Survey response: Acibadem Mehmet Ali Aydinlar University, Rare Disease and Orphan Drug Research and Application Center AND School of Medicine. Istanbul.	
1. Brief description of the "Clinical Center" or "Services on undiagnosed rare diseases" (max 1000 characters). Please add a title to the short article/description you are going to send. Description should include information on: location contact person researchers staff existing program on undiagnosed rare diseases number of undiagnosed cases collected/year number of diagnosis obtained/year scientific advances/discoveries clinical research initiatives educational and collaborative opportunities	**Location.** Istanbul. Acibadem Mehmet Ali Aydinlar University, Rare Disease and Orphan Drug Research and Application Center AND School of Medicine (ACUSoM) (www.acibadem.edu.tr/en/school-of-medicine-about-faculty), **Contact Person:** ACU-RARE director Prof.Dr. Ugur Ozbek, MD, PhD AND Prof. Yasemin Alanay, MD, PhD is the Dean of ACUSoM. **Existing program on undiagnosed rare disease:** ACURARE team have expertise on delineation of the genetics and molecular biological mechanisms of unresolved the disorders of childhood and adolescence in clinical and laboratory genetics as well as Biostatistics and Medical Informatics. Specialized in analysis of omics data in a pathway related context. Prof. Alanay is one of the most experienced groups specialized in research and clinical management of Rare Diseases in Turkey. **Number of Undiagnosed case collected a year:** 100 (500 new patients a year). **Number of Diagnosis a year:** 400
2. Past (six months) and future/upcoming events related to undiagnosed rare disease. Please indicate date and venue.	Istanbul Medical Genetics case consultation meetings are organized twice a year. Our team regularly attend the meeting and present unresolved cases. Last meeting was organized in March 2018.
3. Relevant scientific publications (selected and / or published by the center)	Van Damme T, Gardeitchik T, Mohamed M, Guerrero-Castillo S, Freisinger P, Guillemyn B, Kariminejad A, Dalloyaux D, van Kraaij S, Lefeber DJ, Syx D, Steyaert W, De Rycke R, Hoischen A, Kamsteeg EJ, Wong SY, van Scherpenzeel M,Jamali P, Brandt U, Nijtmans L, Korenke GC, Chung BHY, Mak CCY, Hausser I, Kornak U, Fischer-Zirnsak B, Strom TM, Meitinger T, Alanay Y, Utine GE, Leung PKC, Ghaderi-Sohi S, Coucke P, Symoens S, De Paepe A, Thiel C, Haack TB, Malfait F, Morava E, Callewaert B, Wevers RA. Mutations in ATP6V1E1 or ATP6V1A Cause Autosomal-Recessive Cutis Laxa. Am J Hum Genet. 100(2):216-227. (2017) Duran I, Martin JH, Weis MA, Krejci P, Konik P, Li B, Alanay Y, Lietman C, Lee B, Eyre D, Cohn DH, Krakow D. A Chaperone Complex Formed by HSP47, FKBP65, and BiP Modulates Telopeptide Lysyl Hydroxylation of Type I Procollagen. J Bone Miner Res. 32(6):1309-1319. (2017) Ranza E, Huber C, Levin N, Baujat G, Bole-Feysot C, Nitschke P, Masson C, Alanay Y, Al-Gazali L, Bitoun P, Boute O, Campeau P, Coubes C, McEntagart M, Elcioglu N, Faivre L, Gezdirici A, Johnson D, Mihci E, Nur BG, Perrin L, Quelin C, Terhal P, Tuysuz B, Cormier-Daire V. Chondrodysplasia with multiple dislocations: comprehensive study of a series of 30 cases. Clin Genet. 91(6):868-880. (2017)
4. Grants. a) grant award to announce funding for projects; b) Received grants from other organizations/institutions for undiagnosed rare diseases research.	DG-SANCO RD-ACTION Project. FP7-ADOPT Project (Rare Disease Biobank Work Package) Multinational, multicenter clinical trial on Achondroplasia treatment (BMN-301).
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