

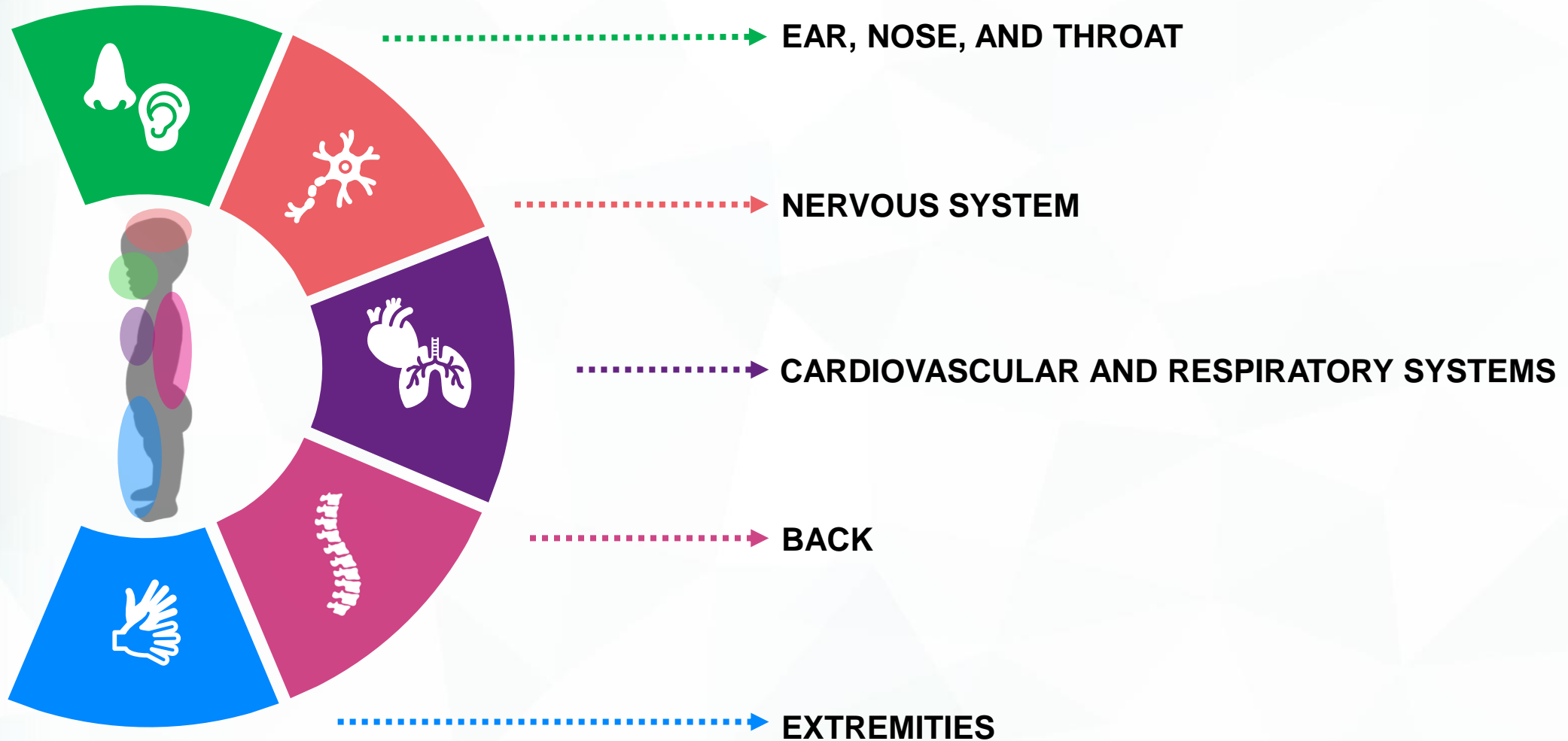
Achondroplasia – understanding a lifetime of management

Dr Nadia Merchant

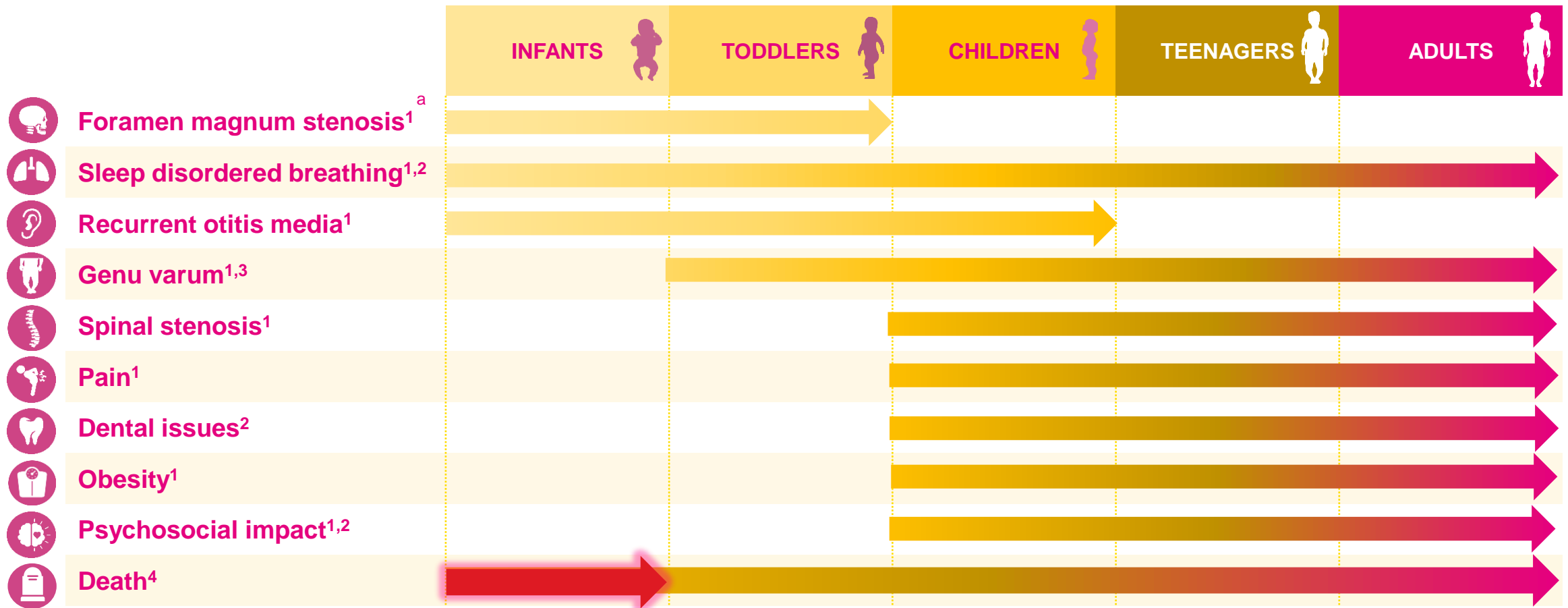
Pediatric Endocrinologist and Geneticist
Children's National Hospital, Washington, DC, USA



Children with achondroplasia may experience lifelong complications in multiple systems



Complications occur during different stages of life



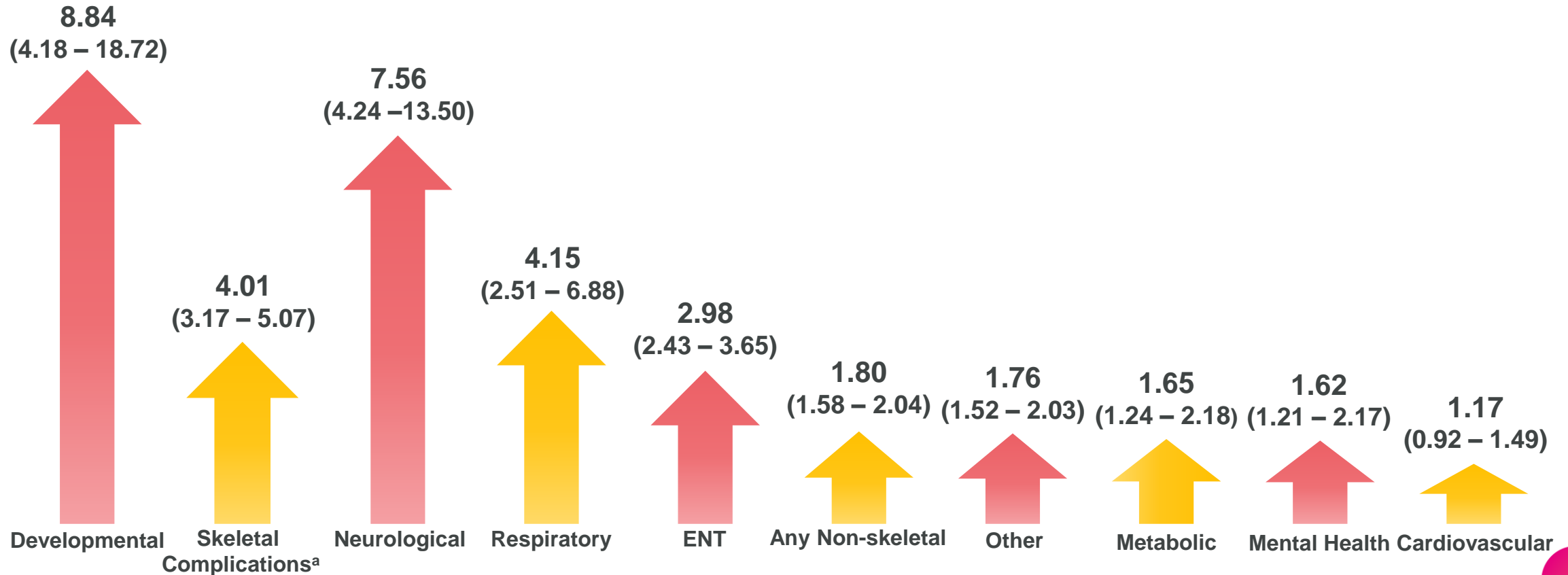
^aMajor concern, especially for sudden infant death syndrome

1. Hoover-Fong J, et al. *Pediatrics* 2020;145(6):e20201010; 2. Wright MJ, et al. *Arch Dis Child* 2012;97:129–134; 3. Lee ST, et al. *J Bone Joint Surg Br* 2007;89:57–61; 4. Simmons K, et al. *Birth Defects Res A Clin Mol Teratol* 2014;100:247–249



Rates of skeletal and non-skeletal complications in patients with achondroplasia are higher than the general population

Rate Ratio for Complications in Achondroplasia Patients (N=541) vs Matched Controls (N=2,052) in the UK



^aSkeletal complications included arthritis, osteoarthritis, falls, osteoporosis, reduced bone density, cauda equina syndrome, incontinence, neurogenic bladder/bowel, paraplegia, parathesia in addition to skeletal complications listed above

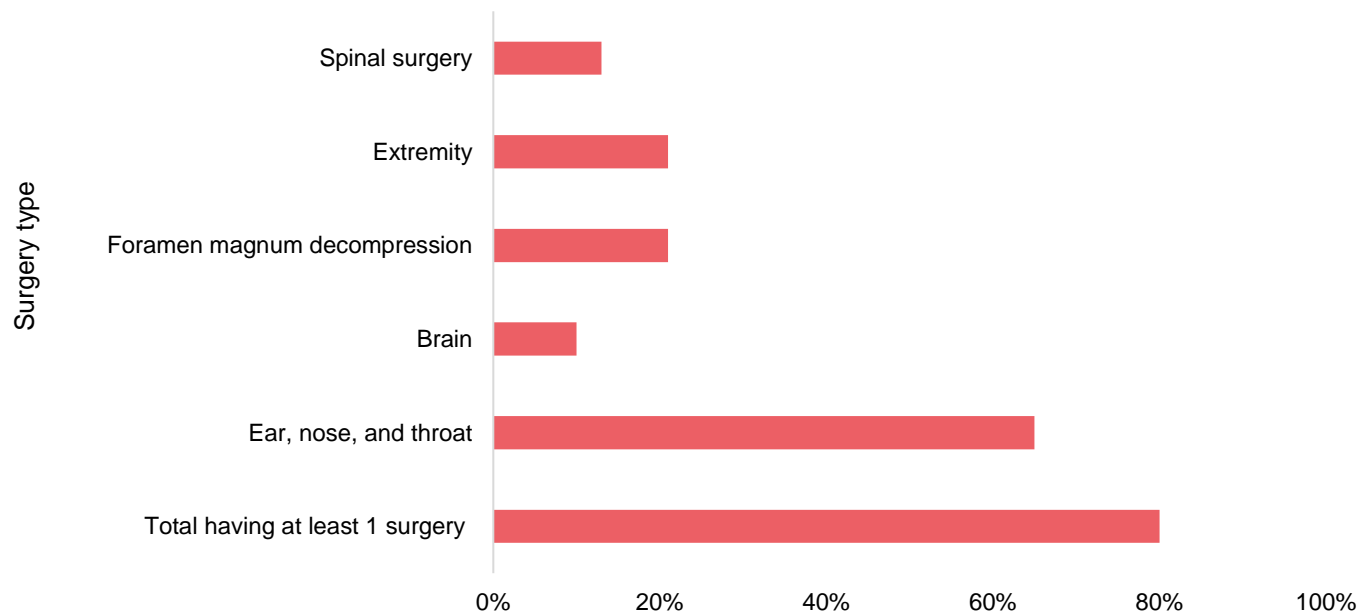
ENT, ear, nose, and throat

Pimenta JM, et al. Presented at Virtual ISPOR Europe, December 2021



Patients with achondroplasia also experience frequent surgeries

Common Surgeries in United States Patients with Achondroplasia, Aged 0 to 80 Years (CLARITY, US- based)¹
N=1,374



Similarly high rates of surgeries have also been found in UK and EU studies^{2,3}

Ear, nose, and throat includes tympanostomy with tube placement, primary tonsillectomy/adenoidectomy, and all revisions; Brain includes ventriculoperitoneal shunt placement and revision and ventriculostomy;

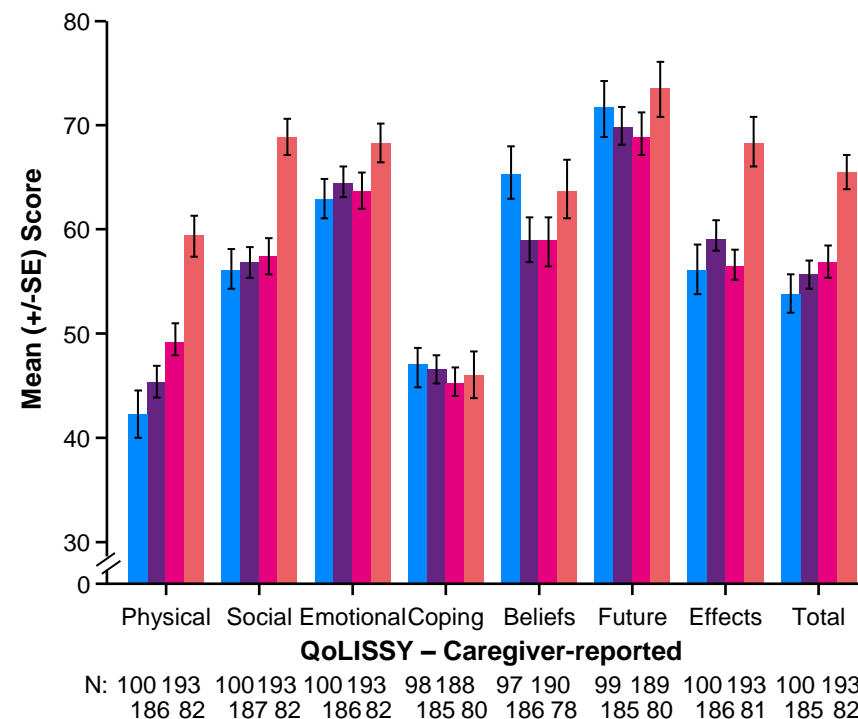
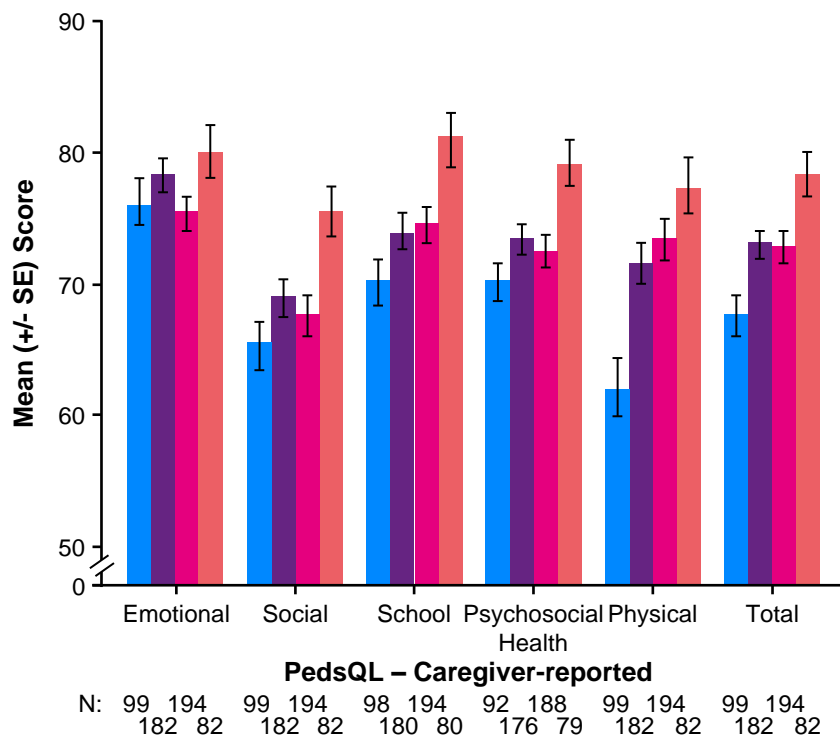
Extremity includes various procedures to correct genu vara and lower extremity bowing; Spinal surgery includes procedures from C3 through the sacrum

1. Hoover-Fong JE, et al. *Genet Med* 2021;23:1498–1505; 2. Pimenta JM, et al. Presented at Virtual ISPOR Europe, December 2021; 3. Maghnie M, et al. *Mol Genet Metab* 2021;132:S65-S66



Achondroplasia is associated with a reduced quality of life

In 901/301/302, mean scores across PRO tools were generally lower in children with greater height deficit (≤ -6 SDS) compared with taller children with height Z-score greater than -4 SDS



■ ≤ -6
 ■ > -6 to ≤ -5
 ■ > -5 to ≤ -4
 ■ > -4



Multisystem disorders require multidisciplinary care¹⁻²

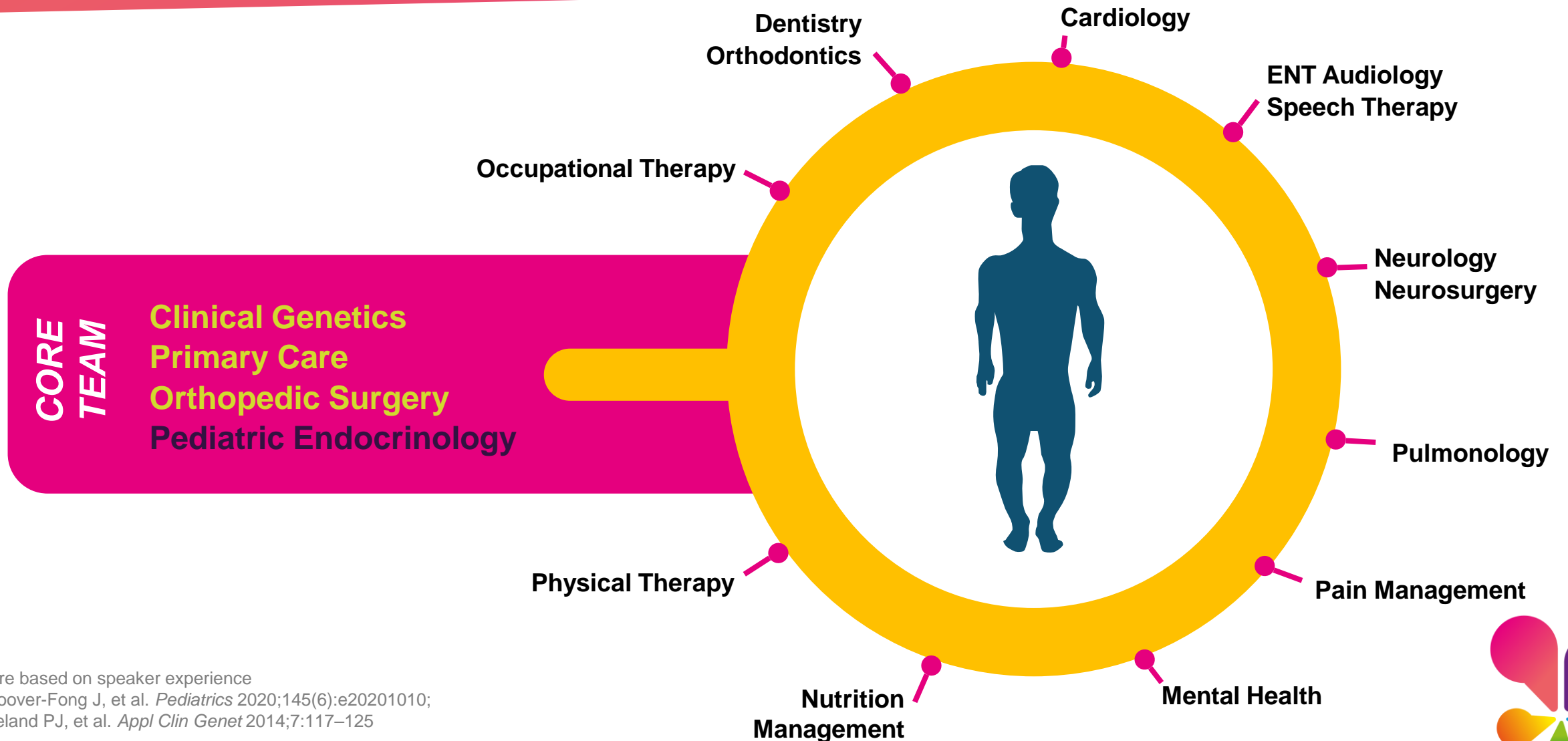


Figure based on speaker experience

1. Hoover-Fong J, et al. *Pediatrics* 2020;145(6):e20201010;

2. Ireland PJ, et al. *Appl Clin Genet* 2014;7:117-125

