Guidance document for processing PM-JAY packages

Inborn Errors of Metabolism

Procedures covered: 1 **Specialty:** Pediatric Medical Management

Package Name	Procedure Name	HBP 1.0 code	HBP 2.0 code	Package price (INR)
Inborn errors of metabolism	Inborn errors of metabolism	M200060	MP036A	General Ward- 1800/- HDU – 2700/- ICU without ventilator– 3600/- ICU with Ventilator– 4500/-

ALOS: 7 days

Minimum qualification of the treating doctor:
Essential: MD/DNB/DCH (Pediatric) and might require a multidisciplinary approach

Special empanelment criteria/linkage to empanelment module: Care at a Tertiary Hospital

Disclaimer:
For monitoring and administering the claim management process for **Inborn Errors of Metabolism,** shall be following these guidelines. This document has been prepared for guidance of PROCESSING TEAM and TRANSACTION MANAGEMENT SYSTEM of AB PM-JAY for the claims of procedures mentioned above. The hospitals can also refer to this document so that they have the insight on how the claims will be processed. However, this document doesn't provide any guidance on clinical and therapeutic management of patient. In that respect the hospitals and physicians may refer to any other relevant material as per the extant professional norms.

PART I: GUIDELINES FOR CLINICAL AND HEALTHCARE PROVIDERS

1.1 Objective:

The purpose of this section is to act as a guidance & a clinical decision support tool for the clinicians in deciding the line of treatment, plan clinical management of patient and decide referral of cases to the appropriate level of care (as required) for treatment of patients under PMJAY and selection of corresponding Health Benefit Package.

It will also serve as a tool for hospitals to determine and submit the mandatory documents required for claiming reimbursement of health benefit package under PMJAY.

1.2 Clinical key pointers:

Inborn errors of metabolism are a heterogeneous group of disorders that may be inherited or may occur as the result of spontaneous mutation. These diseases involve failure of the metabolic pathways involved in either the break-down or storage of carbohydrates, fatty acids, and proteins. They can present at any age, and therefore, a working knowledge of these diseases, their presentations, and their evaluation is critical for the emergency provider.

Inborn errors of metabolism are inherited disorders caused by mutations in genes coding for proteins that function in metabolism. Most are inherited as autosomal recessive. Rarely, they are autosomal dominant and X-linked. Environmental, epigenetic, and microbiome factors and additional genes are potential modifying etiologic factors in those with inborn errors of metabolism.

Clinical presentation:

- Patients with severe errors in carbohydrate metabolism present early (typically in the neonatal period) and catastrophically. Clinically, they can be indistinguishable from septic neonates, with hypoglycemia, brady/and tachydysrhythmias, hypothermia or hyperpyrexia, seizures, and poor tone.
- Patients with severe errors in excretion pathways will present typically with intoxication, with lethargy and altered mental status, seizures, vomiting, and vital sign abnormalities.
- Patients with errors in pathways involved in accessing stored energy may appear well for prolonged periods of time and can be asymptomatic as long as they have an ongoing intake of carbohydrates.

Commonly presented as

- Although all these presentations exist on a spectrum depending upon degree and type of enzyme involvement, inborn errors of metabolism have some common features.
- The most common presentation of these diseases taken as a whole is neurologic abnormalities, which occurs in about 80% of individuals. These abnormalities include developmental delay, loss of milestones, poor tone, poor suck, and seizure.
- The second most common presentation is related to gastrointestinal symptoms, including vomiting, hepatomegaly, food intolerance, diarrhea, food aversion, exercise intolerance, and dehydration.
- More than half of children have both neurologic and gastrointestinal abnormalities.

Investigation:

- Routine screening test at birth
- Electrocardiogram (ECG)
- USG (cranial etc.)
- Complete septic screening
- Cerebrospinal fluid (CSF) examination
- LFT/KFT/ blood gas/ammonia/lactate
- Urine for ketones and reducing substance.

Management

The management of patients with inborn errors of metabolism is extremely complex and challenging. In many cases, there is no specific treatment and symptoms are managed as they arise. The disorders are best managed by an interprofessional team that includes pharmacists, nurses, therapists, dietitians, and social workers.

- Supportive therapy
- Enzyme replacement therapy
- Substrate reduction

1.3 Mandatory documents- For healthcare providers

Following documents should be uploaded by the concerned hospital staff at the time of pre-authorization and claims submission:

Mandatory document	Inborn Errors of Metabolism
i. At the time of Pre-authorization	
Clinical notes including history, evaluation findings, and planned line of management	Yes
Based on Etiology Cerebrospinal fluid (CSF) examination Liver Function Test Kidney Function Test Arterial blood gas Ammonia Urine for ketones and reducing substance USG CT/MRI X Ray Tandem Mass Spectrometer (TMS) Gas chromatography-mass spectrometry (GCMS) Gene testing Exome sequencing	Yes
ii. At the time of claim submission	
Detailed ICPs mentioning the treatment details	Yes
Detailed Discharge Summary	Yes

PART II: GUIDELINES FOR PROCESSING TEAM

2.1 **Objective:** To provide guidance to the pre-authorization and claims processing team in ascertaining the medical necessity of procedure carried out vis a vis the patient's medical condition as evidenced by supporting documents/investigation reports etc., in deciding the admissibility and quantum of claim and compliance with mandatory documents by the hospital.

2.2 **Following mandatory documents to be diligently reviewed by the pre-auth / claims processing personnel:**

2.2.1 **At the time of pre-authorization processing- For pre-authorization processing doctor (PPD)**:

a. Clinical notes - detailed history especially prenatal, perinatal, family history, signs & symptoms, vital monitoring, planned treatment line, and advice for admission?
b. Did clinical presentation and evaluation findings suggestive of Inborn Errors of Metabolism?

2.2.2 **At the time of claim processing- For claims processing doctor (CPD):**

a. Are the detailed ICPs with daily vitals and treatment details submitted?
b. Was investigations/imaging supportive of diagnosis?
c. Is the Discharge summary with follow-up advice at the time of discharge?

PART III: GUIDELINES FOR TRANSACTION MANAGEMENT SYSTEM (TMS)

3.1 **Objective:** To enable setting up of cross check mechanisms/rule engines within the IT platform (TMS) to ensure compliance with STGs and to prevent fraud / abuse of the Health Benefit Package.

3.2 **Below mentioned are the scenarios where a provision would be built in TMS for pop-ups:**

I. Was clinical presentation, evaluation findings, and imaging/investigations suggestive of diagnosis? Yes

Till the time the functionality is being developed, the processing doctors shall check the above manually.

Reference

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3. Agana M, Frueh J, Kamboj M, Patel DR, Kanungo S. Common metabolic disorder (inborn errors of metabolism) concerns in primary care practice. Ann Transl Med. 2018;6(24):469. doi:10.21037/atm.2018.12.34
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5. El-Hattab AW. Inborn errors of metabolism. Clin Perinatol. 2015 Jun;42(2):413-39, x. doi: 10.1016/j.clp.2015.02.010. Epub 2015 Apr 8. PMID: 26042912.