symptom to two health states and minor revisions to the language translation. ***Conclusions:*** While this project supported the face and content validity of the vignettes and attributes previously developed for the UK, it also showed that it is important to validate with local experts the language when planning to derive utilities for a cost-effectiveness model of an AADC deficiency treatment in a specific country.

PRO135
A DISCRETE CHOICE EXPERIMENT TO DERIVE HEALTH STATE UTILITIES FOR AROMATIC L-AMINO ACID DECARBOXYLASE (AADC) DEFICIENCY IN THE UNITED KINGDOM

Smith A,[1] Hanbury A,[1] Whitty J,[2] Buesch K[3]
[1]*York Health Economics Consortium, York, UK,* [2]*Norwich Medical School, University of East Anglia, Norwich, NFK, Great Britain,* [3]*PTC Therapeutics, Steinhausen, Switzerland*

Objectives: Deriving health utilities for ultra-rare medical conditions such as aromatic L-amino acid decarboxylase (AADC) deficiency poses challenges. The rarity of AADC deficiency and the fact that this genetic condition is predominantly manifested in infants means that robust utility values cannot be derived from the child or their parent/caregiver. Alternative approaches, e.g. discrete choice experiments (DCE), are required in order to provide health utilities for cost-effectiveness evaluations interventions. The study aim was to generate health utilities for AADC deficiency using a DCE. ***Methods:*** An orthogonal design was created (NGene). The DCE comprised 6 key AADCd attributes (2-6 levels): mobility, muscle weakness, oculogyric crises, feeding ability, cognitive impairment and screaming. These had been identified from published literature, clinician input, parent interviews and expert opinion. Participants were presented with 10 choice sets, including 1 with reversed levels to evaluate choice consistency. Participants were presented with 5 health state vignettes prior to the DCE. These were used to elicit utilities using time-tradeoff. The utilities for the worst/best health states were used as anchors to convert indirect DCE part-worth utilities to health utilities. Multinomial logit models were estimated (NLogit6). The DCE was completed online by panel participants from a UK representative sample. ***Results:*** A total of 1596 participants completed the DCE. The majority (70.7%) gave consistent responses to the repeated choice task; only 1.7% (27) always chose the same alternative for every choice set. Five models were evaluated. There was one preference reversal ("sitting unaided"/"standing with assistance") occurring in all models; these 2 mobility level coefficients were set to be equal in the final model. Rescaled utilities ranged from 0.4217 to 0.6703, corresponding to the worst (633233) and best (111111) health states. ***Conclusions:*** Health utilities were derived for AADC deficiency through a DCE. These will be used for a cost-effectiveness model of an AADC deficiency treatment.

PRO136
SYMPTOMS AND IMPACTS OF NONSENSE MUTATION DUCHENNE MUSCULAR DYSTROPHY AT DIFFERENT STAGES OF AMBULATION

Williams K,[1] Buesch K,[2] Piglowska N,[1] Acaster S[3]
[1]*Acaster Lloyd Consulting Ltd., London, LON, UK,* [2]*PTC Therapeutics Switzerland GmbH, Steinhausen, Switzerland,* [3]*Acaster Lloyd Consulting Ltd, London, LON, UK*

Objectives: Duchenne muscular dystrophy (DMD) is a rare genetic neuromuscular disorder which causes progressive muscle degeneration and weakness, and leads to loss of motor function and premature death. The aim of this study was to understand the symptoms and impacts of nonsense mutation DMD (nmDMD) at different stages of ambulation. ***Methods:*** Semi-structured qualitative telephone interviews were conducted with caregivers of individuals with nmDMD treated with ataluren in the UK. The interviews explored the symptoms and impacts of nmDMD, as well as impacts on caregivers. Participants also completed a background questionnaire with questions which enabled categorisation into three different health states (HS) according to level of ambulation. Qualitative data were analysed using thematic analysis and symptoms/impacts were compared across HS. ***Results:*** Ten interviews were conducted with parents of individuals aged 4-19 years. Six participants had children who were in an early ambulatory HS (can rise from supine, stand and walk 10 metres), three in a late ambulatory HS (can stand and walk 10 metres) and one in an intermediate HS (can stand). In line with the HS definitions, increasing HS severity was related to a decreasing physical function and independent action, such as the ability to walk, run/jump, climb stairs and get up off the floor, and increasing fatigue. This impacted the individuals' ability to take part in daily and social activities, and their emotional wellbeing. This declining physical function was reflected in an increased level of care and emotional burden reported by caregivers. ***Conclusions:*** As individuals with nmDMD lose ambulation, their decline in physical function can lead to impairments in other areas of life, which can impact their health-related quality of life (HRQoL) and that of their caregivers. Treatments which delay progression have the potential to prevent loss of function and decline in HRQoL of individuals and their caregivers.

Rare & Orphan Diseases - Real World Data & Information Systems

PRO137
PREVALENCE AND BURDEN OF ALPHA-1 ANTITRYPSIN DEFICIENCY IN FRANCE: AN ANALYSIS FROM THE FRENCH NATIONAL HEALTH DATABASE (SNDS)

Borget I,[1] Willemin MC,[2] Pison C,[3] Szeftel D,[4] Bourkhis E,[5] Moumane N,[5] Matheron A,[5] Lepage V[5]
[1]*Gustave Roussy Institute, Villejuif, France,* [2]*CHU Angers, Angers, France,* [3]*CHU Grenoble Alpes, Grenoble, France,* [4]*Semeia, Paris, France,* [5]*CSL Behring, Paris, France*

Objectives: Alpha-1 antitrypsin deficiency (AATD) is a rare disease leading to emphysema in severe form. AATD is associated with loss of lung tissue, progressive disability and increased mortality. We aimed to estimate the number of patients treated with replacement therapies for AATD and its economic burden. ***Methods:*** An analysis was performed using the French National Health Database (SNDS) which covered health care expenses and hospital-discharge summaries for 99 % of the French population. Treated patients were identified based on delivery of replacement therapy from 2014 to 2017. Costs were estimated from the Health Insurance perspective. Direct costs were valued using national tariffs, whereas indirect cost included paid sick leaves. ***Results:*** 365 AATD patients treated by replacement therapies were identified with mean age of 56.6 years, with a majority of male (61%). The analysis showed more frequent serious comorbidities than in the general population including high transplantation rate. Mean annual cost per patient was €13,680 (excluding A1AT therapy) driven by ambulatory related costs (45%) and hospital related costs (35%). Paid sick leaves represent 20% of the total annual cost. ***Conclusions:*** This study gives the first data regarding the number of patients treated and the economic burden of AATD in France. Understanding the profile and the burden of patients treated informs on AATD patient management.

PRO138
INSURANCE CLAIM-BASED ALGORITHMS TO IDENTIFY PATIENTS WITH NEUROMYELITIS OPTICA SPECTRUM DISORDER (NMOSD) AND RELAPSES – A SYSTEMATIC LITERATURE REVIEW

Schneider S,[1] van Beek J,[1] Exuzides A,[2] von Büdingen H-C,[1] Mouchet J[1]
[1]*F. Hoffmann-La Roche Ltd, Basel, Switzerland,* [2]*Genentech Inc, South San Francisco, CA, USA*

Objectives: Insurance claim databases are a potentially valuable source for evidence generation in NMOSD, a rare autoimmune condition of the central nervous system with scarce real-world data. The use of one International Classification of Diseases (ICD-10) code for NMOSD (G36) has shown limited validity in detecting NMOSD patients from insurance claims. To date, there is no specific ICD code to identify NMOSD relapses. This systematic literature review aimed to identify algorithms to detect NMOSD patients and relapses using insurance claims data. ***Methods:*** We searched the MEDLINE, Embase and SciSearch databases using a combination of indexed and free terms to identify articles published between 2010–2020. Complementary searches of relevant congress abstracts were conducted. Given the paucity of NMOSD literature and similarities in disease presentation, the scope was extended to multiple sclerosis (MS) to gain insights from a more advanced field. Two independent reviewers screened the abstracts for relevance against pre-determined criteria, followed by an in-depth full-text review and data extraction. ***Results:*** Ten publications reported NMOSD algorithms, developed using various criteria including prescription claims and exclusion of MS diagnosis, as well as by combining inpatient/outpatient claims with ICD-9/ICD-10 NMO/NMOSD codes and core symptoms. Only one publication validated algorithms against chart reviews to identify NMOSD patients. Two publications described a similar, non-validated algorithm to identify NMOSD relapses. The literature was more abundant for identifying MS patients and relapses, with 13 and 8 publications reporting validated algorithms, respectively. Most were evaluated by external validation; two used indirect validation. Extending the timeframe of encounters, inclusion of prescription data, and combining inpatient and outpatient claims all increased sensitivity, whereas increasing the number of claims improved positive predictive value at the cost of sensitivity. ***Conclusions:*** Due to the limited evidence, further research is needed to establish robust methods to detect both NMOSD patients and their relapses.

PRO139
IDENTIFICATION AND DESCRIPTION OF GLOBAL REAL-WORLD DATA (RWD) SOURCES FOR IGA NEPHROPATHY (IGAN), MEMBRANOUS NEPHROPATHY (MN) AND C3 GLOMERULOPATHY (C3G)

Rajput T,[1] Agrawal R,[1] Rovira G,[2] Loefroth E,[2] Studer R[2]
[1]*Novartis Healthcare Pvt. Ltd., Hyderabad, India,* [2]*Novartis Pharma AG, Basel, Switzerland*

Objectives: To identify RWD sources available for the rare renal diseases IgAN, MN and C3G globally in order to summarize and understand the availability of RWD in these rare diseases and support the design and potentially selection of data sources for future observational studies. ***Methods:*** A literature review was conducted to

identify RWD sources for IgAN, MN and C3G using Medline and EMBASE from inception until May 2019. Retrieved results were screened to devise a list of unique data sources and relevant meta-data (type of data source, study design, population size, epidemiology, demographics, clinical, economic and humanistic burden, follow-up duration, data access and linkage, etc.) was extracted. ***Results:*** Of the 2,926 retrieved publications, 173 unique RWD sources (IgAN only: 28; MN only: 32; C3G only: 3; IgAN & MN: 91; IgAN, MN & C3G: 19) were identified. There were 61% generic and 39% disease specific RWD sources. Half of the data sources (50%) covered Asian countries, followed by Europe (33%) and Americas (12%). Majority were administrative data sources (70%), followed by registries (20%). Data sources recorded minimum 5% and maximum 56% of the 68 assessed variables. The most commonly reported (>90%) variables were age, sex and biopsy as mode of diagnosis; quality of life, caregiver burden, and details of hospitalizations were the least reported (<1%) ones across all the three indications. Patients' baseline eGFR and proteinuria levels were captured in >60% and >80% data sources, respectively. Possibility to access was actively reported in 21% of data sources. ***Conclusions:*** This comprehensive overview identified RWD sources for IgAN and MN in certain geographies, with limited availability of existing RWD for C3G. The overview might provide the base for identification of opportunities for future partnerships and more efficient and sustainable use of RWD.

PRO140
HEALTH-RELATED QUALITY OF LIFE ASSOCIATED WITH MYCOSIS FUNGOIDES-TYPE CUTANEOUS T-CELL LYMPHOMA PATIENTS: DETERMINATION OF UTILITY VALUES FROM A UK-BASED CLINICIAN QUESTIONNAIRE

Scarisbrick J.[1] Schmidt F,[2] Turini MM,[3] D'agostino P,[3] Thornton S,[4] Evans J,[5] Ader J,[6] van Hest N,[6] Brazier JE[7]
[1]*University Hospital Birmingham, Birmingham, UK,* [2]*Recordati Rare Diseases, Puteaux, France,* [3]*Helsinn Healthcare SA, Pazzallo - Lugano, Switzerland,* [4]*Cutaneous Lymphoma Foundation, Birmingham, MI, USA,* [5]*Costello Medical, Cambridge, CAM, UK,* [6]*Costello Medical, London, UK,* [7]*University of Sheffield, Sheffield, UK*

Objectives: Utility studies quantify the impact of disease on patients' health-related quality of life (HRQoL). In rare diseases particularly, utility data are often limited. Where direct elicitation of data from patients is unfeasible, indirect methods are required to determine health state utility values (HSUVs). Here, we present indirect elicitation of HSUVs for patients with mycosis fungoides (MF), a rare skin lymphoma characterised by painful, itchy skin patches and plaques, which can progress into tumours and may involve blood, lymph nodes and viscera. ***Methods:*** Twelve distinct vignettes were prepared, based on published literature and expert opinion. The vignettes were validated by a MF patient expert and described typical MF patients with varying disease stage (IA-IVB) and skin burden, as assessed by the modified Severity Weighted Assessment Tool (mSWAT). Other included patient characteristics (age, Eastern Cooperative Oncology Group [ECOG] score) were informed by a patient registry. UK clinicians, working at key NHS lymphoma treatment centres (N=7), acted as respondents and completed the EQ-5D-5L questionnaire (proxy version 2) electronically for each vignette, responding as they believed MF patients would. Responses were converted into HSUVs using the UK value set. ***Results:*** In every disease stage, HSUVs decreased with increasing mSWAT score. Similarly, for each mSWAT range HSUVs decreased as disease stage advanced. This demonstrates the effect of increasing skin burden and disease progression on HRQoL. Despite this, HSUVs associated with mSWAT score=0 were similar, regardless of disease stage (0.945 in Stage IA, 0.924 in Stage IB-IIA and 0.917 in Stage IIB-IV). ***Conclusions:*** The results of this investigation suggest that proxy-reporting via EQ-5D represents a potential option for generating HRQoL data, if published estimates are not available. Despite the small sample size and indirect elicitation of data, the finding that HRQoL decreases with increased mSWAT score is aligned with published literature.

PRO141
TREATMENT PATTERNS PRIOR TO THE USE OF THROMBOPOIETIN RECEPTOR AGONISTS IN PATIENTS WITH IMMUNE THROMBOCYTOPENIA IN GERMANY

Schill M.[1] Taieb V,[2] Richter H,[3] Lebioda A[1]
[1]*Amgen GmbH, Munich, Germany,* [2]*Amgen Ltd, Uxbridge, UK,* [3]*IQVIA Epidemiology, Frankfurt, Germany*

Objectives: To describe real-world treatment patterns and steroid usage as well as the steroid prescription alignment to clinical guidelines, prior to the use of thrombopoietin receptor agonists (TPO-RAs) in patients with immune thrombocytopenia (ITP) in Germany. ***Methods:*** The IMS® LRx prescription database was used to identify patients with at least one TPO-RA (romiplostim or eltrombopag) prescription between 7/2016 and 6/2019 in Germany. ITP treatments received prior to TPO-RA therapy were assessed. ***Results:*** In total 3,553 patients were included in the analysis (2,752 eltrombopag, 801 romiplostim). Overall, 48% of patients were male and 57% of patients were >60 years when initiating TPO-RA. Oral steroids were the most commonly used therapy before TPO-RAs, in 2,289 of patients (64%; 69% eltrombopag, 48% romiplostim), followed by intravenous immunoglobulins (IVIg) in 249 of patients (7%; 6% eltrombopag, 10% romiplostim). Rituximab or other ITP treatments (e.g. azathioprine, mycophenolate mofetil, cyclophosphamide, dapsone) were rarely used before TPO-RAs. Rituximab was used in 2% of patients (1% eltrombopag, 4% romiplostim), other prior treatments in 5% of patients (5% eltrombopag, 6% romiplostim). Mean total days of steroid use prior to TPO-RAs was 169 days (median 105 days, maximum 540 days) and mean cumulative steroid (prednisolone equivalent) dose before TPO-RA therapy was 3,328 mg (median 2,522 mg, maximum 39,702 mg). ***Conclusions:*** Oral steroids were the most commonly used medication before TPO-RA therapy, reflecting their continued importance in the management of newly diagnosed ITP. However, treatment duration and cumulative steroid dose exceeded current guideline recommendations (ie from the American Society of Hematology 2019) and international consensus on ITP treatment. This over-use of steroids has the potential to lead to serious side effects and negatively impact on mental health, mood, sleep or weight gain for patients.

PRO142
IDENTIFICATION AND DESCRIPTION OF REAL-WORLD DATA SOURCES FOR PAROXYSMAL NOCTURNAL HEMOGLOBINURIA (PNH): A REVIEW OF LITERATURE

Lavudiya S.[1] Agrawal R,[1] Rovira G,[2] Studer R[2]
[1]*Novartis Healthcare Pvt. Ltd., Hyderabad, India,* [2]*Novartis Pharma AG, Basel, Switzerland*

Objectives: To identify global real-world data (RWD) sources available for the rare blood disease **Paroxysmal Nocturnal Hemoglobinuria** (PNH) in order to summarize and understand the availability of RWD in this rare disease. ***Methods:*** A literature review was conducted to identify RWD sources for PNH using Medline and EMBASE from inception until June 2019. Identified literature was reviewed and a list of unique data sources was derived. Data sources were examined and meta-data for 90 variables was extracted to describe details around type of data source, study design, population size, epidemiology, clinical, economic and humanistic burden, follow-up duration, data access and linkage. ***Results:*** A total of 657 identified publications were screened. Among these, 45 potential RWD sources were identified - 80% were generic and 20% disease specific. Almost one-third of the data sources covered Asia, followed by Europe (24%), Americas (20%) and Oceania (2%); 7% were multi-regional. The majority of the RWD sources were administrative. Approximately half of the data sources (51%) reported a sample size in the range of 101 to 500. Data sources recorded minimum 2% and maximum 61% of the 90 assessed variables. More than 60% of RWD sources reported age, gender, diagnosis, PNH clone size and incidence of thrombotic events; the least (<1%) reported variables were risk factors, caregiver involvement, cost and resource use. Lactate dehydrogenase (LDH) levels cut-off and hemoglobin levels were reported in 60% and 53% of data sources respectively. ***Conclusions:*** It seems that there is only a limited collection of RWD sources available based on our current review. Thus, to close the gap in existing evidence in this disease area the importance of collaboration to leverage and generate relevant RWD in this rare disease needs to be highlighted.

PRO143
COSTS OF MULTIPLE MYELOMA PATIENTS DIAGNOSED WITH PERIPHERAL NEUROPATHY IN GERMANY

Lebioda A.[1] Popfinger K,[2] Weiß V,[3] Scheider M[3]
[1]*Amgen GmbH, Munich, Germany,* [2]*University of Bayreuth, Bayreuth, Germany,* [3]*Team Gesundheit GmbH, Essen, Germany*

Objectives: Based on data from the German statutory health insurance system (GKV), we evaluated the costs and health resource utilization (HRU) associated with peripheral neuropathy (PNP), a commonly reported adverse event in patients (pts) with multiple myeloma (MM). ***Methods:*** A claims database was used to identify pts with MM (ICD-10 C90.0-), which includes around 3.7 million insured pts representing approximately 5% of the GKV-population. Among MM pts who experienced PNP between 01/2014-12/2017 (coded as ICD-10 G62.0, G62.8, or G62.9), PNP-associated costs were evaluated using the uniform physician's fee scale and Diagnosis Related Groups. Only the costs reimbursed by GKV were considered; additional out-of-pocket expenditures by patients were not captured. ***Results:*** In total, 618 MM pts were included, 58% were male and 72% aged ≥65 years. Most pts (88%) were treated in outpatient setting. For 12% (n=76) of MM pts, PNP was documented during their first-line treatment. A bortezomib-based regimen was the most common treatment regimen (83% of all patients) in first line with a median treatment duration of 92 days. Of the 76 PNP cases, 29 were treated in a hospital and 52 in ambulatory care. Total costs of care for patients with a PNP diagnosis varied according to age: mean hospital costs were €39,454 in the elderly pts (aged ≥65) and €7,956 in younger pts. Mean ambulatory costs of PNP treatment were higher for the elderly pts (€77) than younger pts (€31). Further costs for other cures, remedies, or individual prevention courses were documented for 37 pts. These costs did not differ by age group (≥65: €323; <65: €372). ***Conclusions:*** In this real-world MM cohort, PNP was reported frequently in pts treated with bortezomib in first line. PNP management showed to be an increased cost burden especially in elderly pts for the German healthcare system.