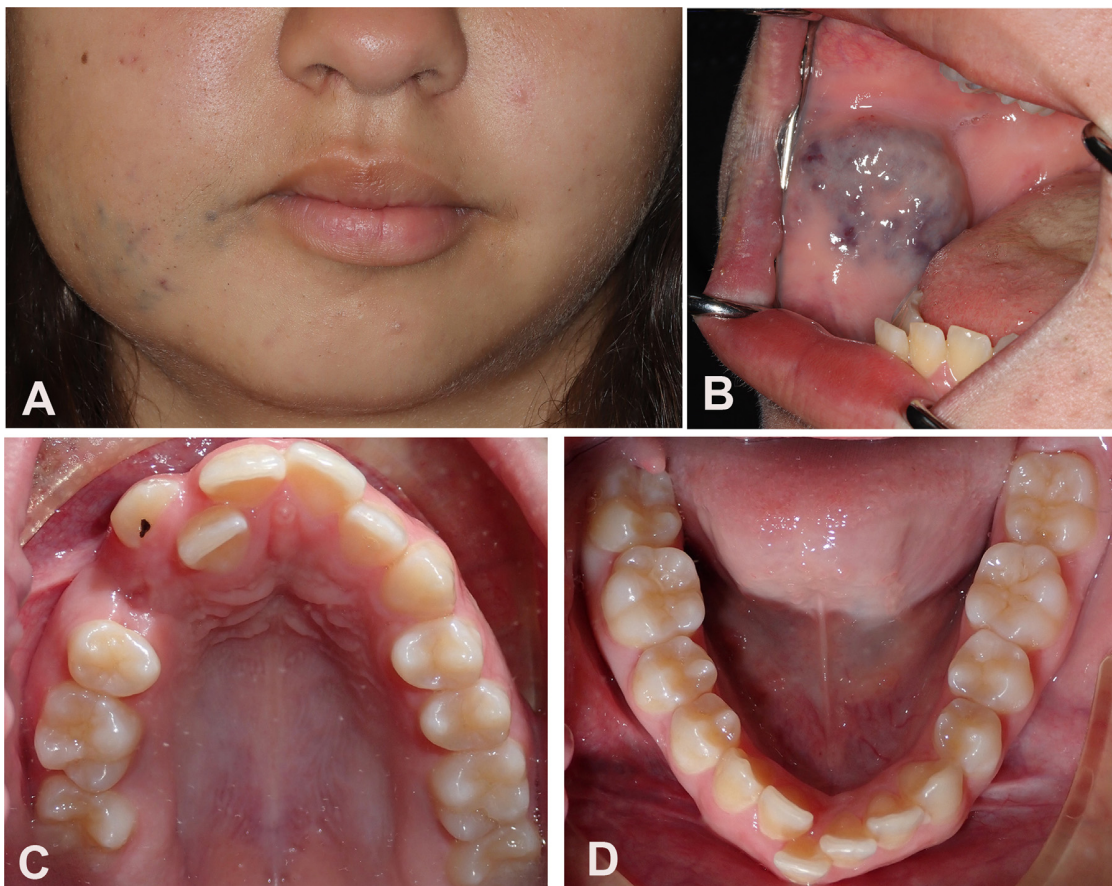


Fig. 1. Clinical photographs at initial presentation. (A) Enlargement of the right cheek area with dilated blue vessels evident on the skin surface. (B) Blue-purple buccal mucosal swelling. (C) Constriction of the right maxillary arch. (D) Constriction of the right mandibular arch.



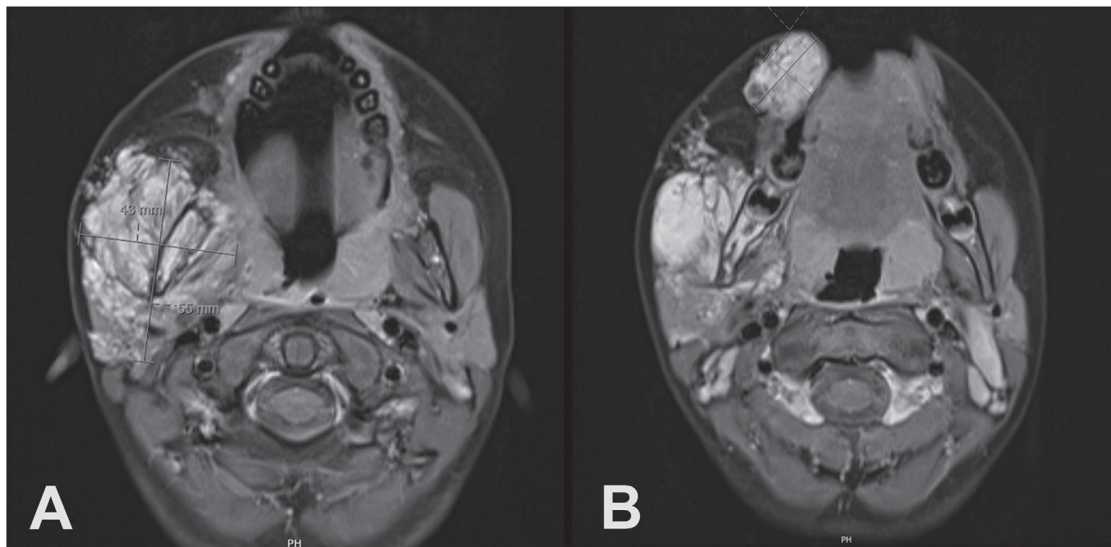


Fig 2. (A and B) Magnetic resonance imaging (T1-weighted), when the patient was 6 years old, showed a right facial mass measuring approximately 5.5 cm in greatest dimension and involving the right medial pterygoid muscle, right pterygoid fossa laterally, and the right masseter muscle and buccal space anteriorly. Compression on the right palatine tonsil caused mild airway narrowing.

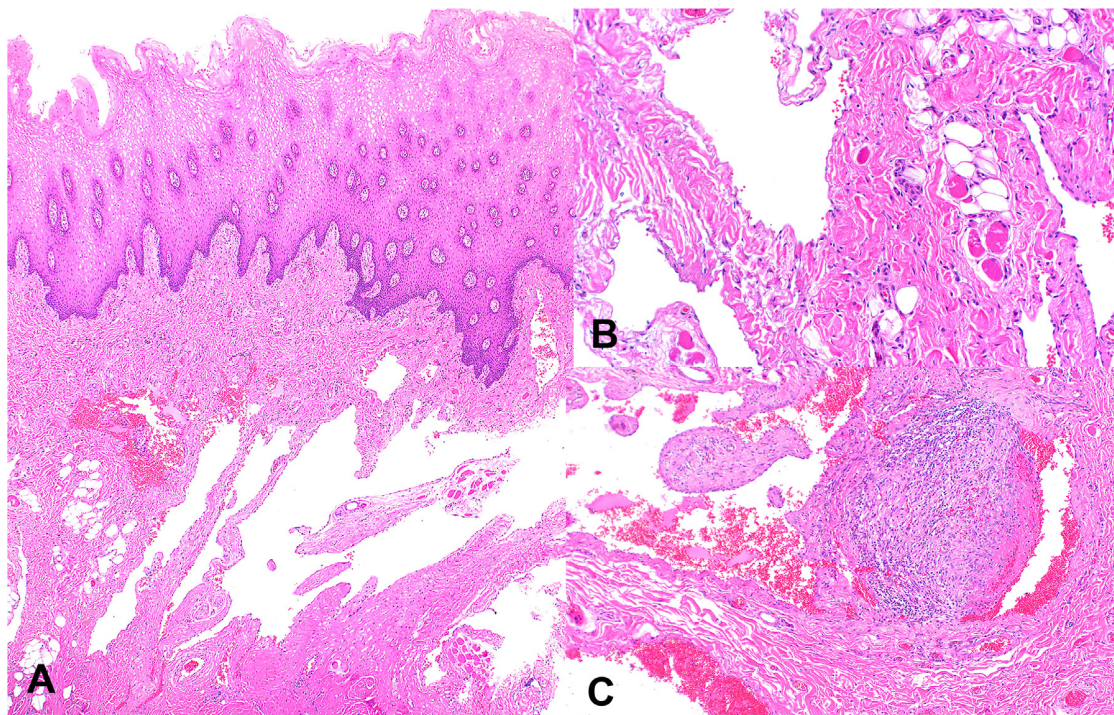


Fig. 3. Photomicrographs of the surgical debulking specimen. (A) Mucosa covered by hyperparakeratotic stratified squamous epithelium and exhibiting numerous thin-walled, dilated vascular channels within the lamina propria (hematoxylin and eosin, original magnification 100 $\times$). (B) Additional dilated vascular channels within the submucosa with adjacent atrophic skeletal muscle bundles and adipocytes (hematoxylin and eosin, original magnification 400 $\times$). (C) Intraluminal thrombosis (hematoxylin and eosin, original magnification 200 $\times$).

Vascular malformations (VMs) often affect the orofacial region, where they can markedly impact the quality of life. They frequently harbor mutations in TEK, PIK3CA, or other genes essential for angiogenesis. Alpelisib is a phosphatidylinositol 3-kinase inhibitor recently shown to be effective for treating VMs with PIK3CA or TEK mutations. Herein we describe a 13-year-old female with an orofacial VM present since birth. Based on clinical, histopathologic, and molecular findings, the final diagnosis was TEK-related venous malformation. She received multimodal therapy, consisting of sclerotherapy, surgical debulking, and systemic alpelisib.

Ten months after surgery and 7 months after terminating systemic therapy, the buccal mucosal aspect of the lesion was barely apparent. Molecular diagnostics and targeted agents have ushered in a new era of precision medicine for VMs. It is important for pathologists and clinicians to be aware of molecular-guided therapy as delivered via a multidisciplinary team-based approach for optimal management of patients with VMs. (Oral Surg Oral Med Oral Pathol Oral Radiol 2025;140:587593)

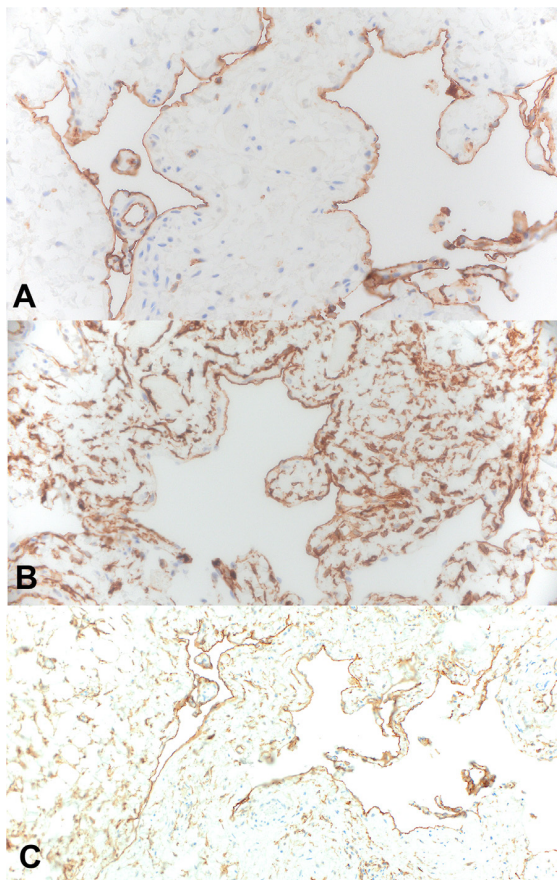


Fig. 4. Immunohistochemistry. (A) Strong immunoreactivity among the lesional endothelial cells for CD31 (DAB, original magnification 400×). (B) Strong immunoreactivity among the lesional endothelial cells and associated stromal cells for CD34 (DAB, original magnification 400×). (C) Strong immunoreactivity among the lesional endothelial cells and occasional low-intensity immunoreactivity among the stromal cells for CD10 (DAB, original magnification 400×).



Fig. 5. Eight months after surgery and 3 months after terminating systemic therapy, the intraoral aspect of the mass was barely apparent, with only a small portion evident toward the anterior aspect.



Where Complexity Meets Compassion: Level IV NICU

In modern neonatology, the intersection of scientific complexity and human compassion defines the standard of care for our most vulnerable patients. As understanding of neonatal conditions evolve and extreme prematurity becomes increasingly survivable, Level IV NICUs carry the responsibility of providing the highest level of coordinated, multidisciplinary care. El Paso Children's Hospital Level IV NICU reflects the mission, managing hundreds of critical newborns each year while advancing evidence-based practices, interdisciplinary collaboration, and family-centered decision-making.

A Regional Perspective: Understanding the Need

The border region faces unique demographic and clinical challenges. Preterm birth remains a significant contributor to neonatal morbidity and mortality. With no other children's hospital on the U.S. side of the border within 200 miles of El Paso, and a diverse population with limited access to specialty care, we manage a higher proportion of both surgical and non-surgical congenital anomalies. These patterns underscore the need for high-level perinatal coordination, early detection of fetal anomalies, and ready access to advanced neonatal services.



NICU Designation and Levels of Care

Texas has adopted neonatal unit designation/assignment of level process since 2013, which needs each NICU in Texas to go through a stringent process of meeting standards for this level of care. As per the DSHS website, EPCH NICU is one of 24 facilities throughout Texas to have a Level IV designation. The same source also mentions currently there are **224 designated neonatal facilities**. This is a nationally recognized need, The American Academy of Pediatrics (AAP) policy statement, “Levels of Neonatal Care,” reinforces the need for such advanced centers, emphasizing institutional commitment, standardized quality improvement programs, and highly specialized personnel. These standards guide the daily operations and long-term strategic development of Level IV NICUs across the nation.

“It Takes a Village”- Role of a Multidisciplinary Team

Exceptional neonatal care depends on the seamless coordination of an expanded clinical team. Beyond neonatologists and neonatal nurse practitioners, our NICU integrates respiratory therapists, pharmacists, dietitians, lactation consultants, therapists (OT/PT/SLP), case managers, social workers, child life specialists, pastoral care, and a host of pediatric subspecialists including interventional pediatric radiologists, geneticist, immunologists, pediatric neurosurgeons and other surgical specialties. Neonatal nurses undergo ongoing certification and AWHONN-directed education to maintain proficiency. This collaborative model allows infants with complex anomalies, surgical needs, and extreme prematurity to receive comprehensive support from birth through discharge.



Complex neonatal care extends beyond the walls of NICU, with involvement of our team beginning as soon as an anomaly is detected antenatally by the Obstetrician or Maternal Fetal Medicine specialists. The NICU further collaborates with neurosurgery, neurology, pediatric surgery, ENT, to have a perinatal plan of care. Fetal MRI helps characterize conditions such as congenital diaphragmatic hernia or neurological anomalies. Monthly perinatal conferences and joint consults ensure unified plans of care and eliminate “silos” that historically fragmented high-risk perinatal management. Such communication improves not only neonatal outcomes but also parental understanding and emotional preparedness. Continuity of physician care is another

foundational principle. Evidence shows that consistent neonatologist involvement reduces complications, improves parental satisfaction, and promotes safer transitions. Recommended staffing models include 1 to 2 week service blocks and limits on individual provider patient loads to mitigate burnout while preserving quality.

Ethical Decision-Making and Palliative Care

A big part of complex care is sometimes re-direction of care (including Do-Not-Resuscitate discussions and palliative pathways) requiring input from multiple disciplines. Continuing care for some conditions might mean worsening the suffering not just for babies but the whole family. Family meetings are conducted with the involvement of nursing, social services, case management, ethics and even sometimes legal teams. While pediatric palliative care expertise is still a limitation of our region, collaboration with community services such as hospice programs ensures compassionate guidance for infants with life-limiting diagnoses such as Trisomy 13 or 18.

Extreme Prematurity: The New Frontier

The management of infants born during the peri viable period **22–24 weeks gestation** (sometimes referred as Uber-preemies) has changed dramatically over the past decade. Improved understanding of antenatal interventions, including steroids, magnesium for neuroprotection, and coordinated maternal transfer to higher-level facilities, has reshaped survival curves. Active resuscitation—once controversial at 22–23 weeks—is now supported by emerging data from U.S. centers and international leaders like Japan and Sweden.

EPCH's outcomes reflect this progress: among 14 actively resuscitated infants born at 22–24 weeks, **57% survived**, with notably high survival among **singletons (86%)**. These results align with the broader trend showing that structured protocols, timely antenatal steroids, and consistent neonatal practices profoundly influence survival.



Photo from 2025 Advance Level IV NICU Reunion

Protocols for “uber-preemies” differ substantially from those for older preterm infants. Standardizing our approach to them along with consistent care in terms of respiratory support, cardiac management, and nutrition support seems to be the key to improving survival. Also important are the non-invasive ventilation strategies such as **LISA (Less Invasive Surfactant Administration)** and upcoming **SALSA techniques** that help reduce intubation during the later gestations. Meticulous thermoregulation, early nutritional optimization, and precise device selection are essential. Nationally recommended adoption of video laryngoscopy use by our unit also improves intubation success and safety.

Education, Outreach, and the Road Ahead

Our goal is to improve overall newborn survival and care and for the Border region and the extended community. EPCH, along with Texas Tech El Paso Physicians, conducts outreach education focusing on neonatal resuscitation, stabilization of extremely low birth weight infants, and hypoxic-ischemic encephalopathy management. Regular case reviews with referring hospitals strengthen regional neonatal systems of care.

Future goals include establishing a neonatal neurology center, optimizing two-year neurodevelopmental outcomes for extremely preterm infants, expanding high-risk clinic services, and integrating emerging evidence into clinical practice. At its core, the mission remains constant: delivering compassionate, state-of-the-art care that honors both the fragility and resilience of newborn life.

Table 1.
EPCH Level IV NICU Snapshot

Category	Details
Annual Admissions	~700
Average Daily Census	30 patients
Critical Care Needs	40–50% of admissions
Surgical Management	20–30% patients
Transfers In (HLOC)	15–20% patients
Transfers Out	1–3% patients
Preterm Birth Rates	10% <37 wks, 1.6% <32 wks, 0.5% <28 wks

Table 2. Key Elements of Extreme Preterm (22–24 Weeks) Management

Domain	Clinical Focus
Antenatal Care	Steroids, magnesium, antibiotics for PPROM, maternal transfer
Delivery Room	Thermoregulation, early monitoring, LISA/SALSA, appropriate interfaces
Respiratory	High frequency Jet Ventilation, Newer NIV support
Cardiac	Tailored PDA strategy differing from 24–27-week infants
Nutrition	Early IL use, modified feeding protocols
Outcomes	EPCH survival 57% (86% in singletons)



A Note from the desk of the CMO

Jeffrey David Schuster, MD
Chief Medical Officer

As Chief Medical Officer of El Paso Children's Hospital, it is my distinct honor to share with you a reflection on our journey and a glimpse into our future. Over the past 14 years, El Paso Children's Hospital has grown from a bold vision into a beacon of hope and healing for families across the region.

What began as the region's only dedicated children's hospital has evolved into a center of excellence—providing advanced, specialized care with compassion, innovation, and integrity.



“Our team has worked tirelessly to ensure that every child, regardless of their circumstance, receives the highest standard of medical care right here at home.”

This year marks another significant chapter in our story. We are proud to welcome three new physicians to the El Paso Children's Hospital team. These highly skilled specialists bring with them not only expertise in their respective fields, but also a deep commitment to our community. Their arrival expands our capabilities and ensures that we continue to meet the growing needs of our pediatric population.

The addition of these talented physicians reinforces our dedication to delivering world-class care without compromise. Each of them plays a vital role in advancing our mission: to bring the highest quality of care to the children of our region.

As we look to the future, we remain grounded in our purpose—to serve every child with excellence, compassion, and a commitment to continuous improvement. We are grateful for the trust you place in us and for the unwavering support of the El Paso community.

“Together, we are building a healthier future for our children.”





Two Texas Tech El Paso Pediatric Residents Selected to Present at Prestigious National Pediatric Conference

The Texas Tech University Health Sciences Center El Paso Pediatric Residency Program is proud to announce that two of its second-year pediatric residents were selected to present their scholarly work at the upcoming American Academy of Pediatrics (AAP) National Conference & Exhibition, taking place September 26–30 in Denver, Colorado. The AAP National Conference is one of the largest and most respected gatherings of pediatric professionals worldwide, and being chosen to present is a significant academic honor.

Dr. Cyrus Blanco, a PGY-2 pediatric resident with a growing interest in Allergy and Immunology, has delivered a presentation on “The Relationship Between Atopic Dermatitis and Food Allergy.” His research was conducted under the mentorship of Dr. David Jara, a specialist in Allergy and Immunology at El Paso Children’s Hospital.

Dr. Deepti Sharma, also a PGY-2 resident, has a strong interest in Child Psychiatry and has presented findings from her ongoing research on the mental health crisis in the Pediatric Intensive Care Unit (PICU). Her mentor, Dr. Avi Kopstick, a Pediatric Intensivist at Texas Tech/El Paso Children’s Hospital, is a dedicated advocate for trauma-informed care and the integration of mental health practices in critical care settings.

Their selection reflects the residency program’s commitment to academic excellence and innovation in pediatric healthcare. Congratulations to Dr. Blanco and Dr. Sharma on this remarkable achievement.



Dr. Cyrus Blanco



Dr. Deepti Sharma



Introducing the new Pediatric Medical Genetics and Genomics Clinic at EPCH

By Michelle Nguyen, MS, CGC and Maria Ethel Aguirre Flores, MD



What is Genetics and Genomics?

Genetics is the branch of biology that studies inheritance, including the interplay of genes, DNA variation and their interactions with environmental factors.

Genomics is a field of biology focused on studying all the DNA of an organism – that is, its genome. The study of genomics includes identifying and characterizing all the genes and functional elements in an organism's genome as well as how they interact. Medical Genetics and Genomics involves care from a Medical Geneticist and Genetic Counselor.

Medical Geneticist

A medical geneticist is a physician specializing in the diagnosis, management, and counseling of patients with known or suspected genetic conditions. They conduct a comprehensive review of medical history, perform detailed physical examinations, and order and interpret appropriate genetic testing. Based on genetic test results and diagnoses, medical geneticists develop personalized care plans that guide surveillance, treatment, and long-term management. Medical geneticists work closely with genetic counselors and a wide range of specialists to ensure coordinated, comprehensive care for individuals with genetic disorders.

Genetic Counseling

Genetic counseling is essential to ensure informed decision-making before genetic testing. Genetic counseling sessions discuss the potential for variants of uncertain significance, incidental findings, and secondary findings, helping families understand both the benefits and limitations of testing. In addition to facilitating informed consent, genetic counselors assess family history and inheritance patterns, estimate recurrence risks, and clearly communicate complex genetic information. They also help patients interpret results within their personal and familial contexts and connect families with appropriate support and advocacy resources.

Indications for referral

Genetic evaluation and genetic testing should be guided by the patient's clinical presentation and suspicion for an underlying genetic condition. Indications include multiple congenital anomalies, developmental delay, intellectual disability, autism spectrum disorder, unexplained seizures, bone fragility, or features suggestive of a specific genetic syndrome. The American College of Medical Genetics and American Academy of Pediatrics now recommend genetic testing for pediatric patients with developmental delay, intellectual



disability, ASD, or congenital anomalies. Early identification of a genetic cause can improve care, guide family planning, and expand access to targeted resources and clinical trials, while complementing ongoing specialty evaluations. An inpatient genetics consultation can be valuable for patients with an unknown etiology for their findings, particularly when a genetic diagnosis could clarify the cause of their findings and directly influence management and treatment decisions while hospitalized.

The Genetics Clinic Workflow

When a patient is referred to the Pediatric Genetics Clinic, the genetic evaluation process starts with determining whether a genetic diagnosis has already been established through prior genetic testing.

- If the patient has a confirmed genetic diagnosis, they will have regular visits with a pediatric geneticist for ongoing surveillance and management. Typically, annual follow-ups are recommended to monitor disease progression and address any emerging clinical concerns.

- If the patient does not have a confirmed genetic diagnosis, the genetics team assesses the extent of the patient's symptoms:

- o Patients with multisystemic involvement or diagnoses such as global developmental delay (GDD), intellectual disability (ID), autism spectrum disorder (ASD), seizures, or multiple congenital anomalies (MCA) will be evaluated by the medical geneticist (MD) and a genetic counselor (GC) for consideration of comprehensive genetic testing.

- o Patients with single-system involvement, such as short stature, kidney disease, birthmarks, or bone fragility, are assessed for prior genetic testing.

- For patients with prior genetic testing results, the genetics team carefully interprets specific genetic variants, including reviewing the latest literature on those variants, to provide a detailed, variant-specific clinical assessment and guide patient management.

- o If results are positive, both the geneticist and genetic counselor will meet with the patient and family to guide clinical management and counseling.

- o If results are negative or show variants of uncertain significance (VUS), the genetic counselor will see the patient and family to provide a discussion of genetic testing conducted, detailed interpretation of results, support coping, and offer referrals to appropriate resources if needed.

- If no prior genetic testing has been performed, evaluation by both geneticist and genetic counselor is initiated to conduct a comprehensive genetics evaluation including a detailed collection of medical history, developmental history, family history, determination of appropriate genetic testing, and next steps.

- If the patient is referred due to abnormal newborn screening results an urgent appointment is scheduled with the geneticist for clinical assessment and follow up testing as indicated. Based on clinical status a patient might be advised to undergo evaluation in the hospital, otherwise geneticist will provide family with information regarding the condition flagged by newborn screening, any recommended interventions and signs to be aware of. A final diagnosis is reached within 2-3 weeks and newborn screening state lab program is notified directly by geneticist. If a patient is identified as having an inborn error of metabolism they will continue to be followed closely with metabolic dietitian services provided during visit with geneticist.



Different Types of Genetic Testing

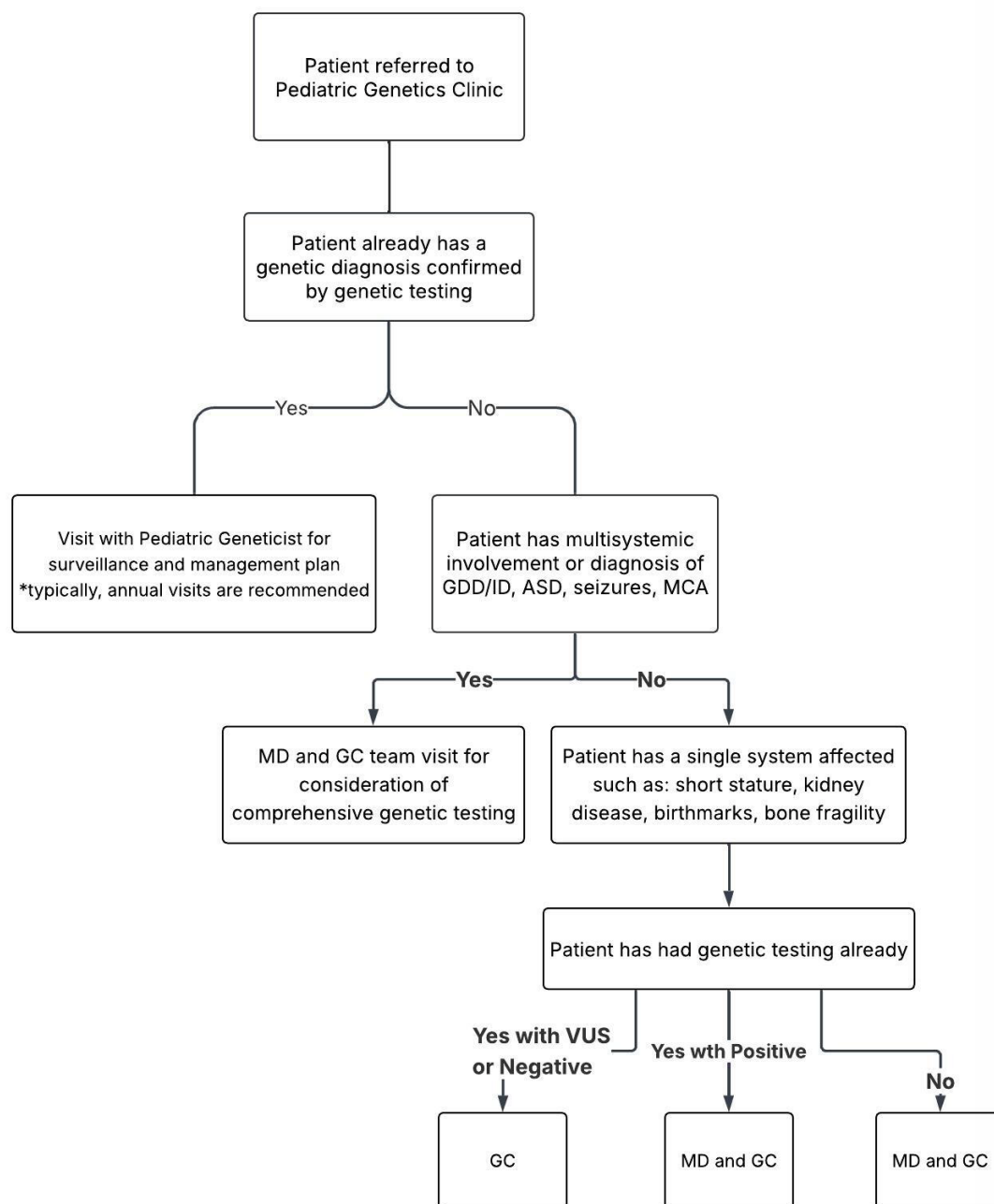


Figure 1. Workflow and care team for patients referred to the genetics clinic. GDD/ID = global developmental delay/ intellectual disability, ASD = Autism spectrum disorder, MCA = multiple congenital anomalies



Gene Therapy and Gene-Targeted Treatments

Gene therapy, which involves the editing or replacement of faulty genes, has emerged as a viable treatment option for several conditions. FDA-approved gene therapies are now available, including for adults with inherited retinal dystrophies and hemophilia B; for pediatric patients with spinal muscular atrophy and cerebral adrenoleukodystrophy; and for individuals with sickle cell disease and Duchenne muscular dystrophy.

Although the number of approved indications remains limited, the gene therapy landscape using is expanding rapidly. Since eligibility for gene therapy requires confirmation of the underlying pathogenic variant, comprehensive genetic testing is a critical component of patient diagnosis, management, and referral.

Beyond gene-editing and gene-replacement approaches, gene-specific pharmacologic treatments and gene specific clinical trials are also emerging. For example, a recently approved therapy for amyotrophic lateral sclerosis (ALS) targets ALS patients with pathogenic SOD1 variants. In other cases, not only the gene but the specific variant within the gene determines eligibility for treatment. Cystic fibrosis transmembrane conductance regulator

Type of genetic Test	Tests for	Indication for testing	Turnaround time
Karyotype	Aneuploidy (Trisomy 21, 18, and 13)	Multiple congenital anomalies (MCA), Physical exam findings consistent with T21, T18, T13	2-4 weeks
Chromosomal microarray	Submicroscopic chromosomal deletions and duplications (ie: 22q11.2 deletion syndrome DiGeorge, 4p deletion syndrome Wolf Hirschhorn)	MCA, short stature, mild developmental delay, failure to thrive	2-4 weeks
Single gene	Variants in one gene	High clinical suspicion for a single condition associated with variants in one gene	1-4 weeks
Panel	Variants in multiple genes associated with a shared phenotype or condition group	Clinical suspicion for a condition within a group of related disorders that may result from variants in multiple genes	2-4 weeks
Whole Genome Sequencing	Variants across the genome	Global developmental delay (GDD), intellectual disability, autism spectrum disorder (ASD), seizures	4-6 weeks
Rapid Whole Genome Sequencing	Variants across the genome (protein coding and non-coding genes)	Critically ill patients currently admitted to the hospital where a genetic diagnosis may impact acute management	7-10 days
Whole exome sequencing	Variants across the exome (protein coding genes which is ~ 1% of the entire genome)	When WGS is not accepted by insurance	4-6 weeks
Methylation studies	Abnormal DNA methylation affecting gene expression	Beckwith Wiedemann, Angelman, Prader Willi, Russel Silver	2-4 weeks

Table 1. Summary of commonly used genetic tests including the target of testing, typical indications, and turnaround times.

modulators, for instance, are designed to correct the defective protein caused by specific CFTR variants of Cystic Fibrosis. Both of these examples underscore the growing importance of genetics and genetic testing for genotype-guided treatment strategies in precision medicine. Although gene therapies and gene-specific treatments are not yet available for every condition, genetic technology continues to advance at a rapid pace, which is why obtaining a genetic diagnosis is important, as it may enable access to tailored therapies in the near future.

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